CytoScan HD
2015 Publications

1-
2q31.1 microdeletion syndrome: case report and literature review
Surasak Puvabanditsin*, Melissa February, Tazeem Shaik, Arun Kashyap, Chantal Bruno andRajeev Mehta
Department of Pediatrics, Rutgers Robert Wood Johnson Medical School, New Brunswick, New Jersey, USA
Clinical Case Reports, June 2015, 3(6): 357–360.
We describe a preterm neonate with bilateral coloboma of the iris, upper and lower limb malformations including rocker bottom feet, camptodactyly, and clinodactyly together with microcephaly and small for gestational age whom cytogenetic diagnosis using SNP microarray detected an interstitial deletion of chromosome 2 between 2q31.1 and 33.1. ... SNP microarray analysis was performed using the Affymetrix CytoScan HD platform. There was a 23.16 megabase (MB) deletion between 2q31.1 and 2q33.1.

2-
5p13.3p13.2 duplication associated with developmental delay, congenital malformations and chromosome instability manifested as low-level aneuploidy
Ivan Y. Iourov123*, Svetlana G. Vorsanova12, Irina A. Demidova12, Galina A. Aliamovskaia2, Elena S. Keshishian2 and Yuri B. Yurov12
1 Mental Health Research Center, Russian Academy of Medical Sciences, Moscow 117152, Russia;
2 Russian National Research Medical University named after N.I. Pirogov, Separated Structural Unit “Clinical Research Institute of Pediatrics”, Ministry of Health of Russian Federation, Moscow 125412, Russia ; 3 Dept of Medical Genetics, Russian Medical Academy of Postgraduate Education, Moscow 123995, Russia
SpringerPlus, 15 October 2015, 4:616.
... High-resolution chromosomal SNP microarray analysis was performed on DNA isolated from peripheral blood lymphocytes using CytoScan ® HD Arrays (Affymetrix ...

3-
7q21.3 Deletion involving enhancer sequences within the gene DYNC1I1 presents with intellectual disability and split hand-split foot malformation with decreased penetrance
Sara Delgado1 and Milen Velinov123*
1 Bronx-Lebanon Hospital Center, New York, Bronx ; 2 Albert Einstein College of Medicine, New York, Bronx ; 3 Dept of Human Genetics, New York State Institute for Basic Research in Developmental Disabilities, 1050 Forest Hill Rd, Staten Island 10314, New York
Molecular Cytogenetics, 13 June 2015, 8:37
... Oligo SNP array, Affimetrix Cytoscan HD (Quest Diagnostic, San Juan CA) identified an approximately 1 Mb deletion in chromosomal region 7q21.3.

4-
7q21.11 Microdeletion in a Neonate With Goldenhar Syndrome: Case Report and a Literature Review
Surasak Puvabanditsin, M.D., Melissa February, M.D., Lissa Francois, M.D., Eugene Garrow, M.D., Chantal Bruno, D.O., Rajeev Mehta, M.D.
The Cleft Palate-Craniofacial Journal, online 11 June 2015
... Chromosome microarray analysis was performed using both copy number and single-nucleotide polymorphism probes on a whole-genome array (Affymetrix CytoScan HD platform).

5-
8p23.1 Interstitial Deletion in a Patient with Congenital Cardiopathy, Neurobehavioral Disorders, and Minor Signs Suggesting 22q11.2 Deletion Syndrome
Molck, Miriam C.; Monteiro, Fabíola P.; Simioni, Milena; Gil-da-Silva-Lopes, Vera L.
Journal of Developmental & Behavioral Pediatrics, September 2015, 36(7): 544–548.
... highlights the application of aGH to investigate 8p23.1 deletion in nonconfirmed 22q11.2 DS patients ... Array genomic hybridization analysis for the patient was performed using CytoScan HD chip (Affymetrix ®) ...

6-

A 12q24.31 interstitial deletion in an adult male with MODY3: Neuropsychiatric and neuropsychological characteristics

Willem M.A. Verhoeven1,2,*, Jos I.M. Egger1,3, Joop P.W. van den Bergh4,5, Ronald van Beek6, Tjitske Kleefstra6 and Nicole de Leeuw6
1Vincent van Gogh Institute for Psychiatry, Centre of Excellence for Neuropsychiatry, Venray, The Netherlands; 2Erasmus University Medical Centre, Department of Psychiatry, Rotterdam, The Netherlands; 3Donders Institute for Brain, Cognition and Behaviour, Behavioural Science Institute, Radboud University Nijmegen, Nijmegen, The Netherlands; 4VieCuri Medical Centre, Department of Internal Medicine, Venlo, The Netherlands; 5Maastricht University Medical Centre, Dept of Internal Medicine, Maastricht, The Netherlands; 6Dept of Human Genetics, Radboud University Medical Centre, Nijmegen, The Netherlands

American Journal of Medical Genetics Part A, January 2015, 167(1):169–173.

... This is the first patient with normal intelligence in whom the presence of subtle facial dysmorphisms were decisive for introducing genetic analysis that, in turn, disclosed a rare form of diabetes necessitating modifications in treatment regimen. ... Genomic array analysis with an average resolution of ~20 kb was performed using the Affymetrix CytoScan HD array platform ...

7-

A Case Report: Jacobsen Syndrome Complicated by Paris-Trousseau Syndrome and Shone's Complex

Malia, Laurie A; Wolkoff, Leslie I.; Mnayer, Laila; Tucker, Joseph W.; Parikh, Nehal S.

Journal of Pediatric Hematology/Oncology, October 2015, 37(7): e429–e432.

... FIGURE 2. Microarray: SNP microarray was performed using the Affymetrix CytoScan HD. The array detected a 12.1 MB deletion (122,797,729-134,938,470) with 108 genes deleted. Microarray showing detailed view of deletion 11q.

8-

A cryptic balanced translocation (5;17), a puzzle revealed through a critical evaluation of the pedigree and a FISH focused on candidate loci suggested by the phenotype

A. Primerano1, E. Colao1, C. Villella1, M. D. Nocera1, A. Ciambrone1, E. Luciano1, L. D'Antona4, M. F. M. Vismara1, S. Loddo2, A. Novelli3, N. Perrotti14† and Paola Malatesta1*†
1 Unit of Medical Gentics, University Hospital Mater Domini, Catanzaro, Italy ; 2 Mendel Laboratory, Casa Sollievo della Sofferenza Hospital, IRCCS, San Giovanni Rotondo, Italy ; 3 Genetics Unit, Bambino Gesu Children's Hospital, IRCCS, Rome, Italy ; 4 Dept of "Scienze della Salute", Università UMG di Catanzaro, Catanzaro, Italy

Molecular Cytogenetics, 2 September 2015, 8:70

...The imbalances were confirmed by a new high resolution SNP array. ... Microarray analysis was performed using CytoScan HD (Affymetrix, ...

9-

A germline chromothripsis event stably segregating in 11 individuals through three generations

Birgitte Bertelsen PhD, Lusine Nazaryan-Petersen PhD, Wei Sun PhD, Mana M. Mehrjouy MSc, Gangcai Xie PhD, Wei Chen PhD, Lena E. Hjermind PhD, Peter E. M. Taschner PhD & Zeynep Tümer PhD, DMSc

Genetics in Medicine, online 27 August 2015

... Chromosomal microarray was performed for II:6 using Affymetrix CytoScan HD array (Affymetrix, Santa Clara, CA), and data were analyzed with ChAS software (Affymetrix) ...

10-

A late onset sickle cell disease reveals a mosaic segmental uniparental isodisomy of chromosome 11p15

Isabelle Vinatier a, Xavier Martin b, Jean-Marc Costa a, Anne Bazin a, Stéphane Giraudier c, Philippe Joly d
a Laboratoire CERBA, 95066 Cergy-Pontoise cedex 9, France; b Service de médecine polyvalente, Centre hospitalier Antoine Gayraud, 11890 Carcassonne cedex 9, France; c Service d'hématologie biologique, GH Henri-Mondor, 51 avenue du Maréchal de Lattre de Tassigny, 94010 Créteil Cedex, France; d Unité de Pathologie Moléculaire du Globule Rouge, Laboratoire de Biochimie et Biologie moléculaire, Hôpital Edouard Herriot, Hospices Civils de Lyon & Université Claude Bernard-Lyon 1, Lyon, France

Blood Cells, Molecules, and Diseases, January 2015, 54(1):53–55.
doi:10.1016/j.bcmd.2014.07.021

… A SNP array analysis (CytoScan HD SNP Affymetrix) confirmed this hypothesis as it revealed a 45.9 Mb LOH on almost the whole short arm of chromosome 11 …

11-

A maternally inherited 16p13.11-p12.3 duplication concomitant with a de novo SOX5 deletion in a male patient with global developmental delay, disruptive and obsessive behaviors and minor dysmorphic features

Ines Quintela1, Francisco Barros2,*, Ramon Lago-Leston3, Manuel Castro-Gago4, Angel Carracedo1,2,5 and Jesus Eiris4

1Grupo de Medicina Xenomica - Universidad de Santiago de Compostela, Centro Nacional de Genotipado - Instituto Carlos III, Santiago de Compostela, Spain; 2Grupo de Medicina Xenomica - USC, CIBERER, Fundacion Publica Galega de Medicina Xenomica - SERGAS, Santiago de Compostela, Spain; 3Grupo de Medicina Xenomica - USC, Fundacion Publica Galega de Medicina Xenomica - SERGAS, Santiago de Compostela, Spain; 4Departamento de Pediatria, Hospital Clinico Universitario de Santiago de Compostela - Unidad de Neurologia Pediatrica, Santiago de Compostela, Spain; 5Center of Excellence in Genomic Medicine Research, King Abdulaziz University, Jeddah, Saudi Arabia

American Journal of Medical Genetics Part A, June 2015, 167(6): 1315–1322.

... DNA samples from both the patient and his parents were obtained from peripheral blood and genotyped using the Cytogenetics Whole-Genome 2.7 M SNP array and the CytoScan High-Density SNP array (Affymetrix, ...

12-

A missense mutation in domain III in HSPG2 in Schwartz–Jampel syndrome compromises secretion of perlecan into the extracellular space

Satoshi Iwata, Mikako Ito, Tomohiko Nakata, Yoichiro Noguchi, Tatsuya Okuno, Bisei Ohkawara, Akio Masuda, Tomohide Goto, Masanori Adachi, Hitoshi Osaka, Risa Nonaka, Eri Arikawa-Hirasawa, Kinji Ohno

Neuromuscular Disorders, August 2015, 25(8): 667–671.

... We also searched for copy number variations (CNVs) by the CytoScan HD array (Affymetrix).

13-

A modified multiplex ligation-dependent probe amplification method for the detection of 22q11. 2 copy number variations in patients with congenital heart disease

Xiaoqing Zhang, Yuejuan Xu, Deyuan Liu, Juan Geng, Sun Chen, Zhengwen Jiang, Qihua Fu, Kun Sun

BMC Genomics, May 8, 2015, 16:364

... A genomic hybridization was performed on each sample with the CytoScan HD array (Affymetrix, ...

14-

A mosaic small supernumerary marker chromosome 17 in a patient with Tourette syndrome, ADHD and intellectual disability: A case story and review of the literature

Nanna Cornelius a, 1, Birgitte Bertelsen a, 1, Linea Melchior a, Lusine Nazaryan a, Zeynep Tümer a, Nanette Mol Debes b, Camilla Groth b, Liselotte Skov b

a Department of Clinical Genetics, Applied Human Molecular Genetics, Kennedy Center, Copenhagen University Hospital, Rigshospitalet, Glostrup, Denmark; b The Tourette Clinic, Department of Pediatrics, Herlev University Hospital, Herlev, Denmark

Psychiatry Research, 30 July 2015, 228(1): 179–181.

...The sSMC was present in mosaic form as 47,XY+mar/46,XY and microarray analysis using Affymetrix CytoScan HD ... a copy number gain corresponding to chromosome 17 spanning an approximately 6.3 Mb region at 17p11.2-q11.2 ...

15- *Meeting abstract*

A new biomarker of response to 5-azacitidine therapy in MDS and AML patients: SIRPB1

Viviana Guadagnuolo1, Cristina Papayannidis1, Ilaria Iacobucci1, Giorgia Simonetti1, Antonella Padella1, Stefania Paolini1, Mariachiara Abbenante1, Sarah Parisi1, Francesca Volpato1, Chiara Sartor1, Maria Chiara Fontana1, Massimo Delledonne2, Michele Malagola3, Carla Filì3, Domenico Russo3, Sandro Grilli4, Michele Cavo1, and Giovanni Martinelli1

1L. & A. Seragnoli, Bologna, Italy; 2Dept of Biotechnology, Verona University, Verona, Italy; 3Unit of Blood Diseases and Cell Therapies, University of Brescia - AO Spedali Civili Brescia, Brescia, Italy; 4Dipartimento di Medicina Specialistica, Diagnostica e Sperimentale, Bologna, Italy.

Cancer Research, 2015, 75(15 Suppl):Abst #4835. [AACR 106th Annual Mtg, Apr 18-22, 2015, Philadelphia, PA]

… To identify the genes mostly predictive of treatment response, we use high-throughput genomic analysis (SNP arrays and/or NGS-RNA-seq and/or NGS-WES) in azacitidine-sensitive and resistant MDS/AML patients. Materials and Methods: NGS-WES or RNA seq HiSeq 2000 (Illumina) was positively done in 35/214 AML samples (16%). SNPs arrays (CytoScan HD Array, Affymetrix Inc.) was done in 125/214 AML samples (58%) and 18/32 MDS samples (56%) at diagnosis, then analyzed by Chromosome Analysis Suite (ChAS) v1.2 (Affymetrix Inc.), Nexus Copy Number™ v7.5 (BioDiscovery) and GeneGo MetaCore™ software. ...

16-

A Novel 23.1 Mb Interstitial Deletion Involving 7q22.3q32.1 in a Girl with Short Stature, Motor Delay, and Craniofacial Dysmorphism.

del Refugio Rivera-Vega M.a· Gómez-del Angel L.A.a· Valdes-Miranda J.M.a· Pérez-Cabrera A.a· Gonzalez-Huerta L.M.a· Toral-López J.b· Cuevas-Covarrubias S.a
aDepartamento de Genética Médica, Hospital General de México/Facultad de Medicina, Universidad Nacional Autónoma de México (UNAM), México, DF, and bDepartamento de Genética Médica, Centro Médico Ecatepec, ISSEMYM, Toluca, México
Cytogenet Genome Research, 2015,145:1-5.
... Here, a Mexican girl with microcephaly, facial dysmorphism, short stature, hand anomalies, and intellectual disability was analyzed by CytoScan HD array. Her phenotype was associated with a de novo 7q22.3q32.1 deletion involving 109 loci ...

17-
A novel large deletion of the ICR1 region including H19 and putative enhancer elements
Helen Fryssira, Stella Amenta, Deniz Kanber, Christalena Sofocleous, Evangelia Lykopoulou, Christina Kanaka-Gantenbein, Flavia Cerrato, Hermann-Josef Lüdecke, Susanne Bens, Andrea Riccio and Karin Buiting
BMC Medical Genetics, May 6, 2015, 16:30
... Molecular karyotyping. Array analysis was carried out using the genome-wide high-resolution SNP array CytoScan HD (Affymetrix) ...

18-
A Novel Whole Exon Deletion in WWOX Gene Causes Early Epilepsy, Intellectual Disability and Optic Atrophy
Salma Ben-Salem1, Aisha M. Al-Shamsi2, Anne John1, Bassam R. Ali1, Lihadh Al-Gazali3
1. Dept of Pathology, College of Medicine and Heath Sciences, United Arab Emirates University, Al-Ain, United Arab Emirates ; 2. Dept of Paediatrics, Tawam Hospital, Al-Ain, United Arab Emirates ; 3. Dept of Paediatrics, College of Medicine and Heath Sciences, United Arab Emirates University, PO box 17666, Al-Ain, United Arab Emirates
Journal of Molecular Neuroscience, May 2015, 56(1):17-23.
... Chromosomal Microarray Genomic DNA from the index patient (Fig. 1, IV-3) was analyzed by chromosomal microarray (CMA) using whole- genome array by means of Affymetrix cytoscan HD platform ...

19-
A pure familial 6q15q21 split duplication associated with obesity and transmitted with partial reduction
Emilie Landais1,2, Camille Leroy1,3, Pascale Kleinfinger4, Stéphanie Brunet1, Valérie Koubi5, Christine Pietrement6, Marie-Laurence Poli-Mérol3,7, Caroline Fiquet7,8, Pierre-François Souchon6, Mylène Beri9, Philippe Jonveaux9, Roselyne Garnotel10, Dominique Gaillard1,3 andMartine Doco-Fenzy1,8,*
1CHU-Reims, HMB, Service de Génétique, France; 2CHU-Reims, HMB, Plateforme Régionale de Biologie Innovante, France; 3Université de Reims Champagne-Ardenne, UFR de médecine, France; 4Laboratoire de Génétique, Pasteur Cerba, Cergy Pontoise, France; 5Service de génétique Médicale, Laboratoire de génétique moléculaire, CHU Hopital Necker enfants malades, Paris, France; 6CHU-Reims, American Memorial Hospital, Service de Pédiatrie A, France; 7CHU-Reims, American Memorial Hospital, Service de Chirurgie pédiatrique, France; 8SFR CAP Santé, Reims, France; 9CHU-Nancy, Laboratoire de Génétique Médicale, Nancy Université, France; 10CHU-Reims, Laboratoire de Biochimie Médicale et Biologie Moléculaire, CNRS UMR 6198, UFR, Médecine, France
American Journal of Medical Genetics Part A, June 2015, 167(6): 1275–1284.
... SNP analysis was then performed using Affymetrix high-density SNP arrays (CytoScan® HD Array Kit, Affymetrix, Santa Clara, CA), which analyze 750,000 genotypable SNPs, and CytoScan HD Array Chromosome Analysis Suite (ChAS) Software.

20-
A rare but recurrent t(8;13)(q24;q14) translocation in B-cell chronic lymphocytic leukaemia causing MYC up-regulation and concomitant loss of PVT1, miR-15/16 and DLEU7 **[free access]**
Gemma Macchia1, Angelo Lonoce1, Santina Venuto1, Ettore Macrí2, Orazio Palumbo3, Massimo Carella3, Crocifissa Lo Cunsolo2, Paolo Iuzzolino2, María Hernández-Sánchez4, Jesus M. Hernandez-Rivas4 and Clelia T. Storlazzi1
1Dept of Biology, University of Bari, Bari, Italy; 2UO Anatomia Patologica, Ospedale S. Martino, Belluno, Italy; 3Medical Genetics Unit, IRCCS Casa Sollievo della Sofferenza Hospital, San Giovanni Rotondo, Italy; 4Servicio de Hematología, Hospital Universitario de Salamanca, IBSAL, IBMCC, Centro de Investigación del Cáncer, Universidad de Salamanca, CSIC, Salamanca, Spain
British Journal of Haematology, Article first published online: 26 May 2015
... Subsequently, single nucleotide polymorphism (SNP) array analysis of the patient's PB DNA, accomplished on a CytoScan® HD Array according to manufacturer protocols using the ChAS

Software (Affymetrix, Santa Clara, CA, USA), confirmed a deletion that included MIR15A/MIR16-1 and DLEU7 on the der(13) (chr13: 49.85–51.07 Mb).

21-
A rare coincidence of different types of driver mutations among uterine leiomyomas (UL)
Carsten Holzmann1, Dominique Nadine Markowski2, Sabine Bartnitzke2, Dirk Koczan3, Burkhard Maria Helmke45 and Jörn Bullerdiek12*
1 Institute of Medical Genetics, University Rostock Medical Center, Ernst-Heydemann-Strasse 8, Rostock, D-18057, Germany; 2 Center of Human Genetics, University of Bremen, Leobener Strasse ZHG, Bremen, D-28359, Germany; 3 Institute of Immunology, University of Rostock, University Rostock Medical Center, Schillingallee 70, Rostock, D-18057, Germany; 4 Institute of Pathology, University of Heidelberg, Heidelberg, Germany
Molecular Cytogenetics, 14 October 2015, 8:76
... CNV (copy number variation) analysis was performed using premade CytoScan HD Arrays (Affymetrix ...

22-
A recurrent deletion syndrome at chromosome bands 2p11.2-2p12 flanked by segmental duplications at the breakpoints and including REEP1
Servi J C Stevens, Eveline W Blom, Ingrid T J Siegelaer and Eric E J G L Smeets
European Journal of Human Genetics, April 2015, 23(4):543-6.
… Genome-wide copy number profiling was done using high-resolution CytoScan HD arrays with 2.7 million markers, according to the manufacturer's protocol (Affymetrix ...

23- *Meeting abstract*
A Specific Pattern of Somatic Mutations Associates with Poor Prognosis Aneuploid Acute Myeloid Leukemia: Results from the European NGS-PTL Consortium
Giorgia Simonetti, PhD*,1, Antonella Padella*,1, Ítalo Faria do Valle*,2, Marco Manfrini, PhD*,3, Cristina Papayannidis, MD PhD3, Carmen Baldazzi, PhD*,1, Maria Chiara Fontana*,4, Viviana Guadagnuolo, PhD*,1, Anna Ferrari, PhD*,1, Elisa Zago, PhD*,5, Marianna Garonzi*,6, Simona Bernardi*,7, Emanuela Ottaviani*,1, Annalisa Astolfi, PhD*,8, Maria Chiara Abbenante, MD*,1, Giovanni Marconi, MD*,1, Simona Soverini9, Michele Cavo, MD*,10, Nicoletta Testoni*,1, Alberto Ferrarini, PhD*,6, Massimo Delledonne, PhD*,5, Torsten Haferlach11, Daniel Remondini, PhD*,2, Ilaria Iacobucci, PhD*,10, and Giovanni Martinelli10
1University of Bologna, Bologna, Italy ; 2Dept of Physics and Astronomy, University of Bologna, Bologna, Italy ; 3Bologna University School of Medicine, Bologna, Italy ; 4"Seràgnoli" Institute of Hematology, Bologna University School of Medicine, Bologna, Italy ; 5Personal Genomics, Verona, Italy ; 6Dept of Biotechnology, University of Verona, Verona, Italy ; 7Unit of Blood Diseases and Stem Cells Transplantation, University of Brescia, Brescia, Italy ; 8Centro Interdipartimentale per la Ricerca sul Cancro "G. Prodi", University of Bologna, Bologna, Italy ; 9Institute of Hematology, S.Orsola-Malpighi University Hospital, Bologna, Italy ; 10"Seràgnoli" Institute of Hematology, University of Bologna, Bologna, Italy ; 11MLL Munich Leukemia Laboratory, Munich, Germany
Blood, 3 December 2015, 126(23): 3840.
… AML samples were genotyped by CytoScan HD Array (Affymetrix). Gene expression profiling (GEP) was also conducted on bone marrow cells from 24 A-AML, 33 E-AML (≥80% blasts) and 7 healthy controls (HTA 2.0, Affymetrix). ...

24-
A t(3;9)(q25.1;q34.3) translocation leading to OLFM1 fusion transcripts in Gilles de la Tourette syndrome, OCD and ADHD
Birgitte Bertelsen, Linea Melchior, Lars Riff Jensen, Camilla Groth, Lusine Nazaryan, Nanette Mol Debes, Liselotte Skov, Gangcai Xie, Wei Sun, Karen Brøndum-Nielsen, Andreas Walter Kuss, Wei Chen, Zeynep Tümer
Psychiatry Research, 28 February 2015, 225(3):268–275.
... Chromosome microarray was carried out using Affymetrix Genome-Wide Human SNP Array 6.0 (translocation patient) or Affymetrix CytoScan HD (GTS-cohort), and data were analyzed using Genotyping Console Software or Chromosome Analysis Suite software (Affymetrix ...

25- *Open access*
A Three-Generation Family with Idiopathic Facial Palsy Suggesting an Autosomal Dominant Inheritance with High Penetrance
Christian Grønhøj Larsen,1 Mette Gyldenløve,2 Aia Elise Jønch,3 Birgitte Charabi,1 and Zeynep Tümer4
1Department of Otorhinolaryngology, Head and Neck Surgery, Copenhagen University Hospital of Rigshospitalet, 2100 Copenhagen, Denmark; 2Department of Dermato-Allergology, Gentofte Hospital, University of Copenhagen, 2900 Hellerup, Denmark; 3Clinical Genetic Clinic, Kennedy Center, Copenhagen University Hospital,

Rigshospitalet, 2600 Glostrup, Denmark; 4Applied Human Molecular Genetics, Kennedy Center, Copenhagen University Hospital, Rigshospitalet, 2600 Glostrup, Denmark
Case Reports in Otolaryngology, 2015, Volume 2015:Article# 683938.
... After two weeks, the patient had almost fully recovered with only minor muscle weakness remaining, which was still present two months later. Chromosome microarray analysis was carried out using CytoScan HD microarray platform (Affymetrix, USA) but did not reveal a microdeletion or duplication segregating with the disease in the family.

26-
A Transition Zone Showing Highly Discontinuous or Alternating Levels of Stem Cell and Proliferation Markers Characterizes the Development of PTEN-Haploinsufficient Colorectal Cancer
Kevin J. Arvai, Ya-Hsuan Hsu, Lobin A. Lee, Dan Jones
PLoS ONE, 22 June 2015, 10(6): e0131108.
... After macrodissection of FFPE tumor sections, genomic DNA was extracted using the QiaAmp DNA FFPE Tissue Kit (Qiagen, Valencia, CA) and assessed using the CytoScan HD 2.6 million-probe microarray platform (Affymetrix, Santa Clara, CA) according to the manufacturer's instructions.

27-
Absence of Heterozygosity due to Template Switching during Replicative Rearrangements
Claudia M.B. Carvalho, Rolph Pfundt, Daniel A. King, Sarah J. Lindsay, Luciana W. Zuccherato, Merryn V.E. Macville, Pengfei Liu, Diana Johnson, Pawel Stankiewicz, Chester W. Brown, DDD Study, Chad A. Shaw, Matthew E. Hurles, Grzegorz Ira, P.J. Hastings, Han G. Brunner, James R. Lupski
American Journal of Human Genetics, 2 April 2015, 96(4): 555–564.
... DNA from subjects BAB3922, BAB3923, and BAB3924 plus parental DNA samples were hybridized on an Affymetrix GeneChip 250k (NspI) SNP array platform or CytoScan array. ... To confirm triplication and AOH independently, samples were run on two different array platforms. For instance, BAB4539 plus parental samples were run on an Affymetrix CytoScan SNP array and samples BAB3922, BAB3923, and BAB3924 plus parental samples were analyzed with an Illumina array HumanOmniExpress-24 Beadchip at the Human Genome Sequencing Center of Baylor College of Medicine.

28-
Acro-spondylo-pubic dysostosis associated with cataracts, microcephaly, and normal intelligence
Oscar F. Chacon-Camacho1, Vanessa Villegas-Ruiz1, Beatriz Buentello-Volante1, Raul E. Piña-Aguilar2, Hugo Peláez-González3, Magdalena Ramírez3, Johanna González-Rodríguez1, Juan Carlos Zenteno1,4,*
1Genetics Dept-Research Unit, Institute of Ophthalmology "Conde de Valenciana", Mexico City, Mexico; 2Dept of Genetics, Centro Médico Nacional "20 de Noviembre", ISSSTE,, Mexico City, Mexico; 3Radiology Dept, Hospital General de Mexico, Mexico City, Mexico; 4Biochemistry Dept, Faculty of Medicine, National Autonomous University of Mexico (UNAM), Mexico City, Mexico
American Journal of Medical Genetics Part A, Feb 2015, 167(2):282–286.
... Molecular karyotyping was performed on the patient's DNA using the genome-wide high-resolution SNP array CytoScan HD (Affymetrix, Santa Clara, CA, USA) chip.

29-
Acrofacial Dysostosis, Cincinnati Type, a Mandibulofacial Dysostosis Syndrome with Limb Anomalies, Is Caused by POLR1A Dysfunction
K. Nicole Weaver1, 9, Kristin E. Noack Watt2, 3, 9, Robert B. Hufnagel1, Joaquin Navajas Acedo2, Luke L. Linscott4, Kristen L. Sund1, Patricia L. Bender1, Rainer König5, Charles M. Lourenco6, Ute Hehr7, Robert J. Hopkin1, Dietmar R. Lohmann8, Paul A. Trainor2, 3, 10, Dagmar Wieczorek8, 10, Howard M. Saal1, 10
1 Div of Human Genetics, Dept of Pediatrics, Cincinnati Children's Hospital Medical Center and University of Cincinnati College of Medicine, MLC 4006, 3333 Burnet Ave, Cincinnati, OH 45229, USA; 2 Stowers Institute for Medical Research, 1000 East 50th Street, Kansas City, MI 64110, USA; 3 University of Kansas Medical Center, Kansas City, MI 66160, USA; 4 Dept of Radiology, Cincinnati Children's Hospital Medical Center, Cincinnati, OH 45229, USA; 5 Institut für Humangenetik, Universitätsklinikum Frankfurt, Theodor-Stern-Kai 7, 60596 Frankfurt, Germany; 6 Neurogenetics Unit, Clinics Hospital of Ribeirao Preto, University of Sao Paulo, Avenue Bandeirantes 3900, Sao Paulo 14049-900, Brazil; 7 Zentrum für Humangenetik, Universitätsklinikum Regensburg, Franz-Josef-StrauB-Allee 11, 93053 Regensburg, Germany; 8 Institut für Humangenetik, Universitätsklinikum Essen, Universität Duisburg-Essen, Hufelandstr 55, 45122 Essen, Germany
American Journal of Human Genetics, 7 May 2015, 96(5):765–774.
... Individual 1A2 had a normal karyotype and Affymetrix Cytoscan HD Array.

30-

Additional patients with 4q deletion: Severe growth delay and polycystic kidney disease associated with 4q21q22 loss

Satoru Sakazume1,*, Yasuhiro Kido1, Nobuyuki Murakami1, Tomoyo Matsubara1, Hironao Numabe2

1Dept of Pediatrics, Koshigaya Hospital, Dokkyo Medical University, Koshigaya, Japan; 2Dept of Genetic Counseling, Graduate School of Human Genetics and Science, Ochanomizu University, Tokyo, Japan

Pediatrics International, October 2015, 57(5): 880–883.

... this is the third report concerning the 4q21q22 deletions. ... we used the same array platform to analyze genetic anomaly of both patients. ... performed at a commercial laboratory (LSI Medience Corporation Japan) using the CytoScan HD array (Affymetrix) ...

31-

Adult Low-Hypodiploid Acute B-Lymphoblastic Leukemia With IKZF3 Deletion and TP53 Mutation

Comparison With Pediatric Patients

Min Fang, MD, PhD1,2,3, Pamela S. Becker, MD, PhD1,2, Michael Linenberger, MD1,2, Keith D. Eaton, MD, PhD1,2,3, Frederick R. Appelbaum, MD1,2,3, ZoAnn Dreyer, MD4, Gladstone Airewele, MD4, Michele Redell, MD, PhD4, Dolores Lopez-Terrada, MD, PhD4, Ankita Patel, PhD5, Karen R. Rabin4, Xinyan Lu, MD6

1From the Fred Hutchinson Cancer Research Center, Seattle, WA; 2University of Washington, Seattle; 3Seattle Cancer Care Alliance, Seattle, WA; 4Texas Children's Cancer and Hematology Centers and Dept of Pediatrics, Baylor College of Medicine, Houston; 5Dept of Molecular and Human Genetics, Baylor College of Medicine, Houston, TX; 6Dept of Hematopathology, University of Texas MD Anderson Cancer Center, Houston, TX.

American Journal of Clinical Pathology, 2015, 144:263-270.

...[chromosome genomic array testing] was critical in the genotype clarification of these cases ...and should be considered performing for B-ALL ... Adult DNA was processed using the CytoScanHD kit from Affymetrix ...Pediatric DNA was assayed using Infinium Cyto12 from Illumina ...

32-

Alternating hypoglycemia and hyperglycemia in a toddler with a homozygous p.R1419H ABCC8 mutation: an unusual clinical picture

Shira Harel1 / Ana S.A. Cohen2, 3 / Khalid Hussain4, 5 / Sarah E. Flanagan4 / Kamilla Schlade-Bartusiak3, 6 / Millan Patel2, 3 / Jaques Courtade3 / Jenny B.W. Li7 / Clara Van Karnebeek8 / Harley Kurata7 / Sian Ellard4 / Jean-Pierre Chanoine1 / William T. Gibson2, 3

1Endocrinology and Diabetes Unit, Dept of Pediatrics, British Columbia Children's Hospital, Univ of British Columbia, Vancouver, Canada; 2Dept of Medical Genetics, University of British Columbia, Vancouver, Canada; 3Child and Family Research Institute, Vancouver, Canada; 4Institute of Biomedical and Clinical Science, Univ of Exeter Medical School, Exeter, UK; 5Developmental Endocrinology Research Group, Clinical and Molecular Genetics, Institute of Child Health, Univ College London, London, UK; 6Dept of Pathology and Laboratory Medicine, Univ of British Columbia, Vancouver, Canada; 7Dept of Anesthesiology, Pharmacology and Therapeutics, Univ of British Columbia, Vancouver, Canada; 8Div of Biochemical Diseases, Dept of Pediatrics, British Columbia Children's Hospital, Univ of British Columbia, Vancouver, Canada

Journal of Pediatric Endocrinology and Metabolism, March 2015 Mar, 28(3-4):345-351.

... This is the first description of a homozygous p.R1419H mutation. Our findings highlight that homozygous loss-of-function mutations of ABCC8 do not necessarily translate into early-onset severe hyperinsulinemia.

... CytoScanHD (Affymetrix, Inc., Santa Clara, CA, USA) single nucleotide polymorphism (SNP) array analysis was done ...

33-

An apoptosis-enhancing drug overcomes platinum resistance in a tumour-initiating subpopulation of ovarian cancer

D. M. Janzen, E. Tiourin, J. A. Salehi, D. Y. Paik, J. Lu, M. Pellegrini & S. Memarzadeh

Nature Communications, 3 August 2015, 6:Article #7956

... DNA was isolated from S#-GODL cells and their respective parent tumours ... then hybridized to the Affymetrix CytoScan HD array ...

34-

An infant with large fontanelles, aplasia cutis congenita, tessier facial cleft, polydactyly inversus, and toe syndactyly: A previously undescribed syndrome?

Jessica Jackson1,*, Paula Delk1, Emily Farrow2, Christopher Griffith1, Melissa Lah1 andDavid D. Weaver1

1Dept of Medical and Molecular Genetics, Indiana University School of Medicine, Indianapolis, Indiana; 2Center for Pediatric Genomic Medicine, Children's Mercy Hospital, Kansas City, Missouri

American Journal of Medical Genetics Part A, April 2015, 167(4):683–687.

... Results of a constitutional chromosomal microarray analysis (CMA) using the Cytoscan HD platform (Affymetrix, Santa Clara, CA) was also normal (Indiana University Cytogenetics Laboratory, Indianapolis, IN).

35-
Anirdia-like phenotype caused by 6p25 dosage aberrations
Karthikeyan Arcot Sadagopan1,2, Grace T. Liu1,3,4,*, Jenina E. Capasso1, Wadakarn Wuthisiri1,5, Rosanne B. Keep1,5, Alex V. Levin1,6,7
1Ocular Genetics, Wills Eye Institute, Philadelphia, PA; 2Dept of Pediatric Ophthalmology, Strabismus and Ocular Genetics, Ocular Genetics Service, Aravind Eye Hospital, Madurai, India; 3Pediatric Ophthalmic Consultants, New York City, NY; 4Dept of Ophthalmology, New York University, New York City, NY; 5Dept of Ophthalmology, Faculty of Medicine, Ramathibodi Hospital Mahidol University, Bangkok, Thailand; 6Quest Diagnostics, Madison, NJ; 7Thomas Jefferson University, Philadelphia, PA
American Journal of Medical Genetics Part A, March 2015, 167(3):524–528
... In Patient 2, cytogenetic analysis with a 550 band resolution revealed a male karyotype with a 6p25.1 interstitial duplication, 46XY, dup(6)(25.1p25.3). SNP microarray analysis (Affymetrix Cytoscan HD platform) using 743,000 SNP probes ...

36-
Aortopathy in the 7q11.23 microduplication syndrome
Ashley Parrott1, Jeanne James1, Paula Goldenberg1, Robert B. Hinton1, Erin Miller1,*, Amy Shikany1, Arthur S. Aylsworth2,3, Kathleen Kaiser-Rogers2,3,4, Sunita J. Ferns2, Seema R. Lalani5, Stephanie M. Ware6
1Dept of Pediatrics, Cincinnati Children's Hospital Medical Center, Heart Institute, Cincinnati, OH; 2Dept of Pediatrics, University of North Carolina, Chapel Hill, NC; 3Dept of Genetics, University of North Carolina, Chapel Hill, NC; 4Dept of Pathology and Laboratory Medicine, University of North Carolina, Chapel Hill, NC; 5Dept of Molecular and Human Genetics, Baylor College of Medicine, Houston, TX; 6Dept of Pediatrics and Medical and Molecular Genetics, Indiana University School of Medicine, Indianapolis, Indiana
American Journal of Medical Genetics Part A, Feb 2015, 167(2): 363–370.
... The 7q11.23 microduplication in patient 9 was also identified using a SNP microarray platform (Affymetrix CytoScan HD, Santa Clara, CA) and confirmed by both metaphase and interphase FISH using RP11–19F19 probe (Empire Genomics Buffalo, NY).

37- *Article in Chinese*
[Application of chromosome microarray analysis for fetuses with increased nuchal translucency and a normal karyotype]
Yang X, Fu F, Li R, Zhang Y, Wan J, Yang X, Han J, Pan M, Zhen L, Liao C.
Zhonghua Yi Xue Yi Chuan Xue Za Zhi. (Chinese Journal of Medical Genetics), June 10, 2015, 32(3):370-4.
... Genomic DNA was extracted, and microarray testing was performed using Affymetrix CytoScan™ HD arrays.

38- *Article in Chinese*
[Application of whole-genome and high-resolution chromosome microarray analysis for the investigation of fetuses with ultrasound abnormalities].
Zhang Y, Fu F, Li R, Xie G, Han J, Pan M, Zhen L, Yang X, Li D, Liao C.
Zhonghua Yi Xue Yi Chuan Xue Za Zhi. (Chinese Journal of Medical Genetics), 2015 Apr;32(2):169-74.
... Whole genome high-resolution CytoScanHD array from Affymetrix was employed to investigate 651 fetuses with structural abnormalities detected by ultrasound, for whom standard G-banded chromosome analysis has revealed a normal karyotype. ...

39-
Assessing the utility of confirmatory studies following identification of large-scale genomic imbalances by microarray.
Sanmann JN, Pickering DL, Golden DM, Stevens JM, Hempel TE, Althof PA, Wiggins ML, Starr LJ, Davé BJ, Sanger WG.
Genetics in Medicine, November 2015, 17(11):875-879.
... Table 1 Microarray platforms and resolutions ... 1.9 million copy-number markers and 750,000 genotypable SNPs: Affymetrix Cytoscan HD (Santa Clara, CA) ...

40-
Association of an α-Globin Gene Cluster Duplication and Heterozygous β-Thalassemia in a Patient with a Severe Thalassemia Syndrome
Hua Jiang, Sha Liu, Yong-Ling Zhang, Jun-Hui Wan, Ru Li, and Dong-Zhi Li

1Department of Hematology/Oncology, Guangzhou Women & Children Medical Center affiliated to Guangzhou Medical University, Guangzhou, Guangdong, People's Republic of China; 2Prenatal Diagnostic Center, Guangzhou Women & Children Medical Center affiliated to Guangzhou Medical University, Guangzhou, Guangdong, People's Republic of China

Hemoglobin, online 18 February 2015, 39(2): 102-106.

... Array comparative genomic hybridization (CGH) (Affymetrix CytoScan® High-Density Array; Affymetrix Inc., Santa Clara, CA, USA) was also used to confirm the α-globin gene rearrangement.

41-

BCL6 – regulated by AhR/ARNT and wild-type MEF2B – drives expression of germinal center markers MYBL1 and LMO2

Jie Ding,1 Wilhelm G Dirks,1 Stefan Ehrentraut,1 Robert Geffers,2 Roderick AF MacLeod,1 Stefan Nagel,1 Claudia Pommerenke,1 Julia Romani,1 Michaela Scherr,3 Lea Al Vaas,1 Margarete Zaborski,1 Hans G Drexler,1 and Hilmar Quentmeier1

1Leibniz-Institute DSMZ-German Collection of Microorganisms and Cell Cultures, Braunschweig, Germany; 2Helmholtz Centre for Infection Research, Genome Analysis Research Group,Braunschweig, Germany; 3Medical School Hannover, Department of Hematology, Hemostasis, Oncology and Stem Cell Transplantation, Hannover, Germany

Haematologica, June 2015, 100(6): 801-809.

... CytoScan HD Array (Affymetrix; Santa Clara, CA, USA) hybridization analysis was performed to identify numerical aberrations.

42-

Bone marrows from neuroblastoma patients: An excellent source for tumor genome analyses

M. Reza Abbasi a, Fikret Rifatbegovic a, Clemens Brunner a, Ruth Ladenstein b, c, Inge M. Ambros a, Peter F. Ambros a, c

a Tumor Biology, Children's Cancer Research Institute, St. Anna Kinderkrebsforschung, Vienna, Austria; b SiRP, Children's Cancer Research Institute, St. Anna Kinderkrebsforschung, Vienna, Austria; c Dept of Pediatrics, Medical University of Vienna, Vienna, Austria

Molecular Oncology, March 2015, 9(3): 545–554.

... Genomic profile of tumor cells was examined by CytoScan™ HD Array (Affymetrix Inc., UK Ltd), a genome-wide UHD-SNP array technique with more than 2.6 million copy number and SNP markers.

43-

Catel–Manzke syndrome: Further delineation of the phenotype associated with pathogenic variants in TGDS

Rachel Pferdehirt a, Mahim Jain a, Maria A. Blazo b, Brendan Lee a, Lindsay C. Burrage a

a Dept of Molecular and Human Genetics, Baylor College of Medicine, Houston, TX 77030; b Dept of Genetics, Baylor Scott & White Health, Texas A&M Health Science Center College of Medicine, Temple, TX 76508, USA

Molecular Genetics and Metabolism Reports, September 2015, 4: 89–91

... Routine chromosome analysis, comparative genomic hybridization microarray (Affymetrix Cytoscan HD Microarray System, Scott and White Healthcare Molecular Genetics Laboratories), ... only the second publication describing molecularly confirmed Catel–Manzke syndrome, ...

44-

CD19-negative B-lymphoblastic leukemia associated with hypercalcemia, lytic bone lesions and aleukemic presentation

Shafinaz Husseina, Kerice Pinkneyb, Vaidehi Jobanputraa, Govind Bhagata & Bachir Alobeida*

Leukemia & Lymphoma, 2015, 56(5): 1533-1537.

... SNP-array analysis, using the Affymetrix CytoScan HD Array (Affymetrix, Santa Clara, CA) performed on DNA extracted from the BM biopsy sample targeting the lytic lesion, revealed no abnormalities.

45-

Cerebral visual impairment, autism, and pancreatitis associated with a 9 Mbp deletion on 10p12.

Bosch, Daniëlle G.M.a,b,c,d; Boonstra, Frouke N.a,c; Pfundt, Rolphb; Cremers, Frans P.M.b,d; de Vries, Bert B.A.b,c

Clinical Dysmorphology, January 2015, 24(1):34-37.

... Genome- wide array analysis was carried out on DNA from peripheral blood using the Affymetrix CytoScan HD array platform (Affymetrix ...

46-

Characterization of a Complex Chromosomal Rearrangement Involving a de novo Duplication of 9p and 9q and a Deletion of 9q

Martín-De Saro M.D.a · Valdés-Miranda J.M.b · Plaza-Benhumea L.c · Pérez-Cabrera A.b · Gonzalez-Huerta L.M.b · Guevara-Yañez R.d · Cuevas-Covarrubias S.A.b
aDept of Medical Genetics, Hospital Materno Infantil ISSEMyM, Toluca, bDept of Medical Genetics, School of Medicine, General Hospital of Mexico, National Autonomous University of Mexico, cGenetics Dept, Children's Hospital IMIEM, and 4Laboratorio BIOGEN, Mexico City, Mexico
Cytogenet Genome Res, 2015, 147:124-129
… we present a CCR in a patient with multiple congenital anomalies who represents the first case with partial 9p trisomy, partial 9q trisomy and partial 9q monosomy. … analysis was carried out using CytoScan HD array …

47-
Chromosomal microarrays testing in children with developmental disabilities and congenital anomalies
Lay-Son G1, Espinoza K2, Vial C2, Rivera JC2, Guzmán ML3, Repetto GM3.
1Center for Human Genetics, Facultad de Medicina, Clínica Alemana Universidad del Desarrollo, Santiago, Chile; Hospital Padre Hurtado, Santiago, Chile. Electronic address: glayson@udd.cl.; 2Center for Human Genetics, Facultad de Medicina, Clínica Alemana Universidad del Desarrollo, Santiago, Chile.; 3Center for Human Genetics, Facultad de Medicina, Clínica Alemana Universidad del Desarrollo, Santiago, Chile; Hospital Padre Hurtado, Santiago, Chile.
J Pediatr (Rio J), March-April 2015, 91(2):189-95.
... Chilean patients with developmental disabilities and congenital anomalies were studied with a high-density microarray (CytoScan™ HD Array, Affymetrix, Inc., Santa Clara, CA, USA).

48-
Chromothriptic Cure of WHIM Syndrome
David H. McDermott, Ji-Liang Gao, Qian Liu, Marie Siwicki, Craig Martens, Paejonette Jacobs, Daniel Velez, Erin Yim, Christine R. Bryke, Nancy Hsu, Zunyan Dai, Martha M. Marquesen, Elina Stregevsky, Nana Kwatemaa, Narda Theobald, Debra A. Long Priel, Stefania Pittaluga, Mark A. Raffeld, Katherine R. Calvo, Irina Maric, Ronan Desmond, Kevin L. Holmes, Douglas B. Kuhns, Karl Balabanian, Françoise Bachelerie, Stephen F. Porcella, Harry L. Malech, Philip M. Murphy
Cell, 12 February 2015, 160(4): 686–699.
... Affymetrix Cytoscan HD array ... was used to identify chromosomal breakpoints .... monoclonal Abs used were CD45.1- PECy7 and CD45.2-eFlour450 from eBioscience ... and biotinylated CD34-streptavidin-PE (clone RAM) from eBioscience.

49-
Clinical and molecular characterization of an infant with a tandem duplication and deletion of 19p13
Ratna N. G. B. Tan1, Ruben S. G. M. Witlox1, Yvonne Hilhorst-Hofstee2, Cacha M. P. C. D. Peeters-Scholte3, Nicolette S. den Hollander2, Claudia A. L. Ruivenkamp2, Mariëtte J.V. Hoffer2, Kerstin B. Hansson2, Mark J. van Roosmalen4, Wigard P. Kloosterman4 and Gijs W. E. Santen2,*
1Dept of Pediatrics, Leiden University Medical Center, Leiden, the Netherlands; 2Dept of Clinical Genetics, Leiden University Medical Center, Leiden, the Netherlands; 3Dept of Neurology, Leiden University Medical Center, Leiden, the Netherlands; 4Center for Molecular Medicine, Dept of Medical Genetics, University Medical Center Utrecht, Utrecht, the Netherlands
American Journal of Medical Genetics Part A, August 2015, 167(8): 1884–1889.
... A Cytoscan HD array (Affymetrix, Santa Clara, CA, USA) was performed according to the manufacturer's instructions. Fluorescence in situ hybridization (FISH) analysis was carried out ...

50-
Clinical and molecular cytogenetic studies of an unrecognised 22q11.2 deletion in three families
LINHUAN HUANG,1,* YINGJUN XIE,1,* YI ZHOU,1 YANMIN LUO,1 XUAN HUANG,1 ZHE XU,2 DANLEI CAI,3 and QUN FANG1
1Fetal Medicine Centre, Dept of Obstetrics and Gynaecology, The First Affiliated Hospital of Sun Yat-Sen University, Guangzhou, Guangdong 510080, P.R. China; 2Div of Cardiac Surgery, First Affiliated Hospital of Sun Yat-Sen Univ, Guangzhou, Guangdong 510080, P.R. China; 3Dept of Ultrasonic Medicine, First Affiliated Hospital of Sun Yat-Sen Univ, Guangzhou, Guangdong 510080, P.R. China
Exp Ther Med. 2015 Mar; 9(3): 823–828.
... The DNA samples (250 ng) were hybridised to CytoScan HD arrays (Affymetrix®, Santa Clara, CA, USA) according to the manufacturer’s instructions.

51- *First CytoScan publication from Mexico*
Clinical and Molecular Characterization of a Patient with 15q21.2q22.2 Deletion Syndrome

Velázquez-Wong A.C.a· Ruiz Esparza-Garrido R.a, b· Velázquez-Flores M.Á.a· Huicochea-Montiel J.C.c· Cárdenas-Conejo A.c· Miguez-Muñoz C.P.e· Araujo-Solís M.A.c· Salamanca-Gómez F.d· Arenas-Aranda D.J.a

aUnidad de Investigación Médica en Genética Humana, bCátedras CONACyT, Consejo Nacional de Ciencia y Tecnología, cServicio de Genética, Hospital de Pediatría, and dCoordinación de Investigación en Salud, Centro Médico Nacional Siglo XXI, Instituto Mexicano del Seguro Social (IMSS), Mexico City, Mexico; eAffymetrix, Latinoamérica, São Paulo, Brazil

Cytogenetic and Genome Reserch, 2014: 144:183-189. Posted 6 Feb 2015.

... The girl was clinically diagnosed with probable Prader-Willi syndrome. Chromosomal analysis showed a de novo deletion 46,XX,del(15)(q21q22). However, the use of the Affymetrix CytoScan HD Array defined the exact breakpoints of the deleted 15q21q22 region. ...

52-

Clinical characterization of a male patient with the recently described 8q21.11 microdeletion syndrome

Ines Quintela1, Francisco Barros2,*, Manuel Castro-Gago3, Angel Carracedo1,2,4, Jesus Eiris3

1Grupo de Medicina Xenomica, Universidade de Santiago de Compostela, Centro Nacional de Genotipado, Plataforma de Recursos Biomoleculares y Bioinformaticos, Instituto de Salud Carlos III (CeGen-PRB2-ISCIII), Santiago de Compostela, Spain; 2Grupo de Medicina Xenomica-USC, CIBERER, Fundacion Publica Galega de Medicina Xenomica-SERGAS, Santiago de Compostela, Spain; 3Departamento de Pediatria, Hospital Clinico Universitario de Santiago de Compostela, Unidad de Neurologia Pediatrica, Santiago de Compostela, Spain; 4King Abdulaziz University, Center of Excellence in Genomic Medicine Research, Jeddah, Saudi Arabia

American Journal of Medical Genetics Part A, June 2015, 167(6): 1369–1373.

... DNA samples from both the patient and his mother were obtained from peripheral blood and genotyped using the Cytogenetics Whole-Genome 2.7 M SNP array and the CytoScan High-Density SNP array (Affymetrix, Santa Clara, CA), respectively.

53-

Clinical Severity of PGK1 Deficiency Due To a Novel p.E120K Substitution Is Exacerbated by Co-inheritance of a Subclinical Translocation t(3;14)(q26.33;q12), Disrupting NUBPL Gene

Dezső David(1), Lígia S. Almeida(1), Maristella Maggi(2)(3), Carlos Araújo(1), Stefan Imreh(4), Giovanna Valentini(2), György Fekete(5), Irén Haltrich(5)

1. Dept of Human Genetics, National Institute of Health Dr Ricardo Jorge, Lisbon, Portugal ; 2. Dept of Biology and Biotechnology “L. Spallanzani”, University of Pavia, Pavia, Italy ; 3. Dept of Molecular Medicine, Unit of Immunology and Pathology, University of Pavia, Pavia, Italy ; 4. Microbiology and Tumour Biology Center, Karolinska Institute, Stockholm, Sweden ; 5. II Dept of Pediatrics, Semmelweis University, Budapest, Hungary

JIMD Reports, 2015, 23: 55-65.

... Genomic amplicons of der(3) and der(14) chromosomes were analyzed by array painting using CytoScan HD array (Affymetrix, Santa Clara, CA, USA).

54-

Clinically relevant copy number variations detected in cerebral palsy

Maryam Oskoui, Matthew J. Gazzellone,Bhooma Thiruvahindrapuram, Mehdi Zarrei, John Andersen, John Wei, Zhuozhi Wang, Richard F. Wintle, Christian R. Marshall, Ronald D. Cohn, Rosanna Weksberg, Dimitri J. Stavropoulos, Darcy Fehlings, Michael I. Shevell & Stephen W. Scherer

Nature Communications, 3 August 2015, 6: Article #7949.

... We utilized additional population control samples ... (both cohorts were run on the Affymetrix 6.0 microarray); ... Ontario Population Genetics Platform (from the CytoScan HD).

55-

Clival Encephalocele and 5q15 Deletion: A Case Report

Surasak Puvabanditsin, MD1, Imran Malik, MD1, Eugene Garrow, MD2, Lissa Francois, MD3, Rajeev Mehta, MD, FRCP1

1Dept of Pediatrics, Rutgers Robert Wood Johnson Medical School, New Brunswick, NJ, USA ; 2Dept of Surgery, SUNY Downstate Medical Center, Brooklyn, New York, NY, USA ; 3Dept of Obstetrics and Genecology, Rutgers RWJ Medical School, New Brunswick, NJ, USA

Journal of Child Neurology, Mar 2015; 30:505-508.

… We report a rare variant of a basal encephalocele … microarray analysis was performed … on a whole-genome array (Affymetrix CytoScan HD platform) showed an interstitial deletion of 496 oligonucleotide probe at 5q15 …

56-

CNV-ROC: A cost effective, computer-aided analytical performance evaluator of chromosomal microarrays

Corey W. Goodman a, c, Heather J. Majore, William D. Wallsa, c, Val C. Sheffieldd, e, Thomas L. Casavanta, b, c, d, Benjamin W. Darbro e

a Dept of Electrical and Computer Engineering, University of Iowa; b Dept of Biomedical Engineering, University of Iowa; c Center for Bioinformatics and Computational Biology, Univ of Iowa; d Ph.D. Program in Genetics, Univ of Iowa; e Dept of Pediatrics, Univ of Iowa

Journal of Biomedical Informatics, April 2015, 54: 106–113.

Chromosomal microarrays (CMAs) are routinely used in both research and clinical laboratories; yet, little attention has been given to the estimation of genome-wide true and false negatives during the assessment of these assays and how such information could be used to calibrate various algorithmic metrics to improve performance. Low-throughput, locus-specific methods such as fluorescence in situ hybridization (FISH), quantitative PCR (qPCR), or multiplex ligation-dependent probe amplification (MLPA) preclude rigorous calibration of various metrics used by copy number variant (CNV) detection algorithms. To aid this task, we have established a comparative methodology, CNV-ROC, which is capable of performing a high throughput, low cost, analysis of CMAs that takes into consideration genome-wide true and false negatives. … comparing one of the NimbleGen CGH microarrays to an array manufactured by Affymetrix (CytoScan HD) ... Forty samples were used for comparisons, ... 20 additional samples for the 720K vs. CytoScan HD microarray comparisons.

57-

Comparative mapping of the 22q11.2 deletion region and the potential of simple model organisms

Alina Guna1, Nancy J. Butcher12 and Anne S. Bassett12345*

1 Clinical Genetics Research Program and Campbell Family Mental Health Research Institute, Centre for Addiction and Mental Health, Toronto, ON, Canada ; 2 Institute of Medical Science, University of Toronto, Toronto, ON, Canada ; 3 Dalglish Family Hearts and Minds Clinic for Adults with 22q11.2 Deletion Syndrome, Div of Cardiology, Dep of Medicine, Dept of Psychiatry, and Toronto General Research Institute, University Health Network, Toronto, ON, Canada ; 4 Dept of Psychiatry, University of Toronto, Toronto, ON, Canada ; 5 Centre for Addiction and Mental Health, 33 Russell Street, Room 1100, Toronto, M5S 2S1, ON, Canada

Journal of Neurodevelopmental Disorders, 1 July 2015, 7:18

... The human 22q11.2DS deletion region, genetic content, and order were mapped from NCBI Gene Homo sapiens Annotation Release 105 using Affymetrix CytoScan HD (Santa Clara, CA, USA) array mean breakpoints (chr22:18,820, 303–21, 489,474) ascertained from 16 patients with confirmed 22q11.2 deletions (Fig. 1).

58-

Comprehensive gene panels provide advantages over clinical exome sequencing for Mendelian diseases

Saudi Mendeliome Group

Saudi Human Genome Project, King Abdulaziz City for Science and Technology, Riyadh, Saudi Arabia

Genome Biology, 26 June 2015, 16:134

... we used SNP genotyping arrays (Affymetrix Axiom GT1 chip ... coming from 21 patients as a second method of testing the analytical sensitivity. ... CytoScan HD (Affymetrix) arrays was used for the majority of our patients. ...

59-

Comprehensive genetic analysis of a pediatric pleomorphic myxoid liposarcoma reveals near-haploidization and loss of the RB1 gene

Jakob Hofvander3,*, Vickie Y. Jo1, Iman Ghanei2, David Gisselsson3, Emma Mårtensson3, Fredrik Mertens3

1Dept of Pathology, Brigham and Women's Hospital, Harvard Medical School, Boston, MA; 2Dept of Orthopedics, Skåne University Hospital, Lund University, Lund, Sweden; 3Dept of Clinical Genetics, University and Regional Laboratories, Skåne University Hospital, Lund University, Lund, Sweden

Histopathology, Accepted manuscript online: 9 Dec 2015.

... Banding analysis revealed a hyperdiploid/hypotriploid karyotype that at SNP array analysis could be shown to derive from a near-haploid ancestral clone ... analyzed using the Affymetrix CytoScan HD array (Affymetrix, ...

60-

Comprehensive Pipeline for Analyzing and Visualizing Agilent and Affymetrix Array-Based CGH Data

Frederic Commo

Inserm U981, Bioinformatics Group, Gustave Roussy, France

White paper, July 9, 2015

... We present *rCGH*, a comprehensive array-based CGH analysis workflow, integrating functionalities specifically designed for precision medicine. *rCGH* ensures a full traceability by saving all the process parameters, and facilitates genomic profiles interpretation and decision-making through interactive visualizations. *rCGH* supports Agilent (from 44K to 400K arrays), as well as Affymetrix, SNP6 and cytoScanHD arrays.

61-
Concurrent triplication and uniparental isodisomy: evidence for microhomology-mediated break-induced replication model for genomic rearrangements
Trilochan Sahoo, Jia-Chi Wang, Mohamed M Elnaggar, Pedro Sanchez-Lara, Leslie P Ross, Loretta W Mahon, Katayoun Hafezi, Abigail Deming, Lynne Hinman, Yovana Bruno, James A Bartley, Thomas Liehr, Arturo Anguiano and Marilyn Jones
European Journal of Human Genetics, 2015, 23:61–66.
… We report on the identification and characterization of two cases with interstitial triplication followed by uniparental isodisomy (isoUPD) for remainder of the chromosomal arm. … The above two cases were identified from over 14500 cases analyzed utilizing the Affymetrix version 6.0 genechip (5700 cases; … or the Affymetrix CytoScan HD Array (8800 cases; whole-genome 2.7M array) …

62-
Confirmation of a maternal cryptal balanced translocation through analysis of a fetus using microarray **[article in Chinese]**
Wu J1, Xie Y, Lin S, Chen B, Chen J, Zhang Z, Ji Y.
1Fetal Medicine Center, Department of Gynaecology and Obstetrics, The First Affiliated Hospital, Sun Yatsen University, Guangzhou, Guangdong 510080, P. R. China. Email: wujianzhu3140@126.com.
Zhonghua Yi Xue Yi Chuan Xue Za Zhi. [Chinese Journal of Medical Genetics}, 10 Feb 2015, 32(1):69-72.
doi: 10.3760/cma.j.issn.1003-9406.2015.01.015.

OBJECTIVE To analyze a fetus with heart defects and to assess the recurrence risk for her family. METHODS Single nucleotide polymorphism-based arrays (SNP-Array) analysis using Affymetrix Genome Wide Human SNP CytoHD was performed to analyze the fetus and her parents. Karyotype analysis was also carried out. RESULTS SNP-Array has detected a 14.5 Mb duplication at 9p and a 14.7 Mb deletion at 11q. Karyotype analysis indicated that the fetus' mother has a karyotype of 46,XX,t(9;11)(p23;q24). Therefore, the fetus has inherited a derivative chromosome 11 derived from the maternal translocation, and her karyotype was 46,XX,der(11)t(9;11)(p23;q24)mat. CONCLUSION SNP-Array combined with high resolution GTG banding has confirmed that the fetus has a derivative chromosome 11 derived from her mother's balanced translocation, resulting in partial 9p trisomy and partial 11q monosomy. This couple therefore have a high recurrence risk. SNP-Array is capable of detecting small chromosomal imbalance in abnormal fetuses and can pinpoint the breakpoints. It therefore has the advantage for the detection of unbalanced translocation which is difficult to detect with GTG banding, which is important for assessment the recurrence risk for cryptic balanced translocation carriers.

63-
Copy number variants including RAS pathway genes—How much RASopathy is in the phenotype?
Christina Lissewski1, Sarina G. Kant2, Zornitza Stark3, Ina Schanze1, Martin Zenker1,*
1Institute of Human Genetics, University Hospital Magdeburg, Magdeburg, Germany; 2Department of Clinical Genetics, Leiden University Medical Center, Leiden, The Netherlands; 3Victorian Clinical Genetics Service, MCRI, Royal Children's Hospital, Melbourne, Australia
American Journal of Medical Genetics Part A, November 2015, 167(11): 2685–269.
… Molecular karyotyping was performed in the index patient using CytoScan HD Array Affymetrix and revealed a duplication 19p13.3 (chr19:3947246-5098336, hg19) of at least 1.2 Mb.

64-
Copy number variations in cryptogenic cerebral palsy
Reeval Segel, MD*, Hilla Ben-Pazi, MD*, Sharon Zeligson, Aviva Fatal-Valevski, MD, Adi Aran, MD, Varda Gross-Tsur, MD, Nira Schneebaum-Sender, MD, Dorit Shmueli, MD, Dorit Lev, MD, Shira Perlberg, Luba Blumkin, MD, Lisa Deutsch, PhD and Ephrat Levy-Lahad, MD
From the Medical Genetics Institute (R.S., S.Z., S.P., E.L.-L.) and Neuropediatric Unit (H.B.-P., A.A., V.G.-T.), Shaare Zedek Medical Center, Jerusalem; Pediatric Neurology Unit (A.F.-V., N.S.-S.), Dana Children's Hospital, Tel Aviv; Jerusalem Child Development Center (D.S.), Clalit, Jerusalem; Metabolic-Neurogenetic Clinic (D.L., L.B.), Wolfson Medical Center, Holon; and Biostatistical Consulting (L.D.), BioStats, Israel.
Neurology April 21, 2015 vol. 84 no. 16 1660-1668

…Participants underwent neurologic and clinical genetic examinations before the genomic testing. Chromosomal microarray analysis to detect CNVs was performed using the Affymetrix platform. …

We recommend CNV testing in individuals with CP of unknown etiology. … Affymetrix Cytoscan HD arrays were used ...

65-
CRB2 Mutations Produce a Phenotype Resembling Congenital Nephrosis, Finnish Type, with Cerebral Ventriculomegaly and Raised Alpha-Fetoprotein
Anne Slavotinek,1,2,* Julie Kaylor,3 Heather Pierce,4 Michelle Cahr,5 Stephanie J. DeWard,4
Dina Schneidman-Duhovny,6,7 Adnan Alsadah,1 Fadi Salem,8 Gabriela Schmajuk,9 and Lakshmi Mehta5
American Journal of Human Genetics, 8 Jan 2015, 96:162–169.
We report five fetuses and a child from three families who shared a phenotype comprising cerebral ventriculomegaly and echogenic kidneys with histopathological findings of congenital nephrosis. … His last renal US continued to show diffusely echogenic kidneys with ascites. A SNP microarray (CytoScanHD,Affymetrix) was normal. ...

66-
Cytogenetic, fluorescence in situ hybridization and genomic array characterization of chronic myeloid leukemia with cryptic BCR/ABL1 fusions
Lina Shao, Sue Miller, Jennifer Keller-Ramey#, Yang Zhang, Diane Roulston
Clinical Cytogenetics Laboratory, Department of Pathology, University of Michigan, Ann Arbor, Michigan
Cancer Genetics, July-Aug 2015, 208(7–8): 396–403.
… This study demonstrates for the first time the acquisition of additional BCR/ABL1 fusion genes through mitotic recombination in CML with cryptic BCR/ABL1. ... Genome-wide SNP array analysis using the Affymetrix CytoScan HD platform ...

67-
De novo 9q gain in an infant with tetralogy of Fallot with absent pulmonary valve: Patient report and review of congenital heart disease in 9q duplication syndrome
Ina E. Amarillo1, Shawn O'Connor2, Caroline K. Lee2, Marcia Willing2 and Jennifer A. Wambach2,*
1Dept of Pathology and Immunology, Cytogenomics Laboratory, Washington University in St. Louis School of Medicine, St. Louis, Missouri; 2Dept of Pediatrics, Washington University in St. Louis School of Medicine, St. Louis, Missouri
American Journal of Medical Genetics Part A, December 2015, 167(12): 2966–2974.
... first report implicating RXRA in CHD with 9q duplication ... highlights the significance of CMA in the clinical diagnosis ... Genomic DNA was isolated from cord blood lymphocytes of the infant (proband) for CMA and data output was analyzed using CytoScan HD (Affymterix, ...

68-
De novo microduplication of CHL1 in a patient with non-syndromic developmental phenotypes
Orazio Palumbo1, Rita Fischetto2, Pietro Palumbo1, Francesco Nicastro2, Francesco Papadia2, Leopoldo Zelante1 and Massimo Carella1*
1 Laboratorio di Genetica Medica, IRCCS Casa Sollievo della Sofferenza, San Giovanni Rotondo, (FG), Italy; 2 Unità Operativa Malattie Metaboliche Genetica Medica, P.O. Giovanni XXIII, A.O.U. Policlinico Consorziale, Bari, Italy
Molecular Cytogenetics, 16 August 2015, 8:66.
... We extracted DNA from the lymphocytes of patient and his parents using BioRobot EZ1 (Qiagen, Solna, Sweden). Genomic screening for copy number variations (CNVs) was carried out using the CytoScan HD array platform (Affymetrix, ...

69-
Deleción en el gen RPS6KA3 en una mujer con un fenotipo clásico del síndrome de Coffin-Lowry incluyendo episodios de caída inducidos por estímulo
Inés Quintela b, Francisco Barros-Angueira b, Laura Pérez-Gay d, Manuel Castro-Gago d, Ángel Carracedo a,b,c, Jesús Eirís-Puñal d
a King Abdulaziz University. Center of Excellence in Genomic Medicine Research. Jeddah, Arabia Saudí. b Grupo de Medicina Xenómica. Universidade de Santiago de Compostela. Centro Nacional de Genotipado, Plataforma de Recursos Biomoleculares y Bioinformáticos, Instituto de Salud Carlos III (CeGen-PRB2-ISCIII). c Grupo de Medicina Xenómica-USC, CIBERER, Fundación Pública Galega de Medicina Xenómica- SERGAS. d Departamento de Pediatría. Unidad de Neurología Pediátrica. Hospital Clínico Universitario de Santiago de Compostela. Santiago de Compostela, A Coruña, España.
Rev Neurol 2015; 61(2): 94-96
... Las muestras de ADN de la paciente y su madre se obtuvieron de sangre periférica y se genotiparon con el panel Affymetrix CytoScan High- Density (Affymetrix, Santa Clara, CA) ...

70-
Deletion of 7q33-q35 in a Patient with Intellectual Disability and Dysmorphic Features: Further Characterization of 7q Interstitial Deletion Syndrome
Kristen Dilzell,1 Diana Darcy,2 John Sum,3 and Robert Wallerstein2
1Dept of Medical Genetics, University of Pennsylvania, Philadelphia, PA 19104, USA; 2Silicon Valley Genetics Center, Santa Clara Valley Medical Center, San Jose, CA 95128, USA; 3Pediatric Neurology, Santa Clara Valley Medical Center, San Jose, CA 95128, USA
Case Reports in GeneticsVolume 2015 (2015), Article ID 131852.
This case report concerns a 16-year-old girl with a 9.92 Mb, heterozygous interstitial chromosome deletion at 7q33-q35, identified using array comparative genomic hybridization. … SNP microarray was performed by Integrated Genetics using the Affymetrix Cytoscan HD platform ...

71-
Deletion of 11q12.3–11q13.1 in a patient with intellectual disability and childhood facial features resembling Cornelia de Lange syndrome
Martine Isabel Boyle, Cathrine Jespersgaard, Lusine Nazaryan, Kirstine Ravn, Karen Brøndum-Nielsen, Anne-Marie Bisgaard, Zeynep Tümer
Applied Human Molecular Genetics, Kennedy Center, Dept of Clinical Genetics, Univ of Copenhagen, Rigshospitalet, Glostrup, Denmark
Gene, Available online 8 July 2015, In Press, Corrected Proof
... Deletions within 11q12.3-11q13.1 are very rare ... a 1.6 Mb deletion at chromosome region 11q12.3-11q13.1 was detected by chromosome microarray. ... Chromosome microarray analysis was performed in the proband using the Affymetrix CytoScan HD platform ...

72-
Deletion of 16q24.1 Supports a Role for the ATP2C2 Gene in Specific Language Impairment
Amena W. Smith, MD, PhD1, Kenton R. Holden, MD1,2,3, Alka Dwivedi, PhD3, Barbara R. Dupont, PhD3, Michael J. Lyons, MD3⇑
1Dept of Neurosciences (Neurology), Medical University of South Carolina, Charleston, SC, USA ; 2Dept of Pediatrics, Medical University of South Carolina, Charleston, SC, USA ; 3Greenwood Genetic Center, Greenwood, SC, USA
J Child Neurol, March 2015, 30(4):517-521.
A 10-year-old boy presented with a history of significant delay in language acquisition … Chromosomal microarray analysis and quantitative polymerase chain reaction determined that he had a de novo 159-kilobase deletion of chromosome 16q24.1 that included the ATP2C2 gene. …: Whole-genome chromosomal microarray analysis (Affymetrix Cytoscan HD Microarray system) revealed a 159-kilobase …

73-
Detailed analysis of therapy-driven clonal evolution of TP53 mutations in chronic lymphocytic leukemia
J Malcikova, K Stano-Kozubik, B Tichy, B Kantorova, S Pavlova, N Tom, L Radova, J Smardova, F Pardy, M Doubek, Y Brychtova, M Mraz, K Plevova, E Diviskova, A Oltova, J Mayer, S Pospisilova and M Trbusek
Leukemia, 2015, 29: 877–885.
… Deep sequencing can shift TP53 mutation identification to a period before therapy administration, which might be of particular importance for clinical trials.... cn-LOH 17p—17p copy-neutral loss of heterozygosity involving TP53 gene detected by Cytoscan Affymetrix arrays.

74-
Detailed Biochemical and Bioenergetic Characterization of FBXL4-Related Encephalomyopathic Mitochondrial DNA Depletion
Ghadi Antoun (1), Skye McBride (2), Jason R. Vanstone (2), Turaya Naas (2) (4), Jean Michaud (3), Stephanie Redpath (2), Hugh J. McMillan (2), Jason Brophy (2), Hussein Daoud (2), Pranesh Chakraborty (2) (4), David Dyment (2), Martin Holcik (2), Mary-Ellen Harper (1), Matthew A. Lines mlines@cheo.on.ca (2)
1. Dept of Biochemistry, Microbiology, and Immunology, Faculty of Medicine, University of Ottawa, Ottawa, ON, Canada; 2. Children's Hospital of Eastern Ontario Research Institute, Ottawa, ON, Canada; 3. Dept of Pathology and Laboratory Medicine, Children's Hospital of Eastern Ontario and Faculty of Medicine, University of Ottawa, Ottawa, ON, Canada; 4. Newborn Screening Ontario, Ottawa, ON, Canada
JIMD Reports, online 25 September 2015
... Microarray was performed clinically (Affymetrix CytoScan HD) according to standard protocols.

75-
Detecting copy-number variations in whole-exome sequencing data using the eXome Hidden Markov Model: an 'exome-first' approach
Satoko Miyatake1, Eriko Koshimizu1, Atsushi Fujita1, Ryoko Fukai1, Eri Imagawa1, Chihiro Ohba1, Ichiro Kuki2, Megumi Nukui2, Atsushi Araki3, Yoshio Makita4, Tsutomu Ogata5, Mitsuko Nakashima1, Yoshinori Tsurusaki1, Noriko Miyake1, Hirotomo Saitsu1 and Naomichi Matsumoto1
1Dept of Human Genetics, Yokohama City University Graduate School of Medicine, Yokohama, Japan; 2Dept of Pediatric Neurology, Pediatric Medical Care Center, Osaka City General Hospital, Osaka, Japan; 3Dept of Pediatrics, Kansai Medical University, Hirakata, Japan; 4Education Center, Asahikawa Medical University, Asahikawa, Japan; 5Dept of Pediatrics, Hamamatsu University School of Medicine, Hamamatsu, Japan
Journal of Human Genetics, 2015, 60: 175–182.
… we performed WES CNV analysis using the eXome Hidden Markov Model (XHMM). We validated its performance using 27 rare CNVs previously identified by microarray as positive controls, ... Either Affymetrix CytoScan HD or Human Mapping 250 K Nsp Array for Cytogenetics (Affymetrix, Santa Clara, CA, USA) were used for all the 21 samples.

76-
DGCR6 at the proximal part of the DiGeorge critical region is involved in conotruncal heart defects
Wenming Gao, Takashi Higaki, Minenori Eguchi-Ishimae, Hidehiko Iwabuki, Zhouying Wu, Eiichi Yamamoto, Hidemi Takata, Masaaki Ohta, Issei Imoto, Eiichi Ishii, Mariko Eguchi
Human Genome Variation, online 12 Feb 2015, 2, Article #15004
… when copy number alteration was analyzed by high-resolution array analysis, a small deletion or duplication in the proximal end of DiGeorge critical region was detected in two patients. … Copy number analysis was performed using a genome-wide high-resolution Affymetrix CytoScan HD array …

77-
Diagnosis of 9q22.3 microdeletion syndrome in utero following identification of craniosynostosis, overgrowth, and skeletal anomalies
Sara Chadwick Reichert1,*, Kristin Zelley2, Kim E. Nichols3, Moriah Eberhard1, Elaine H. Zackai4 andJuan Martinez-Poyer1
1Center for Fetal Diagnosis and Treatment, Children's Hospital of Philadelphia, Philadelphia, PA; 2Div of Oncology, Children's Hospital of Philadelphia, Philadelphia, PA; 3Div of Cancer Predisposition, St Jude Children's Research Hospital, Memphis, TN; 4Div of Clinical Genetics, Children's Hospital of Philadelphia, Philadelphia, PA
American Journal of Medical Genetics Part A, April 2015, 167(4):862–865.
…We describe a patient with prenatally diagnosed 9q22.3 microdeletion syndrome following the ultrasonographic identification of trigonocephaly, macrosomia, organomegaly, ventriculomegaly, and anomalous vertebrae. … Follow-up parental SNP microarray was performed on whole blood by Quest Diagnostics Nichols Institute using the Affymetrix Cytoscan HD platform.

78-
Diagnosis of D-Bifunctional Protein Deficiency through Whole-Genome Sequencing: Implications for Cost-Effective Care
Khromykh A.a· Solomon B.D.a,b,d· Bodian D.L.a· Leon E.L.f· Iyer R.K.a, e· Baker R.L.b, c· Ascher D.P.b, d· Baveja R.b, c· Vockley J.G.a, c· Niederhuber J.E.a
aInova Translational Medicine Institute, bInova Children's Hospital, Inova Health System, and cFairfax Neonatal Associates, Falls Church, Va., Depts of dPediatrics and eObstetrics and Gynecology, Virginia Commonwealth University School of Medicine, Richmond, VA, and fDiv of Genetics and Metabolism, Children's National Medical Center, Washington, D.C., USA
Molecular Syndromology, 2015, 6:141-146.
... Commercially performed cytogenetic or molecular testing on peripheral blood included: initial FISH for Trisomy 21, karyotyping, high-density microarray (oligonucleotide-single nucleotide polymorphism microarray via Affymetrix CytoScan HD), spinal muscular atrophy via ...

79-
Diagnosis of intrachromosomal amplification of chromosome 21 (iAMP21) by molecular cytogenetics in pediatric acute lymphoblastic leukemia
Nicolas Duployez1,2,3,*, Elise Boudry-Labis1,3, Gauthier Decool2, Guillaume Grzych2, Nathalie Grardel2, Wadih Abou Chahla4, Claude Preudhomme2,3 and Catherine Roche-Lestienne1,3
1Medical Cytogenetics Institute, Jeanne de Flandre Hospital, Lille, France; 2Laboratory of Hematology, Biology and Pathology Center, Lille, France; 3INSERM UMR-S 1172, Cancer Research Institute, Lille, France; 4Pediatric Hematology Department, Jeanne de Flandre Hospital, Lille, France
Clinical Case Reports, October 2015, 3(10): 814–816.

... Single-nucleotide polymorphism (SNP)-array was performed ... using Affymetrix® Cytoscan High Density Array. ... SNP-array is a useful method to confirm the accuracy of iAMP21 diagnosis ... its use is of increasing importance to refine risk stratification in pediatric BCP-ALL ...

80-
Diagnostic value of multiple café-au-lait macules for neurofibromatosis 1 in Chinese children
Ruen Yao1, Lili Wang2, Yongguo Yu2, Jian Wang1 and Yiping Shen1,3,4,*
1Shanghai Children's Medical Center, Shanghai Jiaotong University School of Medicine, Shanghai, China; 2Xinhua Hospital, Institute for Pediatric Research, Shanghai, China; 3Boston Children's Hospital, Boston, MA, USA; 4Claritas Genomics, Cambridge, MA, USA.
The Journal of Dermatology, first published online: 13 October 2015
... Molecular testing is necessary ... as an adjunct and sometimes as the main tool for confirming and diagnosing children of NF1 at early age ... Chromosomal microarray analysis was applied to detect large structural variants using a CytoScan HD® chip (Affymetrix, ...

81-
Differentiation of genetic abnormalities in early pregnancy loss
S. T. Romero1,2,*, K. B. Geiersbach3, C. N. Paxton4, N. C. Rose2, E. F. Schisterman5, D. W. Branch2 and R. M. Silver1
1Dept of Obstetrics and Gynecology, University of Utah School of Medicine, Salt Lake City, UT, USA; 2Div of Maternal Fetal Medicine, Intermountain Medical Center, Salt Lake City, UT, USA; 3Dept of Pathology, University of Utah School of Medicine, Salt Lake City, UT, USA; 4ARUP Institute for Clinical and Experimental Pathology®, University of Utah School of Medicine, Salt Lake City, UT, USA; 5Epidemiology Branch, Eunice Kennedy Shriver National Institute of Child Health and Human Development, National Institutes of Health, Bethesda, MD
Ultrasound in Obstetrics & Gynecology [Special issue: Fetal cfDNA and microarray], January 2015, 45(1):89–94.
… Chromosomal microarray analysis (CMA) was performed on 74 samples (including two samples from a twin pregnancy); 15 were pre-embryonic (no visible embryo on ultrasound examination), 31 were embryonic (embryo; 6 + 0 to 9 + 6 weeks' gestation) and 28 were fetal (fetus; 10 + 0 to 19 + 6 weeks' gestation) losses. … Labeled DNA fragments were denatured and hybridized to CytoScan SNP arrays (Affymetrix ...

82-
Differing Microdeletion Sizes and Breakpoints in Chromosome 7q11.23 in Williams-Beuren Syndrome Detected by Chromosomal Microarray Analysis
Li L., Huang L., Luo Y., Huang X., Lin S., Fang Q.
Dept of Gynecology and Obstetrics, Fetal Medicine Center, First Affiliated Hospital of Sun Yat-sen University, Guangzhou, PR China
Molecular Syndromology, 2015, 6: 268-275.
... We analyzed 10 patients diagnosed with 7q11.23 microdeletion syndrome by chromosomal microarray analysis. ... hybridized to the Affymetrix CytoScan HD array chip ...

83-
Digital PCR Validates 8q Dosage as Prognostic Tool in Uveal Melanoma
Mieke Versluis, Mark J. de Lange , Sake I. van Pelt, Claudia A. L. Ruivenkamp, Wilma G. M. Kroes, Jinfeng Cao, Martine J. Jager, Gre P. M. Luyten, Pieter A. van der Velden
PLoS ONE, March 12, 2015, 10(3): e0116371.
… We used SNP microarray data that was acquired for clinical purposes on UM samples to determine chromosomal aberrations. Two types of SNP microarray chips were used. The Affymetrix 250K_NSP, chip, which contains roughly 250 000 probes across the genome and the Affymetrix Cytoscan HD chip, with approximately 750 000 probes across the genome. A first set of 28 samples was analyzed with the Affymetrix 250K_NSP chip. Since this chip was no longer available, the remaining 36 samples were measured with the Affymetrix Cytoscan HD chip.

84-
Discordant noninvasive prenatal testing and cytogenetic results: a study of 109 consecutive cases
Jia-Chi Wang MD, PhD, Trilochan Sahoo MD, Steven Schonberg PhD, Kimberly A. Kopita MS, Leslie Ross MS, Kyla Patek MS & Charles M. Strom MD, PhD
Genetics in Medicine, March 2015, 17(3):234-236.
… These findings raise concerns about the limitations of noninvasive prenatal testing and the need for analysis of a larger number of false-positive cases to provide true positive predictive values … and/or oligo–single-nucleotide polymorphism microarrays (CytoScanHD; Affymetrix, Santa Clara, CA) after NIPT.

85-

Discovery of a novel cystathionine-beta-synthase mutation and diagnosis of homocystinuria by whole exome sequencing in a family from rural Honduras

Scott A. Turner1, Mary Beth P. Dinulos2, 3, Stephanie E. Vallee2, 3, Linda Kennedy4, Peter Mason5, Dean Seibert5, Marco Tulio Martinez4, Heather B. Steinmetz2, Gregory J. Tsongalis1, 2, 4, Joel A. Lefferts1, 2

1Dept of Pathology, Geisel School of Medicine at Dartmouth, Hanover, NH, USA. 2Dept of Pathology, Dartmouth-Hitchcock Medical Center, Lebanon, NH, USA. 3Dept of Pediatrics, Dartmouth-Hitchcock Medical Center, Lebanon, NH. 4Norris Cotton Cancer Center, Geisel School of Medicine at Dartmouth, Hanover, NH, USA. 5Dept of Medicine, Geisel School of Medicine at Dartmouth, Hanover, NH, USA.

Case Reports in Clinical Pathology, published online 17 April 2015, 2(3):59-65.

… Microarray analysis was done using CytoScanHD Array (Affymetrix, Santa Clara, CA) and Chromosomal Analysis Suite (ChAS) v1.2 (Affymetrix) software.

86-

Distal 22q11.2 microduplication combined with typical 22q11.2 proximal deletion: A case report

Miriam Coelho Molck1, Társis Paiva Vieira1, Milena Simioni1, Ilária Cristina Sgardioli1, Ana Paula dos Santos1, Ana Carolina Xavier2 and Vera Lúcia Gil-da-Silva-Lopes1,3,*

1Dept of Medical Genetics, Faculty of Medical Sciences, University of Campinas (UNICAMP), Sao Paulo, Brazil; 2Center for Research and Rehabilitation of Lip and Palate Lesions (CRRLPL) Centrinho Prefeito Luiz Gomes, Joinville, Santa Catarina, Brazil; 3Assistance Center for Cleft Lip and Palate (CAIF), Curitiba, Parana, Brazil

American Journal of Medical Genetics Part A, January 2015,167(1): 215-220.

… Array genomic hybridization (aGH) analysis for the patient was performed using the Genome-Wide Human SNP Array 6.0 (Affymetrix®) and analyzed by GeneChip® Operating Software (GCOS) (Affymetrix®). aGH analysis for patient's mother and father was performed using the CytoScan HD chip (Affymetrix®) ...

87-

Duplication Xp11.22-p14 in females: Does X-inactivation help in assessing their significance?

Christina Evers1, Diana Mitter2, Gertrud Strobl-Wildemann3, Ulrich Haug4, Karl Hackmann5, Bianca Maas1, Johannes W. G. Janssen1, Anna Jauch1, Katrin Hinderhofer1, Ute Moog1,*

1Institute of Human Genetics, Heidelberg University, Heidelberg, Germany; 2Institute of Human Genetics, University Hospital Leipzig, Leipzig, Germany; 3Humangenetik Ulm MVZ, Ulm, Germany; 4Center for Child Neurology and Social Pediatrics Maulbronn, Maulbronn, Germany; 5Institut fuer Klinische Genetik, Medizinische Fakultaet Carl Gustav Carus, Technische Universitaet Dresden, Dresden, Germany

American Journal of Medical Genetics Part A, March 2015, 167(3):553–562.

... Array analysis of Patient 1 and her parents was performed using the Affymetrix® CytoScan HD Array. Patients 2 and 3 and parental samples were analyzed with the Affymetrix® Genome-Wide Human SNP Array 2.7 ...

88-

Effects of Integrating and Non-Integrating Reprogramming Methods on Copy Number Variation and Genomic Stability of Human Induced Pluripotent Stem Cells

Xiangjin Kang, Qian Yu, Yuling Huang, Bing Song, Yaoyong Chen, Xingcheng Gao, Wenyin He, Xiaofang Sun, Yong Fan

PLoS ONE, 1 July 2015, 10(7): e0131128.

... using the Affymetrix Cytoscan HD array, we investigated the genomic aberration profiles of 19 human cell lines ... from which the iPSCs were derived. ... importance of determining the molecular mechanisms ... in reprogramming somatic cells to be pluripotent ...

89-

EMT-associated factors promote invasive properties of uveal melanoma cells

Laura Asnaghi,1 Gülçin Gezgin,2 Arushi Tripathy,1 James T. Handa,3 Shannath L. Merbs,3,4 Pieter A. van der Velden,2 Martine J. Jager,2 J. William Harbour,5 Charles G. Eberhart1,3,4

1Dept of Pathology, Johns Hopkins University, School of Medicine, Baltimore, MD; 2Dept of Ophthalmology, Leiden University Medical Center, Leiden, The Netherlands; 3Dept of Ophthalmology, Johns Hopkins University, School of Medicine, Baltimore, MD; 4Dept of Oncology, Johns Hopkins University, School of Medicine, Baltimore, MD; 5Ocular Oncology, Bascom Palmer Eye Institute and Sylvester Comprehensive Cancer Center, University of Miami Miller School of Medicine, Miami, FL

Molecular Vision, 25 August 2015, 21:919-929.

... The Affymetrix 250K_NSP chip was used to analyze 28 tumor specimens. The remaining 36 tumor specimens were analyzed with the Affymetrix Cytoscan HD chip ...

90- *Meeting abstract*

Establishment, characterization, and clinical correlation of a platform of ovarian patient-derived xenograft (PDX) models

Sangeetha S. Palakurthi1, Joyce F. Liu2, Qing Zeng1, Shan Zhou1, Wei Huang1, Elena Ivanova1, Cloud Paweletz1, John R. Murgo1, Justin Evangelista1, Melissa Buttimer1, Jennifer Curtis3, Huiying Piao2, Prafulla Gokhale1, Colin Pritchard4, Jessie M. English1, Paul Kirschmeier1, Kwok-Kin Wong1, Ursula A. Matulonis2, and Ronny Drapkin5
1Belfer Institute for Applied Cancer Science, DFCI, Boston, MA; 2Dept of Medical Oncology, Dana-Farber Cancer Institute, Boston, MA; 3Dept of Cancer Biology, Dana-Farber Cancer Institute, Boston, MA; 4Dept of Laboratory Medicine, University of Washington, Seattle, WA; 5Dept of Medical Oncology, Dana-Farber Cancer Institute and Department of Pathology, Brigham and Women's Hospital, Boston, MA.

Cancer Research, 2015, 75(15 Suppl):Abst #1471. [AACR 106th Annual Mtg, Apr 18-22, 2015, Philadelphia, PA]

... Ovarian PDXs were established by implanting mice intraperitoneally with fresh human ovarian cancer cells purified from operative or paracentesis samples under an IRB-approved protocol. ... DNA from patient material and matched established PDX models was extracted and subjected to CNV on the Affymetrix Cytoscan™ HD microarray platform and BROCA cancer risk panel targeted sequencing. Data from these analyses indicated that the PDXs are molecularly diverse and faithfully maintain the genetic alterations and copy number variation profiles of the patient tumors. ...

91- *Meeting abstract*

Evaluation of SNP Genomic Microarray Analysis as an Alternative to FISH Analysis of Pediatric Solid Tumors

Anthony Arnoldo, James Stavropoulos, Paul Thorner, Cynthia Hawkins, Gino R. Somers, Mary Shago

Cancer Genetics, June 2015, 208(6): 361–362.

... We have validated the use of the genomic microarray platform Affymetrix Cytoscan HD and Chromosome Analysis Suite (ChAS) software as an alternative to FISH analysis for the assessment of copy number changes in these tumors.

92-

Evidence for the multiple hits genetic theory for inherited language impairment: a case study

Tracy M. Centanni, Jordan R. Green, Jenya Iuzzini-Seigel, Christopher W. Bartlett, Tiffany P. Hogan

Frontiers in Genetics, 24 August 2015, 6: 272.

... High-resolution genome-wide analysis was performed on genomic DNA using the CytoScanHD™ array (Affymetrix, Santa Clara, CA, USA) according to manufacturer's instruction.

89- *Open access*

Exceptional Complex Chromosomal Rearrangements In Three Generations

Hannie Kartapradja, Nanis Sacharina Marzuki, Mark Pertile, David Francis, Lita Putri Suciati, Helena Woro Anggaratri, Debby Dwi Ambarwati, Firman Prathama Idris, Harry Lesmana, Hidayat Trimarsanto, Chrysantine Paramayuda, and Alida Roswita Harahap

Case Reports in Genetics, accepted 19 January 2015.

We report an exceptional Complex Chromosomal Rearrangement (CCR) found in three individuals in a family that involves 4 chromosomes with 5 breakpoints. The CCR was ascertained in a phenotypically abnormal newborn with additional chromosomal material on the short arm of chromosome 4. Maternal karyotyping indicated that the mother carried an apparently balanced CCR involving chromosomes 4, 6, 11 and 18. Maternal transmission of the derivative chromosome 4 resulted in partial trisomy for chromosomes 6q and 18q, and a partial monosomy of chromosome 4p in the proband. Further family studies found that the maternal grandmother carried the same apparently balanced CCR as the proband's mother, which was confirmed using whole chromosome painting (WCP) FISH. High resolution whole genome microarray analysis of DNA from the proband's mother found no evidence for copy number imbalance in the vicinity of the CCR translocation breakpoints, or elsewhere in the genome, providing evidence that the mother and grandmother's CCRs were balanced at a molecular level. This structural rearrangement can be categorized as an

exceptional CCR due to its complexity and is a rare example of an exceptional CCR being transmitted in balanced and/or unbalanced form across three generations.
Excerpt: To investigate whether the BCCR was balanced at a molecular level, we analysed DNA from the proband's mother using Affymetrix CytoScan HD microarray (Affymetrix, Santa Clara, CA, USA), with interpretation based on the NCBI36/hg18 (March 2006) human reference sequence. The microarray result showed no clinically significant genomic imbalance.

90-
Exome sequencing identifies a novel heterozygous TGFB3 mutation in a disorder overlapping with Marfan and Loeys-Dietz syndrome
Alma Kuechler, Janine Altmüller, Peter Nürnberg, Stefan Kotthoff, Christian Kubisch, Guntram Borck
Molecular and Cellular Probes, Available online 13 July 2015, Accepted Manuscript
Marfan syndrome (MFS) and Loeys-Dietz syndrome (LDS) are clinically related autosomal dominant systemic connective tissue disorders. ...We performed whole exome sequencing in two of three affected individuals from a family with phenotypic features overlapping MFS and LDS. ... Molecular karyotyping (Affymetrix CytoscanHD array) was also normal.

91-
Exome sequencing of osteosarcoma reveals mutation signatures reminiscent of BRCA deficiency
Michal Kovac, Claudia Blattmann, Sebastian Ribi, Jan Smida, Nikola S. Mueller, Florian Engert, Frances Castro-Giner, Joachim Weischenfeldt, Monika Kovacova, Andreas Krieg, Dimosthenis Andreou, Per-Ulf Tunn, Hans Roland Dürr, Hans Rechl, Klaus-Dieter Schaser, Ingo Melcher, Stefan Burdach, Andreas Kulozik, Katja Specht, Karl Heinimann, Simone Fulda, Stefan Bielack, Gernot Jundt, Ian Tomlinson, Jan O. Korbel, Michaela Nathrath & Daniel Baumhoer
Nature Communications, 3 December, 6: Article #8940.
... Affymetrix CytoscanHD arrays were processed with ChaS 2.1 and Nexus 7.5 software such that SCNA events larger than 50 kb with a minimum support of 21 probes were considered for subsequent analysis.

92-
Expanding the Molecular and Clinical Phenotype of SSR4-CDG
Bobby G. Ng1, Kimiyo Raymond2, Martin Kircher3, Kati J. Buckingham4, Tim Wood5, Jay Shendure3, Deborah A. Nickerson3, Michael J. Bamshad3,4, University of Washington Center for Mendelian Genomics1, JonathanT.S. Wong1, Fabiola Paoli Monteiro6,7, Brett H. Graham8, Sheryl Jackson9, Rebecca Sparkes9, Angela E. Scheuerle10, Sara Cathey5, Fernando Kok7,11, James B. Gibson12 and Hudson H. Freeze1,*
1Human Genetics Program, Sanford – Burnham – Prebys Medical Discovery Institute, La Jolla, CA; 2Biochemical Genetics Laboratory, Mayo Clinic College of Medicine, Rochester, MN; 3Dept of Genome Sciences, University of Washington, Seattle, WA; 4Dept of Pediatrics, University of Washington, Seattle, WA; 5Dept of Clinical Genetics, Greenwood Genetic Center, Charleston Office, North Charleston, SC; 6Dept of Medical Genetics, University of Campinas (UNICAMP), São Paulo, Brazil; 7Mendelics Genomic Analysis, São Paulo, São Paulo, Brazil; 8Dept of Molecular and Human Genetics, Baylor College of Medicine, Houston, TX; 9Dept of Medical Genetics, University of Calgary, Calgary, AB, Canada; 10Dept of Pediatrics, University of Texas Southwestern Medical Center, Dallas, TX; 11Dept of Neurology, University of São Paulo, São Paulo, Brazil; 12Clinical and Metabolic Genetics, Specially for Children, Austin, TX
Human Mutation, November 2015, 36(11): 1048–1051.
... Family 405 is of European ancestry and are the only family in which exome sequencing was not performed. Instead, an Affymetrix CytoScan HD microarray was performed at Greenwood Genetics Center and showed that the affected male (P4) was ...

93-
Familial transmission of 5p13.2 duplication due to maternal der(X)ins(X;5)
Lauren C. Walters-Sen, Kathy Windemuth, Katie Angione, Jenisha Nandhlal, Jeff M. Milunsky,
Center for Human Genetics, Inc., Cambridge, MA, USA
European Journal of Medical Genetics, May 2015, 58(5):305–309.
... first report of an inherited duplication of 5p13.2 with multiple affected family members. ... Analysis by SNP 6.0 Microarray (Affymetrix ... in the consultand discovered a 341 kb duplication at 5p13.2 ... the proband's array was performed using the Cytoscan HD ...

94-
First contiguous gene deletion causing biotinidase deficiency: The enzyme deficiency in three Sri Lankan children **[open access]**

Danika Nadeen Senanayake a, Eresha. A. Jasinge b, Kirit Pindolia c, d, Jithangi Wanigasinghe e, Kristin Monaghan f, 1, Sharon F. Suchy f, Sainan Wei g, Subashini Jaysena b, Barry Wolf c, d

a Dept of Chemical Pathology, North Columbo Teaching Hospital, Columbo, Sri Lanka; b Chemical Pathology, Lady Ridgeway Hospital for Children, Colombo, Sri Lanka; c Dept of Research Administration, Henry Ford Health System, Detroit, MI 48202; d Center for Molecular Medicine and Genetics, Wayne State Univ School of Medicine, Detroit, MI 48201; e Pediatric Neurology, University of Colombo, Columbo, Sri Lanka; f GeneDx, Gaithersburg, MD 20877; g Dept of Pediatrics and Human Development, Michigan State Univ, East Lansing, MI 48824

Molecular Genetics and Metabolism Reports, May 2015, 2: 81-84.

We report three symptomatic children with profound biotinidase deficiency from Sri Lanka. All three children presented with typical clinical features of the disorder. … High density SNP microarray studies were performed using Affymetrix Cytoscan HD SNP Array™.

95-

Five patients with a chromosome 1q21.1 triplication show macrocephaly, increased weight and facial similarities

Anke Van Dijck a, b, Ilse M. van der Werf a, Edwin Reyniers a, b, Stefaan Scheers a, b, Meron Azage c, Kiana Siefkas d, Nathalie Van der Aa a, Amy Lacroix e, Jill Rosenfeld f, Bob Argiropoulos g, h, Kellie Davis g,A.Micheil Innes g, h, Heather C. Mefford d, e, Geert Mortier a, b, Marije Meuwissen b, R.Frank Kooy a

a Dept of Medical Genetics, University of Antwerp, Belgium; b Dept of Medical Genetics, University Hospital Antwerp, Belgium; c Dept of Medical Genetics, Children's Hospital of Pittsburgh, PA, USA; d Dept of Medical Genetics, Seattle Children's Hospital, Seattle, WA, USA; e Dept of Pediatrics, University of Washington, Seattle, WA, USA; f Signature Genomic Laboratories, Spokane, WA, USA; g Dept of Medical Genetics, Cumming School of Medicine, University of Calgary, Calgary, Alberta, Canada; h Alberta Children's Hospital Research Institute, Cumming School of Medicine, University of Calgary, Calgary, Alberta, Canada

European Journal of Medical Genetics, October 2015, 58(10): 503–508

... For the first time, we describe five patients, including monozygotic twins, with a triplication of the 1q21.1 chromosomal segment. ... SNP array analysis of the patient C-II.1 was performed with an Affymetrix Cytoscan HD. ...

96-

Genetic alterations in renal cell carcinoma with rhabdoid differentiation

Carmen M. Perrino, MDa, 1, Vishwanathan Hucthagowder, PhDa, 1, Michael Evenson, BSa, Shashikant Kulkarni, PhDa, Peter A. Humphrey, MD, PhDb*

*Dept of Pathology, Yale School of Medicine, 310 Cedar St, LB11, PO Box 208023, New Haven, CT, 06520-8023.

Human Pathology, January 2015, 46(1):9–16.

Also presented at U.S.and Canadian Academy of Pathology Annual Meeting (USCAP), Mar 1-7, 2014, San Diego, CA, Abstr #1042.

... After extraction, DNA was subjected to CytoScan HD array (Affymetrix, Santa Clara, CA) assay using manufacturer's recommended protocol at Washington University's microarray core facility …

97- *Meeting abstract*

Genetic Analysis Using a High Density SNP Array in Myelodysplastic Syndrome: Clinical Utility and Comparative Analysis Study Compared to Metaphase Chromosome Analysis

Sally Jeffries*,1, Nicola Trim*,2, Emma Huxley*,2, Laura Ford*,2, Manoj Raghavan*,3, Jane Soden*,2, and Michael John Griffiths, BSc*,2

1Birmingham Women's NHS Foundation Trust, West Midlands Regional Genetics Laboratory, Birmingham, UK; 2West Midlands Regional Genetics Laboratory, Birmingham, UK; 3Centre for Clinical Haematology, University Hospitals Birmingham NHS Foundation Trust, Birmingham, UK

Blood, 3 December 2015, 126(23): 5259.

There is no single technology capable of detecting the various genetic and genomic aberrations observed in patients with neoplasia. Patients with myelodysplastic syndrome (MDS) may present with chromosomal copy number changes (duplication, deletion, and amplification), balanced chromosome rearrangements, copy neutral loss of heterozygosity (CN-LOH) and/or gene mutations. Currently only microscopic chromosomal changes, as dictated by the international prognostic scoring system (IPSS-R), are used to determine the genetic risk in MDS. ... The Affymetrix CytoScan® HD Array is a high definition array with over 2.6 million markers (both copy number and SNP) allowing resolution capabilities way beyond that of metaphase chromosome analysis. The incorporation of 750,000 SNPs also allows for detection of CN-LOH, regions known to harbour bi-allelic gene mutations. A real-time comparative study using the Affymetrix CytoScan® HD Array against traditional metaphase chromosome analysis is being performed on patients with confirmed or highly suspected MDS referred for genetic analysis at the West Midlands Regional Genetics Laboratory, UK. The study is expected to utilise 600 arrays over two years at presentation and on serial surveillance samples. The preliminary results available after the first 100 patients are presented with examples demonstrating the capabilities and clinical utility of SNP array genetic analysis. ... The study so far, in patients with MDS at presentation, has demonstrated: •An increased number of genetic aberrations (CN changes

and CN-LOH) detected by SNP array (38/105 (36%) by metaphase analysis and 62/105 (60%) by SNP array analysis). ... Patient benefits are expected to be the potential to improve patient outcomes through improved confidence in diagnosis, prognosis and monitoring. Disclosures Jeffries: Affymetrix: Research Funding.

98-

Genetic variation analysis in a Chinese Maffucci syndrome patient

Yang Xue, MDa, Jinwen Ni, MSa, Mi Zhou, MSa, Weiqi Wang, MSb, Yuan Liu, MDc, Yaowu Yang, MDb, , (Professor), Xiaohong Duan, PHDa, , (Professor)
a State Key Laboratory of Military Stomatology, Department of Oral Biology, Clinic of Oral Rare and Genetic Diseases, School of Stomatology, the Fourth Military Medical University, 145 West Changle Road, Xi'an 710032, P. R. China; b State Key Laboratory of Military Stomatology, Department of Oral and Maxillofacial Surgery, School of Stomatology, the Fourth Military Medical University, 145 West Changle Road, Xi'an 710032, P. R. China; c State Key Laboratory of Military Stomatology, Department of Oral Histology and Pathology, School of Stomatology, the Fourth Military Medical University, 145 West Changle Road, Xi'an 710032, P. R. China

Journal of Cranio-Maxillofacial Surgery, September 2015, 43(7): 1248–1255.

… Genomic DNA was extracted from blood and a hemangioma sample from the patient, and also from her mother's blood, for chromosome microarray analysis (CMA) by Affymetrix CytoScan HD array. … This is the first molecular genetic analysis report on a Chinese patient with Maffucci syndrome …

99-

Genome-wide analysis of the role of copy number variation in schizophrenia risk in chinese

Zhiqiang Li, Ph.Da, b, c, Jianhua Chen, Ph.Da, d, Yifeng Xu, MDd, Qizhong Yi, MDe, Weidong Ji, MDc, f, Peng Wang, MDg, Jiawei Shen, Ph.Da, Zhijian Song, Ph.Da, Meng Wanga, Ping Yang, MDg, Qingzhong Wang, Ph.Da, Guoyin Fengd, Benxiu Liu, MDh, Wensheng Sun, MDh, Qi Xu, MDi, Baojie Li, Ph.Da, Lin He, Ph.Da, c, j, Guang He, Ph.Da, Wenjin Li, Ph.Da, Zujia Wen, Ph.Da, Ke Liu, Ph.Da, Fang Huanga, Juan Zhoua, Jue Jia, Xingwang Li, Ph.Da, Yongyong Shi, Ph.Da, b, c, e, f, k
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Biological Psychiatry, online 26 November 2015, In Press, Accepted Manuscript

... Our data confirm increased genome-wide CNVs (> 500 Kb and < 1%) burden in schizophrenia, and the increasing trend was more significant when only > 1 Mb CNVs were considered. ... CNV validation by Affymetrix CytoScan HD Arrays. ...

100- *Meeting abstract*

Genome-Wide Molecular Portrait of Aggressive Systemic Mastocytosis and Mast Cell Leukemia Depicted By Whole Exome Sequencing and Copy Number Variation Analysis

Simona Soverini, PhD1, Caterina De Benedittis, PhD*,1, Manuela Mancini, PhD*,1, Michela Rondoni, MD*,2, Cristina Papayannidis, MD PhD3, Antonella Padella*,4, Giorgina Specchia, Pr, MD5, Roberta Zanotti, MD*,6, Livio Pagano*,7, Viviana Guadagnuolo, PhD*,1, Maria Chiara Fontana*,1, Massimo Delledonne, PhD*,8, Alberto Ferrarini, PhD*,9, Italo Do Valle*,10, Daniel Remondini, PhD*,11, Gastone Castellani*,12, Raffaele Calogero*,13, Serena Merante, MD*,14, Chiara Elena, MD*,14, Peter Valent, MD15, Michele Cavo*,16, and Giovanni Martinelli, MD PhD3
1"Seràgnoli" Institute of Hematology, Bologna University School of Medicine, Bologna, Italy; 2Area Vasta Romagna, Centro ServiziPievesestina, AUSL Romagna, Pievesestina di Cesena (FC), Italy; 3Bologna University School of Medicine, Bologna, Italy; 4University of Bologna, Bologna, Italy; 5Department of Emergency and Organ Transplantation, Section of Hematology with Transplantation, Medical Sch, University of Bari, Bari, Italy; 6Section of Haematology, Verona, Italy; 7Institute of Hematology, Catholic University, Rome, Italy;8Personal Genomics, Verona, Italy; 9Dep of Biotechnology, University of Verona, Verona, Italy; 10CAPES Foundation, Ministry of Education of Brazil, Brasília, Brazil; 11Dept of Physics and Astronomy, University of Bologna, Bologna, Italy;12Department of Physics and Astronomy, University of Bologna, Bologna, Italy; 13University of Turin, Bioinformatics and Genomics Unit, Turin, Italy; 14Policlinico S. Matteo IRCCS, Pavia, Italy;15Ludwig Boltzmann Cluster Oncology, Medical University of Vienna, Vienna, Austria; 16Seràgnoli Institute of Hematology, Bologna University School of Medicine, Bologna, Italy.

Blood, 3 December 2015, 126 (23): 4085.

… A discovery panel including 6 patients with ASM and 6 patients with MCL was studied using whole exome sequencing (WES) and copy number variation (CNV) analysis. WES (80x) was performed on a Hiseq 2500 (Illumina). CNV was done using Cytoscan HD Arrays (Affymetrix). ...

101-
Genome-wide profiling of HPV integration in cervical cancer identifies clustered genomic hot spots and a potential microhomology-mediated integration mechanism
Zheng Hu, Da Zhu, Wang, Weiyang Li, Wenlong Jia, Xi Zeng, Wencheng Ding, Lan Yu, Xiaoli Wang, Liming Wang, Hui Shen, Changlin Zhang, Hongjie Liu, Xiao Liu, Yi Zhao, Xiaodong Fang, Shuaicheng Li, Wei Chen, Tang Tang, Aisi Fu, Zou Wang, Gang Chen, Qinglei Gao, Shuang Li, Ling Xi
Nature Genetics, 2015, 47: 158–163.
... Ligated DNA was amplified, fragmented, end-labeled with biotin and then hybridized to an Affymetrix CytoScan HD Array.

102-
Genomic Alterations in Ethanol-Exposed Trophoblast Cell Lines Induced by Chronic Ethanol Treatment
Sergi Clave1,2,7, Xavier Joya1,2, Anna Puiggros3, Marta Bódalo4, Judith Salat-Batlle1,2, Marta Salido3, Blanca Espinet3, Óscar Garcia-Algar1,2,5,6,* and Oriol Vall1,2,5,6
1Grup de Recerca Infància i Entorn (GRIE), Institut Hospital del Mar d'Investigacions Mèdiques (IMIM), Barcelona, Spain; 2Red de Salud Materno-Infantil y del Desarrollo (SAMID), Programa RETIC, Fondos FEDER, Instituto Carlos III, Madrid, Spain; 3Laboratori de Citogenètica Molecular. Servei de Patologia, Hospital del Mar, Barcelona, Spain; 4Servei d'Anàlisis de Microarrays, Institut Hospital del Mar d'Investigacions Mèdiques (IMIM), Barcelona, Spain; 5Departament Pediatria, Obstetricia i Ginecologia, i Medicina Preventiva, Universitat Autònoma Barcelona (UAB), Bellaterra, Spain; 6Servei de Pediatria, Parc de Salut Mar, Barcelona, Spain; 7Laboratori de Citogenètica Molecular. Servei de Patologia, Hospital del Mar, Barcelona, Spain
Journal of Steroids & Hormonal Science, 2015, 6: 3.
… The aim of this study is to determine ethanol-caused genomic alterations in placental cell lines after a repeated ethanol treatment in order to describe new genomic targets of cell damage. ... Genomewide high resolution Single Nucleotide Polymorphisms (SNPs) array CytoScan HD (Affymetrix, Santa Clara, CA, USA) was used ...

103-
Genomic analysis of clonal origin of Langerhans cell histiocytosis following acute lymphoblastic leukaemia
Motohiro Kato1,2,3,†, Masafumi Seki1,†, Kenichi Yoshida4, Yusuke Sato4, Ryo Oyama5, Yuki Arakawa5, Hiroshi Kishimoto6, Tomohiko Taki7, Masaharu Akiyama8, Yuichi Shiraishi9, Kenichi Chiba9, Hiroko Tanaka10, Noriko Mitsuiki11, Michiko Kajiwara11, Shuki Mizutani11, Masashi Sanada4,12, Satoru Miyano9,10, Seishi Ogawa4, Katsuyoshi Koh5 and Junko Takita1
1Dept of Paediatrics, University of Tokyo, Tokyo, Japan; 2Div of Transplantation and Cell Therapy, Children's Cancer Centre, Tokyo, Japan; 3Dept of Paediatric Haematology and Oncology Research, Natl Centre for Child Health and Development, Tokyo, Japan; 4Dept of Pathology and Tumor Biology, Kyoto Univ, Kyoto, Japan; 5Dept of Haematology/Oncology, Saitama Children's Medical Centre, Saitama, Japan; 6Dept of Pathology, Saitama Children's Medical Centre, Saitama, Japan; 7Dept of Molecular Diagnostics and Therapeutics, Kyoto Prefectural Univ of Medicine Graduate School of Medical Science, Kyoto, Japan; 8Dept of Paediatrics, Jikei University School of Medicine, Tokyo, Japan; 9Laboratory of DNA Information Analysis, Human Genome Centre, Institute of Medical Science, University of Tokyo, Tokyo, Japan; 10Laboratory of Sequence Data Analysis, Human Genome Centre, Institute of Medical Science, University of Tokyo, Tokyo, Japan; 11Dept of Paediatrics, Tokyo Medical and Dental University, Tokyo, Japan; 12Dept of Advanced Diagnosis, Clinical Research Centre, National Hospital Organization, Nagoya Medical Centre, Nagoya, Japan.
British Journal of Haematology, first published online: 5 November 2015
... DNA extracted from the obtained samples was analysed using SNP-chip (GeneChip Human Mapping 250K NspI/Cytoscan HD; Affymetrix, ... LCH is not a secondary malignancy ... but was instead derived from a founding clone with genomic alterations common to ALL.

104--
Genomic copy number variation affecting genes involved in the cell cycle pathway: implications for somatic mosaicism
Ivan Y. Iourov,1,2,3 Svetlana G. Vorsanova,1,2 Maria A. Zelenova,1,2 Sergei A. Korostelev,4 and Yuri B. Yurov1,2
1Mental Health Research Center, Moscow 117152, Russia; 2Separated Structural Unit "Clinical Research Institute of Pediatrics", Pirogov Russian National Research Medical University, Ministry of Health of Russian Federation, Moscow 125412, Russia; 3Dept of Medical Genetics, Russian Medical Academy of Postgraduate Education, Moscow 123995, Russia; 4I.M. Sechenov First Moscow Medical University, Moscow 119991, Russia
International Journal of Genomics, 2015, 2015:Article ID 757680.

... Here, we have evaluated genomic copy number variation (CNV) in genes implicated in the cell cycle pathway (according to Kyoto Encyclopedia of Genes and Genomes/KEGG) within a cohort of patients with intellectual disability, autism, epilepsy and/or congenital malformations, in which the phenotype was not associated with genomic rearrangements altering this pathway. ... Genomic CNVs were analyzed using CytoScan HD Arrays (Affymetrix, Santa Clara, CA) consisting of approximately 2.7 million markers for CNV evaluation and approximately 750,000 SNPs.

105-
Genomic Instability of Osteosarcoma Cell Lines in Culture: Impact on the Prediction of Metastasis Relevant Genes
Roman Muff, Prisni Rath, Ram Mohan Ram Kumar, Knut Husmann, Walter Born, Michael Baudis, Bruno Fuchs
PLoS ONE, May 19, 2015, 10(5): e0125611.
… Microarray CGH was performed with the CytoScan HD Array Kit (Affymetrix, Santa Clara, CA) according to the instructions of the manufacturer at the Laboratory for Oncology Diagnostics (Kinderspital Zurich, Switzerland).

106-
Genomic Landscape of Primary Mediastinal B-Cell Lymphoma Cell Lines
Haiping Dai, Stefan Ehrentraut, Stefan Nagel, Sonja Eberth, Claudia Pommerenke, Wilhelm G. Dirks, Robert Geffers, Srilaxmi Kalavalapalli, Maren Kaufmann, Corrina Meyer, Silke Faehnrich, Suning Chen, Hans G. Drexler, Roderick A. F. MacLeod
PLOS ONE, 23 November 2015, 10(11): e0139663.
... CytoScan High Density Arrays which combine oligonucleotide and SNP probes (Affymetrix, High Wycombe/UK) were used to detect genomic copy number alterations/gains/losses (CNA/G/L), losses of heterozygosity (LOH) and unbalanced chromosome translocation breakpoints at high resolution. ... Samples were hybridized to Affymetrix GeneChip HG-U133 2.0 Plus for 16 h at 45°C. ... For microRNA profiling the Affymetric GeneChip miRNA 2.0 system was used together with samples drawn from the DSMZ cell bank as comparison data sets.

107- *Meeting abstract*
Genomic Landscape Predictive of Minimal Residual Disease (MRD) in Multiple Myeloma (MM)
Mehmet K Samur, PhD*,1,2, Stephane Minvielle*,3, Florence Magrangeas*,3, Giovanni Parmigiani, PhD*,1, Kenneth C Anderson4, Philippe Moreau, MD*,5, Michel Attal6, Hervé Avet-Loiseau, MD*,7, and Nikhil C. Munshi, MD8,2
1Biostatistics and Computational Biology, Dana-Farber Cancer Institute, Boston, MA ; 2LeBow Institute for Myeloma Therapeutics and Jerome Lipper Multiple Myeloma Center, Dana-Farber Cancer Institute, Harvard Medical School, Boston, MA ; 3Centre Hospitalier Universitaire de Nantes, Unité Mixte de Genomique du Cancer, Nantes, France ; 4The Jerome Lipper Multiple Myeloma Center, Dana-Farber Cancer Institute, Harvard Medical School, Boston, MA ; 5Nantes University Hospital, Hôtel Dieu, Nantes, France ; 6Institut Universitaire du Cancer de Toulouse-Oncopole, Toulouse, France ; 7Centre de Recherche en Cancérologie de Toulouse Institut National de la Santé, Toulouse, France ; 8VA Medical Hospital, Boston, MA
Blood, 3 December, 2015, 126(23): 4212.
… Here we are evaluating the genomic alterations that may predict attainment of MRD negative status in MM. MRD status was evaluted in 279 patients from IFM/DFCI 2009 trial. We obtained gene expression by RNA-seq, and copy number profile by cytoScan HD array to evaluate genomic differences between MRD negative and MRD positive groups. We generated copy number data for 175 / 279 patients (72 MRD- and 103 MRD+) with Affymetrix Cytoscan HD array and compared genome wide copy number alterations. ... In conclusion, we here report a first genomic landscape predictive of minimal residual disease (MRD) in Multiple Myeloma (MM). ...

108-
Genomic microarray in fetuses with increased nuchal translucency and normal karyotype - a systematic review and meta-analysis
Maribel Grande1, Fenna A.R. Jansen2, Yair J. Blumenfeld3, Allan Fisher4, Anthony O. Odibo5, Monique C. Haak2 and Antoni Borrell1,*
1Dept of Maternal-Fetal Medicine, Institute Gynecology, Obstetrics and Neonatology, Hospital Clinic of Barcelona, Catalonia, Spain; 2Leiden University Medical Center, Department of Obstetrics and Fetal Medicine, Leiden, The Netherlands; 3Dept of Obstetrics & Gynecology, Stanford University School of Medicine, Stanford, California, USA; 4Elliot Health System, Manchester, New Hampshire, USA; 5Dept of Obstetrics & Gynecology, Division of Maternal Fetal Medicine, University of South Florida, Tampa, Florida, USA
Ultrasound in Obstetrics & Gynecology, Accepted manuscript online: 20 April 2015
...systematic review of the literature ...and estimate the incremental yield of genomic microarray over karyotyping in fetuses with increased nuchal translucency diagnosed by first trimester prenatal ultrasound. ...Whole Genome 2.7M SNP array or Cytoscan HD SNP array ...

109-
Genomic profile of a Li-Fraumeni-like syndrome patient with a 45,X/46,XX karyotype, presenting neither mutations on TP53 nor clinical stigmata of Turner syndrome
Tatiane R. Basso, Rolando A.R. Villacis, Luisa M. Canto, Vinicius M.F. Alves, Rainer M.L. Lapa, Amanda F. Nobrega, Maria I. Achatz, Silvia R. Rogatto
Cancer Genetics, June 2015, 208(6): 341–344.
... Genome-wide analysis of copy number variations (CNVs) was assessed in DNA from peripheral blood cells and saliva (Affymetrix CytoScan HD).

110- *One sentence comment only*
Genomic Variants Revealed by Invariably Missing Genotypes in Nelore Cattle
Joaquim Manoel da Silva, Poliana Fernanda Giachetto, Luiz Otávio Campos da Silva, Leandro Carrijo Cintra, Samuel Rezende Paiva, Alexandre Rodrigues Caetano, Michel Eduardo Beleza Yamagishi
PLOS ONE, 25 August 2015, 10(8): e0136035.
... Our study has shown that often-discarded missing genotypes can be effectively used to identify population-specific genomic variants which in turn can be used in a wide range of applications. ... Companies that manufacture genotyping chips could develop denser HD genotyping chips and minimize this weakness by designing probes to cover every non-repetitive loci in the genome under study. This prospect is a trend at least in humans as the CytoScanHD Human array from Affymetrix has 2.67 million probes, 1.9 of which are non-polymorphic and designed to empower the results of CNV studies, but which are also compatible with our approach.

111- *Meeting abstract*
Genomic-Wide Analysis By High Resolution SNP Array Identifies Novel Genomic Alteration in Acute Myeloid Leukemia
Maria Chiara Fontana*,1, Viviana Guadagnuolo, PhD*,2, Cristina Papayannidis, MD PhD3, Giorgia Simonetti, PhD*,2, Antonella Padella*,2, Anna Ferrari, PhD*,2, Marco Manfrini, PhD*,3, Italo Faria do Valle*,4,5, Barbara Santacroce*,6, Margherita Perricone*,7, Giovanni Marconi, MD*,2, Emanuela Ottaviani*,2, Jelena D. Milosevic Feenstra, PhD*,8, Doris Chen, PhD*,8, Nicole C.C. Them*,8, Simona Soverini9, Daniel Remondini, PhD*,5, Gerardo Musuraca*,10, Michele Cavo, MD*,11, Robert Kralovics, PhD8, and Giovanni Martinelli11
1Seragnoli Institute, University of Bologna, Bologna, Italy; 2University of Bologna, Bologna, Italy; 3Bologna University School of Medicine,; 4Ministry of Education of Brazil, CAPES Foundation, Brazilia, Brazil; 5Dept of Physics and Astronomy, University of Bologna, Bologna, Italy; 6DIMES - Department of Experimental, Diagnostic and Specialty Medicine, Institute of Hematology, University of Bologna, Bologna, Italy; 7Institute of Hematology "Seragnoli", University of Bologna, Bologna, Italy; 8CeMM Research Center for Molecular Medicine of the Austrian Academy of Sciences, Vienna, Austria; 9Institute of Hematology, S.Orsola-Malpighi University Hospital, Bologna, Italy; 10Istituto Scientifico Romagnolo Per Lo Studio E La Cura Dei Tumori, Meldola (FC), Meldola, Italy; 11"Seràgnoli" Institute of Hematology, University of Bologna, Bologna, Italy
Blood, 3 December 2015, 126(23): 2600.
Introduction: Novel array-based technique-single-nucleotide polymorphism (SNP) microarray can detect cytogenetic lesions mostly involving structural alterations with losses or gains of chromosomic material. .. SNP microarray can also detect copy-neutral loss of heterozygosity (CN-LOH), … We analyzed 279 AML patients (pts) at diagnosis by SNP Array 6.0 or Cytoscan HD Array (Affymetrix). … By SNP array we have identified CNAs involving novel potential leukemia-related genes. Our results suggest that the comparison between SNP and WES data could provide important findings on the prognosis of AML pts. …

112-
Giant breast tumors in a patient with Beckwith–Wiedemann syndrome
Gerarda Cappuccio1,*, Agostina De Crescenzo2, Giuseppe Ciancia3, Luigi Canta4, Marzia Moio4, Ilaria Mataro4, Valeria Varone3, Guido Pettinato3, Orazio Palumbo5, Massimo Carella5, Andrea Riccio2,6, Nicola Brunetti-Pierri1,7
1Dept of Translational Medicine, Section of Pediatrics, Federico II University, Naples, Italy; 2Dept of Environmental, Biological and Pharmaceutical Sciences and Technologies at the Second University of Naples, Naples, Italy; 3Dept of Pathology, Federico II University, Naples, Italy; 4Dept of Plastic Surgery, Federico II University, Naples, Italy; 5Medical Genetics Unit, IRCCS Casa Sollievo Della Sofferenza Hospital, San Giovanni Rotondo, Italy; 6Institute of Genetics and Biophysics A. Buzzati–Traverso at the Consiglio Nazionale delle Ricerche (CNR), Naples, Italy; 7Telethon Institute of Genetics and Medicine, Naples, Italy
American Journal of Medical Genetics Part A, January 2014, 164(1):182–185.

... Further investigations with a SNP array performed by CytoScan HD array (Affymetrix, Santa Clara, CA) ruled out the presence of smaller (<100 kb) deletion/duplications on chromosome 11p15.5 which are found very rarely in BWS [Begemann et al., 2012].

113-
Global patterns of large copy number variations in the human genome reveal complexity in chromosome organization
AVINASH M. VEERAPPAa1, RAVIRAJ V. SURESHa1 †, SANGEETHA VISHWESWARAIAHa1 †, KUSUMA LINGAIAHa1 †, MEGHA MURTHYa1 †, DINESH S. MANJEGOWDAa2, PRAKASH PADAKANNAYAa3 and NALLUR B. RAMACHANDRAa1 c1
a1 Genetics and Genomics Lab, Dept of Studies in Genetics & Genomics, University of Mysore, Manasagangotri, Mysore-06, Karnataka, India; a2 Nitte University Centre for Science Education & Research, K. S. Hegde Medical Academy, Nitte, University, Deralakatte, Mangalore-18, Karnataka, India; a3 Dept of Studies in Psychology, University of Mysore, Manasagangotri, Mysore-06, Karnataka, India
Genetics Research (Cambridge), online 22 September 2015, 97: e18.
... analysis was performed using the Affymetrix Genome-Wide Human SNP Array 6.0 chip and CytoScan High-Density arrays. ... established the first drafts of population-specific CNV maps providing a rationale for prioritizing chromosomal regions.

114-
Global Spectrum of Copy Number Variations Reveals Genome Organizational Plasticity and Proposes New Migration Routes
Avinash M. Veerappa, Sangeetha Vishweswaraiah, Kusuma Lingaiah, Megha Murthy, Raviraj V. Suresh, Dinesh S. Manjegowda, Nallur B. Ramachandra
PLoS ONE, 24 April 2015, 10(4): e0121846.
… Genome-wide genotyping was performed using an Affymetrix Genome-wide Human SNP Array 6.0 chip and Affymetrix CytoScan High-Density (HD) Array having 1.8 million and 2.6 million combined SNP and CNV markers with the median inter- marker distance of 500–600 bases ...These chips provide maximum panel power and the highest physical coverage of the genome [47–50].

115-
Heterozygous deletion at the SOX10 gene locus in two patients from a Chinese family with Waardenburg syndrome type II
He Wenzhi a, 1, Wen Ruijin b, 1, Li Jieliang a, 2, Ma Xiaoyan a, Liu Haibo a, Wang Xiaoman a, Xian Jiajia a,
Li Shaoying a, Li Shuanglin a, Li Qing a,
a Key Laboratory for Major Obstetric Diseases of Guangdong Province, Key Laboratory of Reproduction and Genetics of Guangdong Higher Education Institutes, Experimental Department of Institute of Gynecology and Obstetrics, The Third Affiliated Hospital of Guangzhou Medical University, Guangzhou 510150, Guangdong, China; b Dept of Otolaryngology, Guangzhou Women and Children's Medical Center, Guangzhou 510623, Guangdong, China
International Journal of Pediatric Otorhinolaryngology, October 2015, 79(10): 1718–1721
... This report is the first to describe SOX10 heterozygous deletions in Chinese WS2 patients. ... Array-CGH was performed using the Affymetrix CytoScan HD platform (Affymetrix, Santa Clara, USA).

116-
High resolution single nucleotide polymorphism array reveals cryptic indicating information about myelodysplastic syndrome-related myeloid malignancies
Kun Chiab, Yang Lib, Qiulan Dingc, Lan Xud, Yu Chend & Xuefeng Wangc*
a Dept of Laboratory Medicine, Qingdao Women & Children's Hospital, Qingdao, China; b State Key Laboratory of Medical Genomics, Shanghai Institute of Hematology, Shanghai, China; c Dept of Laboratory Medicine, Shanghai, China; d Dept of Hematology, Ruijin Hospital, Shanghai Jiaotong University School of Medicine, Shanghai, China
Leukemia & Lymphoma, 2015, 56(10): 2969-2972.
... Genomic hybridization of the extracted DNA was performed with a CytoScan™ HD array (Affymetrix ... ... UPDs were detected in 20 patients (60.6%) by CytoScan™ HD array.

117-
Histo-genomic stratification reveals the frequent amplification/overexpression of CCNE1 and BRD4 genes in non-BRCAness high grade ovarian carcinoma
Oumou Goundiam1,2,3,†,*, Pierre Gestraud4,5,6,†, Tatiana Popova7,†, Thibault De la Motte Rouge8, Virginie Fourchotte9, David Gentien3, Philippe Hupé4,5,6,10, Véronique Becette1, Claude Houdayer1,7,11, Sergio Roman-Roman3, Marc-Henri Stern1,7, Xavier Sastre-Garau1,2
1Dept of Biopathology, Institut Curie, Paris, France; 2EA4340-BCOH, Versailles Saint-Quentin-en-Yvelines University, Guyancourt, France; 3Department of Translational Research, Institut Curie, Paris, France; 4Bioinformatics and Computational Systems Biology of Cancer, Institut Curie, Paris, France; 5Mines Paris Tech,

Paris, France; 6Inserm U900, Paris, France; 7Inserm U830 Institut Curie, Paris, France; 8Dept of Medical Oncology, Institut Curie, Paris, France; 9Dept of Surgery, and on behalf of the Gynecologic Study Group, Institut Curie, Paris, France; 10CNRS UMR 144; 11Université Paris Descartes, Sciences Pharmaceutiques et Biologiques, Sorbonne Paris Cité, Paris, France
International Journal of Cancer, 15 October 2015, 137(8): 1890–1900.
… Copy number and expression arrays DNA samples (105 tumors) were hybridized on Cytoscan® HD arrays (Affymetrix

118-
Homozygosity Mapping and Targeted Sanger Sequencing Reveal Genetic Defects Underlying Inherited Retinal Disease in Families from Pakistan
Maleeha Maria , Muhammad Ajmal , Maleeha Azam , Nadia Khalida Waheed, Sorath Noorani Siddiqui,
Bilal Mustafa, Humaira Ayub, Liaqat Ali, Shakeel Ahmad, Shazia Micheal, Alamdar Hussain, Syed Tahir Abbas Shah, Syeda Hafiza Benish Ali, Waqas Ahmed, Yar Muhammad Khan, Anneke I. den Hollander, Lonneke Haer-Wigman, Rob W. J. Collin, Muhammad Imran Khan , Raheel Qamar , Frans P. M. Cremers
PLoS ONE 10(3): e0119806.
… Genetic linkage analysis was carried out for 53 of 81 families using microsatellite markers or whole genome SNP array platforms such as Illumina_10K, Affymetrix_6K, Human Omni express_700k and Cytoscan HD (Fig. 1, Table 1).

119-
Homozygosity mapping of autosomal recessive intellectual disability loci in 11 consanguineous Pakistani families
Iltaf Ahmed 1,2, Muhammad Arshad Rafiq 1, John B. Vincent 1,3,4, Attya Bhatti 2, Muhammad Ayub 5, Peter John 2
1 Molecular Neuropsychiatry & Development (MiND) Lab, Centre for Addiction and Mental Health, Campbell Family Mental Health Research Institute, Toronto, Ontario, Canada ; 2 Atta-ur-Rehman School of Applied Biosciences (ASAB), Natl University of Sciences and Technology (NUST), Islamabad, Pakistan ; 3 Dept of Psychiatry, University of Toronto, Toronto, Ontario, Canada ; 4 Institute of Medical Science, University of Toronto, Toronto, Ontario, Canada; 5 Dept of Psychiatry, Div of Developmental Disabilities, Queen's University, Kingston, Ontario, Canada
Acta Neuropsychiatrica, February 2015, 27(1):38-47.
… Microarray genotyping (Affymatrix 250K) was performed to identify homozygosity-by-descent (HBD) in all affected families. ... In total, 15 healthy and 44 affected individuals were genotyped, using either Affymetrix 250K NspI (Affymetrix, Santa Clara, CA, USA) or Affymetrix® CytoScan™ HD Array (Affymetrix), and for one family using Illumina CoreExome beadchip arrays ...

120-
Identification of disrupted AUTS2 and EPHA6 genes by array painting in a patient carrying a de novo balanced translocation t(3;7) with intellectual disability and neurodevelopment disorder
Anouck Schneider1,*, Jacques Puechberty2, Bee Ling Ng3, Christine Coubes2, Vincent Gatinois1, Magali Tournaire1, Manon Girard1, Bruno Dumont1, Pauline Bouret1, Julia Magnetto4, Amaria Baghdadli4, Franck Pellestor1 and David Geneviève1,2
1Laboratoire de Génétique Chromosomique, Plateforme de puces à ADN, CHRU de Montpellier, France; 2Département de Génétique Médicale, CHRU de Montpellier, France; 3Cytometry Core Facility, The Wellcome Trust Sanger Institute, Wellcome Trust Genome Campus, Hinxton, Cambridge, UK; 4CRA, Département de Psychiatrie de l'Enfant et de l'Adolescent, Centre de Ressources Autisme, CHRU de Montpellier, France
American Journal of Medical Genetics Part A, December 2015, 167(12): 3031–3037.
... we used the high resolution GeneChip® Human Mapping Cytoscan HD Array (Affymetrix).

121-
Identifying the similarities and differences between single nucleotide polymorphism array (SNPa) analysis and karyotyping in acute myeloid leukemia and myelodysplastic syndromes
Thiago Rodrigo de Noronha, Sandra Serson Rohr, Maria de Lourdes Lopes Ferrari Chauffaille
Revista Brasileira de Hematologia e Hemoterapia, Jan-Feb 2015, 37(1):48-54.
…Twenty-two patients diagnosed with acute myeloid leukemia and three with myelodysplastic syndromes were studied. The G-banding karyotyping and single nucleotide polymorphism array analysis (CytoScan® HD) were performed using cells from bone marrow, DNA extracted from mononuclear cells from bone marrow and buccal cells (BC).

122-
Impaired PRC2 activity promotes transcriptional instability and favors breast tumorigenesis

Michel Wassef1,2,3, Veronica Rodilla1,2,3, Aurélie Teissandier1,4,5, Bruno Zeitouni1,4,5, Nadege Gruel1, Benjamin Sadacca1, Marie Irondelle1, Margaux Charruel1,2,3, Bertrand Ducos6,7,8, Audrey Michaud1,2,3, Matthieu Caron1,2,3, Elisabetta Marangoni1, Philippe Chavrier1, Christophe Le Tourneau9,10, Maud Kamal9, Eric Pasmant11,12,13, Michel Vidaud11,12,13, Nicolas Servant1,4,5, Fabien Reyal1, Dider Meseure1,14, Anne Vincent-Salomon1, Silvia Fre1,2,3 and Raphaël Margueron1,2,3

1Institut Curie, Paris Sciences et Lettres Research University, 75005 Paris, France; 2U934, Institut National de la Santé et de la Recherche Médicale, 75005 Paris, France; 3UMR3215, Centre National de la Recherche Scientifique, 75005 Paris, France; 4U900, Institut National de la Santé et de la Recherche Médicale, 75005 Paris, France; 5Mines ParisTech, 77300 Fontainebleau, France; 6Laboratoire de Physique Statistique-Ecole Normale Supérieure de Paris, Centre National de la Recherche Scientifique, 75005 Paris, France; 7UMR 8550, Centre National de la Recherche Scientifique, 75005 Paris, France; 8Plateforme de PCR Quantitative à Haut Débit Genomic Paris Centre, Institut de Biologie de l'École Normale Supérieure, 75005 Paris, France; 9Dept of Medical Oncology, Institut Curie, 75005 Paris, France; 10EA7285, Université de Versailles, Saint-Quentin-en-Yvelines, 78000 Versailles, France; 11UMR_S745, EA7331, Institut National de la Santé et de la Recherche Médicale, 75006 Paris, France; 12Facultée des Sciences Pharmaceutiques et Biologiques, Université Paris Descartes, Sorbonne Paris Cité, 75006 Paris, France; 13Service de Biochimie et Génétique Moléculaire, Assistance Publique-Hôpitaux de Paris, Hôpital Cochin, 75014 Paris, France; 14Platform of Investigative Pathology, 75005 Paris, France

Genes & Development, 2015, 29: 2547-2562.

... provides an unexpected understanding of EZH2's contribution to solid tumors ... in breast cancer metastases previously analyzed by Affymetrix CytoScan arrays ... Transcriptome data analysis ... using Affymetrix Mouse Gene 1.1 ST arrays ...

123-

Influence of total genomic alteration and chromosomal fragmentation on response to a combination of azacitidine and lenalidomide in a cohort of patients with very high risk MDS

Christina Ganster a, Katayoon Shirneshan a, Gabriela Salinas-Riester b, Friederike Braulke a, Julie Schanz a, Uwe Platzbecker c, 1, Detlef Haase a, 1

a Dept of Hematology and Medical Oncology, University Hospital, University Göttingen, Göttingen, Germany; b DNA Microarray Facility, Georg August Univ, Göttingen, Germany; c Medical Clinic and Polyclinic I, Univ Hospital, Technical University Dresden, Dresden, Germany

Leukemia Research, October 2015, 39(10): 1079–1087.

... The CytoScan HD array (17 samples) and the Genome-Wide Human SNP 6.0 array (one sample) were used for MK (both Affymetrix, Santa Clara, CA, USA).

124-

Inherited 15q24 microdeletion syndrome in twins and their father with phenotypic variability

Lena Samuelsson, Theofanis Zagoras, Maria Hafström

European Journal of Medical Genetics, Feburuary 2015, 58(2):111–115.

regarding cases of intellectual deficiency in children.

... Samples were analysed by high-density SNP array using the Genome-Wide Human SNP 6.0 Array, and CytoScan HD from Affymetrix. ... high-resolution SNP array (CytoScan HD from Affymetrix) in the twin brother and father showed that deletion breakpoints are similar in the three cases, with no differences in genetic content ...

125-

Integrated genetic and epigenetic analysis defines novel molecular subgroups in rhabdomyosarcoma

Masafumi Seki, Riki Nishimura, Kenichi Yoshida, Teppei Shimamura, Yuichi Shiraishi,Yusuke Sato, Motohiro Kato, Kenichi Chiba, Hiroko Tanaka, Noriko Hoshino, Genta Nagae, Yusuke Shiozawa, Yusuke Okuno, Hajime Hosoi, Yukichi Tanaka, Hajime Okita, Mitsuru Miyachi, Ryota Souzaki, Tomoaki Taguchi, Katsuyoshi Koh, Ryoji Hanada, Keisuke Kato,Yuko Nomura, Masaharu Akiyama, Akira Oka,Takashi Igarashi, Satoru Miyano, Hiroyuki Aburatani, Yasuhide Hayashi, Seishi Ogawa, Junko Takita

Nature Communications, 3 July 2015, 6:Article #7557

... DNA extracted from RMS samples was subjected to SNP array analysis using Affymetrix GeneChip 250K Nsp or CytoScan HD (Affymetrix) according to the manufacturer's protocol.

126-

Integrated genomics elucidates relative spatial homogeneity of embryonal brain tumors

Marc Remke12*, Florence Cavalli1, A Sorana Morrissy1, Vijay Ramaswamy1, David Jones3, Roger Packer4, Eric Bouffet1, Gary Bader5, Arndt Borkhardt2, Stefan Pfister3, Nada Jabado6, Marco Marra7, Michael D Taylor1

1 Hospital for Sick Children, Toronto, Ontario, Canada; 2 University Hospital Düsseldorf (UKD) and German Cancer Consortium (DKTK), Düsseldorf, Germany; 3 German Cancer Research Center (DKFZ), Heidelberg, Germany; 4 Children's National Medical Center, Washington, D.C., USA; 5 University of Toronto, Toronto, Ontario,

Canada; 6 McGill University and the McGill University Health Center Research Institute, Montreal, Quebec, Canada; 7 Michael Smith Genome Sciences Centre, BC Cancer Agency, Vancouver, British Columbia, Canada
Molecular and Cellular Pediatrics, 1 July 2015, 2(Suppl 1):A10 [This article is part of the supplement: Abstracts of the 51st Workshop for Pediatric Research Meeting abstract]
... we conducted multiregion whole exome sequencing, high-resolution DNA copy number analysis (Cytoscan HD), and transcriptional profiling on 39 distinct pediatric and adult tumors with a median of six spatially distant biopsies per tumor (range 4-11). ...

127- *Meeting abstract*
Integrated molecular characterization of mast cell leukemia reveals recurrent inactivation of the SETD2 tumor suppressor gene
Simona Soverini1, Caterina De Benedittis1, Michela Rondoni2, Manuela Mancini1, Cristina Papayannidis1, Luca Zazzeroni1, Viviana Guadagnuolo1, Elisa Zago3, Francesca Griggio3, Alberto Ferrarini3, Marianna Garonzi3, Massimo Delledonne3, Giorgina Specchia4, Roberta Zanotti5, Omar Perbellini5, Livio Pagano6, Michele Cavo1, Peter Valent7, and Giovanni Martinelli1
1University of Bologna, Bologna, Italy; 2Area Vasta Romagna, Centro Servizi Pievesestina,, Cesena, Italy; 3Univ of Verona, Verona, Italy; 4Univ of Bari, Bari, Italy; 5Univ of Verona, UO Hematology, Verona, Italy; 6Hematology, Universita' Cattolica S. Cuore, Rome, Italy; 7University of Wien, Wien, Austria.
Cancer Research, 2015;75(15 Suppl):Abst #3957. [AACR 106th Annual Mtg, Apr 18-22, 2015, Philadelphia, PA]
Systemic mastocytosis (SM) includes a heterogeneous group of disorders ranging from indolent SM to the rare and aggressive mast cell leukemia (MCL). Somatic mutations in the KIT receptor kinase (most frequently, D816V) can be detected in >90% of patients and are thought to play an important pathogenetic role. ... we undertook an integrated molecular genetic study of a KIT gene mutation-negative MCL case who came to our attention in 2012. ... High resolution karyotyping was also performed with Cytoscan HD arrays (Affymetrix, Santa Clara CA). ... In line with the role of SETD2-dependent H3K36Me3 in DNA repair and genome stability, Cytoscan HD arrays and WES showed that several losses at many chromosomal loci, together with more than 70 additional point mutations, undetectable at diagnosis, were acquired at the time of progression. ...

128-
Integration of cytogenomic data for furthering the characterization of pediatric B-ALL: a multi-institution, multi-platform microarray study
L.B. Baughn, J.A. Biegel, S.T. South, T. Smolarek, S. Volkert, A. Carroll, N.A. Heerema, K.R. Rabin, P.A. Zweidler-McKay, M. Loh, B. Hirsch
Cancer Genetics, Jan-Feb 2015, 208(1–2):1–18.
… The present investigation demonstrates the feasibility and usefulness of integrating array results from multiple laboratories (ARUP, Children's Hospital of Philadelphia, Cincinnati Children's Hospital Medical Center, and University of Minnesota Medical Center) that utilize different array platforms (Affymetrix, Agilent, or Illumina) in their respective clinical settings. ... At ARUP, DNA was extracted using a Gentra Puregene kit from Qiagen, and patient DNA was analyzed using the CytoScan HD® microarray assay (Affymetrix, Santa Clara, CA) which contains 743,304 SNP-based oligonucleotides ...

129-
Intellectual disability secondary to a 16p13 duplication in a 1;16 translocation. Extended phenotype in a four-generation family
Amal Mahmoud Mohamed1,*, Alaa Kamel1, Wael Mahmoud1, Ehab Abdelraouf2 and Nagwa Meguid2
1Human Cytogenetics Department, National Research Centre, Cairo, Egypt; 2Research on Children with Special Needs Department, National Research Centre, Cairo, Egypt
American Journal of Medical Genetics Part A, January 2015, 167(1):128–136.
… Array CGH analysis by Cytoscan HD (Affymetrix) was used according to the manufacturer manual. Microarrays were scanned with GeneChip® Scanner 3000 7G.

130-
Interstitial 1p32.1p32.3 deletion in a patient with multiple congenital anomalies
Martin Kehrer1, Karin Schäferhoff1, Michael Bonin1,2, Anna Jauch3, Andrea Bevot4, Andreas Tzschach1,5,*
1Institute of Medical Genetics and Applied Genomics, University of Tuebingen, Tuebingen, Germany; 2IMGM Laboratories GmbH, Martinsried, Germany; 3Institute of Human Genetics, University of Heidelberg, Heidelberg, Germany; 4University Children's Hospital, University of Tuebingen, Tuebingen, Germany; 5Institute of Clinical Genetics, Technische Universität Dresden, Dresden, Germany
American Journal of Medical Genetics Part A, October 2015, 167(10): 2406–2410.
... Figure 2. Graphical representation of chromosome 1 and the hybridization results of the patient and his parents (Affymetrix CytoScan SNP Array). The deletion in 1p32.1p32.3 is highlighted by an arrow.

131-
Interstitial duplication of chromosome region 1q25.1q25.3: Report of a patient with mild cognitive deficits, tall stature and facial dysmorphisms
Martin Kehrer1, Thomas Liehr2,*, Tanja Benkert1, Sylke Singer1, Ute Grasshoff1, Karin Schaeferhoff1, Michael Bonin1, Annette Weichselbaum3, Andreas Tzschach1
1Institute of Medical Genetics and Applied Genomics, University of Tuebingen, Tuebingen, Germany; 2Jena Univ Hospital, Friedrich Schiller University, Institute of Human Genetics, Jena, Germany; 3University Children's Hospital, University of Tuebingen, Tuebingen, Germany
American Journal of Medical Genetics Part A, March 2015, 167(3):653–656.
… High-resolution chromosome analysis (SNP array analysis) was performed using the Affymetrix Human Mapping CytoScan™ HD Array ...

132-
Intratumoral genome diversity parallels progression and predicts outcome in pediatric cancer
Linda Holmquist Mengelbier,Jenny Karlsson,David Lindgren,Anders Valind,Henrik Lilljebjörn,Caroline Jansson,Daniel Bexell,Noémie Braekeveldt,Adam Ameur,Tord Jonson,Hanna Göransson Kultima,Anders Isaksson,Jurate Asmundsson,Rogier Versteeg,Marianne Rissler,Thoas Fioretos,Bengt Sandstedt,Anna Börjesson,Torbjörn Backman,Niklas Pal,Ingrid Øra,Markus Mayrhofer& David Gisselsson
Nature Communications, online 27 January 2015, 6: Article #6125.
… was hybridized to HumanCNV370-Duo/Quad Genotyping BeadChips, 1 M Genotyping BeadChip HumanOmni1-Quad (Illumina Inc., San Diego, CA), or Affymetrix Cytoscan HD ...

133- *Book chapter*
Introduction: Rationale for Precision Medicine Clinical Trials
Christophe Le Tourneau MD, PhD christophe.letourneau@curie.fr 3,4
3. Dept of Medical Oncology, Institut Curie, Saint-Cloud, Paris, France; 4. EA7285, Versailles-Saint-Quentin-en-Yvelines University, Versailles, France
In Pan-Cancer Integrative Molecular Portrait Towards a New Paradigm in Precision Medicine, pp 1-8
Springer : 2015.
... Molecular alterations were evaluated on a tumor sample from a metastatic site using NGS for mutations screening, CytoScan HD for gene copy number variations, and IHC for estrogen, progesterone, and androgen receptor expression analyses.

134-
Is Chromosome 15q13.3 Duplication Involving CHRNA7 Associated With Oral Clefts?
Yingjun Xie, PhD1,2
1Dept of Prenatal Diagnosis, The First Affiliated Hospital of Sun Yat-sen University, Guangzhou, China; 2Key Laboratory for Major Obstetric Diseases of Guangdong Province, Key Laboratory of Reproduction and Genetics of Guangdong Higher Education Institutes, The Third Affiliated Hospital of Guangzhou Medical University, Guangzhou, China
Child Neurology Open, October-December 2015, 2(4): 2329048X15618918
... peripheral blood or umbilical cord blood was collected ... and hybridized to the CytoScan HD array platform (Affymetrix, ... our study is the first to establish the relationship between 15q13.3 microduplication involving CHRNA7 and oral clefts.

135-
Jacobsen Syndrome: Surgical Complications due to Unsuspected Diagnosis, the Importance of Molecular Studies in Patients with Craniosynostosis
Linares Chávez E.P.a · Toral López J.c · Valdés Miranda J.M.a · González Huerta L.M.a · Perez Cabrera A.a · del Refugio Rivera Vega M.a · Messina Baas O.M.b · Cuevas-Covarrubias S.A.a
Departamentos de aGenética Médica and bOftalmología, Hospital General de México, Facultad de Medicina, Universidad Nacional Autónoma de México (UNAM), Mexico City, and cDepartamento de Genética Médica, Centro Médico Ecatepec, ISSEMYM, Edomex México, México
Molecular Syndromology, 2015, 6: 229-235.
... Oligonucleotide-SNP array analysis using the GeneChip Human Cytoscan HD was carried out for the patient from genomic DNA. The SNP array showed a 14.2-Mb deletion in chromosome 11q23.3q25 ...

136-

Keratoconus in an adult with 22q11.2 deletion syndrome
Norman Saffra, Benjamin Reinherz
Department of Ophthalmology, St John's Episcopal Hospital, Far Rockaway, New York, USA
BMJ Case Reports, published 16 January 2015; doi:10.1136/bcr-2014-203737
22q11.2 Deletion syndrome is one of the most common microdeletional syndromes, with an incidence of 1:4000 live-births, and potentially affects every organ in the body. … Keratoconus has been reported once before in association with 22q11.2 deletion syndrome in a young adult. We report the second case of keratoconus in association with 22q11.2 deletion syndrome. ... High-resolution chromosome analysis and oligo-SNP (oligonucleotide, single nucleotide polymorphism, Affymetrix CytoScan HD) assay were performed in a Clinical Laboratory Improvement Amendments (CLIA)-approved laboratory.

137-
KIT Mutation and Loss of 14q May Be Sufficient for the Development of Clinically Symptomatic Very Low-Risk GIST
Olaf Karl Klinke, Tuba Mizani, Gouri Baldwin, Brigitte Bancel, Mojgan Devouassoux-Shisheboran, Jean-Yves Scoazec, Pierre-Paul Bringuier, Regina Feederle, Anna Jauch, Katrin Hinderhofer, Philippe Taniere , Henri-Jacques Delecluse
PLoS ONE, 23 June 2015, 10(6): e0130149.
... Molecular karyotyping using 50 ng of tumour DNA was performed with the Affymetrix CytoScan HD Array ...

138-
Langerhans Cell Histiocytosis: Diagnosis on Thyroid Aspirate and Review of the Literature
Anjali Saqi1, Adriana P. Kuker2, Susana A. Ebner2, John Ausiello2, Vaidehi Jobanputra1, Govind Bhagat1,
Tamar A. Giorgadze3
1. Dept of Pathology and Cell Biology, Columbia University College of Physicians and Surgeons, New York, NY, USA ; 2. Div of; Endocrinology, Columbia University College of Physicians and Surgeons, New York, NY ; 3. Dept of Pathology and Laboratory Medicine, Weill Cornell Medical College, Cornell University, 525 East 68th St, Suite F-766, New York, NY, 10065
Head and Neck Pathology, December 2015, 9(4): 496-502.
... On performing SNP array analysis, using the CytoScan HD array (Affymetrix), no distinct somatic copy number alteration (chromosome gain or loss) or loss of heterozygosity was observed, supporting the G-band karyotype results.

139-
Language impairment in a case of a complex chromosomal rearrangement with a breakpoint downstream of FOXP2 **[open access]**
Daniela Moralli, Ron Nudel, May T. M. Chan, Catherine M. Green, Emanuela V. Volpi, Antonio Benítez-Burraco, Dianne F. Newbury, Paloma García-Bellido
Molecular Cytogenetics, 10 June 2015, 8:36.
... Figure S3. CNVs were mapped in the probands and parents using Affymetrix Cytoscan and copy number changes were called within Affymetrix Chromosome Analysis Suite (ChAS).

140-
Limb-girdle weakness in a marfanoid man: distinguishing calpainopathy from Becker's muscular dystrophy
Gasnat Shaboodien1,2, David A Watkins1,2, Komala Pillay3, Peter Beighton4, Jeannine M Heckmann5, Bongani M Mayosi1,2
1Cardiovascular Genetics Laboratory, Hatter Institute of Cardiovascular Research in Africa, Cape Town, South Africa ; 2Dept of Medicine, Groote Schuur Hospital and University of Cape Town, Cape Town, South Africa ; 3Div of Anatomical Pathology, Dept of Clinical Laboratory Sciences, Natl Health Lab Service and Univ of Cape Town, Cape Town, South Africa ; 4Div of Human Genetics, Univ of Cape Town, Cape Town, South Africa ; 5Div of Neurology, Dept of Medicine, Groote Schuur Hospital and University of Cape Town, Cape Town, South Africa
Practical Neurology, Mar 2015, 15:152-154.
... We proceeded with a genome-wide high-resolution single nucleotide polymorphism (SNP) array, CytoScan HD (Affymetrix), with 99% sensitivity and specificity for copy number changes ...

141-
Long contiguous stretches of homozygosity spanning shortly the imprinted loci are associated with intellectual disability, autism and/or epilepsy
Ivan Y. Iourov123*, Svetlana G. Vorsanova12, Sergei A. Korostelev4, Maria A. Zelenova12 and Yuri B. Yurov12
1 Mental Health Research Center, Moscow, 117152, Russia ; 2 Separated Structural Unit “Clinical Research Institute of Pediatrics”, Russian National Research Medical University named after N.I. Pirogov, Ministry of Health of Russian Federation, Moscow, 125412, Russia ; 3 Dept of Medical Genetics, Russian Medical Academy of

Postgraduate Education, Moscow, 123995, Russia ; 4 Research Centre for Medical Genetics, Moscow, 115478, Russia
Molecular Cytogenetics, 15 October 2015, 8:77
... SNP chromosomal microarrays may lead to a 5 % improvement in etiological yield by uncovering LCSH at imprinted loci. ... CNV and LCSH were analyzed by CytoScan HD Arrays (Affymetrix ...

142- *Only 1 sentence mentions CytoScan*
Low Rates of Genetic Testing in Children With Developmental Delays, Intellectual Disability, and Autism Spectrum Disorders
John Peabody, MD, PhD1,2, Lisa DeMaria, MA1, Diana Tamandong-LaChica, MD1, Jhiedon Florentino, MS1, Maria Czarina Acelajado, MD1, Trever Burgon, PhD1
1QURE Healthcare, San Francisco, CA, USA ; 2University of California, San Francisco and Los Angeles, USA
Global Pediatric Health January-December 2015, 2 : 2333794X15623717
… The physicians used standard platform CMAs (45.3%) rather than enhanced CMA testing (10.4%) with more probes (eg, tests that include Cytoscan HD and FirstStepDx PLUS; P < .0001).

143- *Invited review*
Malignant hematopoietic cell lines: In vitro models for the study of primary mediastinal B-cell lymphomas
Hans G. Drexler, Stefan Ehrentraut, Stefan Nagel, Sonja Eberth, Roderick A.F. MacLeod
Leibniz-Institute DSMZ-German Collection of Microorganisms and Cell Cultures, Dept. Human and Animal Cell Lines, Braunschweig, Germany
Leukemia Research, January 2015, 39(1):18–29. doi:10.1016/j.leukres.2014.11.002
… CytoScan high density genomic/SNP arrays were obtained from Affymetrix (High Wycombe, UK) and analyzed using Chromosome Analysis Suite software (Affymetrix).

144--
Mayer-Rokitansky-Küster-Hauser syndrome discordance in monozygotic twins:matrix metalloproteinase 14, low-density lipoprotein receptor–related protein 10, extracellular matrix, and neoangiogenesis genes identified as candidate genes in a tissue-specific mosaicism
Katharina Rall, M.D.a, Simone Eisenbeis, Ph.D.a, Gianmaria Barresi, M.D.a, Daniel Rückner, M.D.a, Michael Walter, Ph.D.b, Sven Pothsb, Diethelm Wallwiener, M.D.a, Olaf Riess, M.D.b, Michael Bonin, Ph.D.b, Sara Brucker, M.D.a
Fertility and Sterility, February 2015, 103(2):494–502.e3.
… For validation of the initial analysis, we reanalyzed the twin pairs' DNA with the Affymetrix Cytoscan HD Array. ... Validation of the initial analysis was possible by reanalyzing the twin pairs' DNA with the use of the Affymetrix CytoScan HD Array. ...

145- *Meeting abstract*
MG-127 Diagnostic accuracy of chromosome microarray in children with epilepsy and neurological abnormalities of unknown aetiology
Sarah E Buerki1, Erin Slade1, Kamilla Schlade-Bartusiak2, Lindsay Brown2, Evica Rajcan-Separovic2, Patrice Eydoux2, Mary B Connolly1, Michelle K Demos1
1Pediatric Neurology, University of BC, BC Children's Hospital, British Columbia, BC, Canada ; 2Cytogenetics, Children's and Women's Health Center of BC, University of BC, Vancouver, BC, Canada
Journal of Medical Genetics, 2015, 52:A7.
… The purpose of this study is to describe the results of performed CMA on 706 children with unexplained epilepsy associated with developmental delay/intellectual disability, autism spectrum disorders and/or multiple congenital anomalies ('epilepsy plus'). Design/method Retrospective chart review on clinical and genetic aspects of CNVs identified in 706 patients with 'epilepsy plus', seen at the Vancouver BC Children's Hospital from 2009 to 2014. All patients had CMA performed using Affymetrix Genome-Wide Human SNP Array 6.0 or CytoScanHD®. ... Conclusions CMA revealed pathogenic CNVs in epilepsy "hotspots", and known microdeletion syndromes like Phelan-McDermid syndrome in 11.3% of children with 'epilepsy plus'. ...

146-
Microarray analysis unmasked two siblings with pure hereditary spastic paraplegia shared a run of homozygosity region on chromosome 3q28–q29
Wenqian Yu a, 1, Xiangdong You b, 1, Dong Wang a, Kai Dong a, Jing Su c, Chuanfen Li c, Jinxiu Liu a, Qianqian Zhang a, Feng You a, Xiangrong Wang a, Jing Huang a, Bin Qiao a, 2, Wenyuan Duan a, 2

a Institute of Cardiovascular Disease, General Hospital of Jinan Military Region, 8 Lashan Road, Jinan 250022, China; b Div of Quality Management, Shandong Center for Disease Control and Prevention, 16992 Jingshi Road, Jinan 250014, China; c Dept of Neurology, General Hospital of Jinan Military Region, 25 Shifan Road, Jinan 250031, China

Journal of the Neurological Sciences, 15 December 2015, 359(1-2): 351–355.

... we describe a Chinese non-consanguineous family with two affected siblings manifesting early-onset autosomal recessive HSP... CytoScan HD array analysis was performed ... first clinical description of a pure form spastic paraplegia in a non-consanguineous family ...

147-

Microduplication of chromosome Xq25 encompassing STAG2 gene in a boy with intellectual disability.

Yingjun X1, Wen T2, Yujian L3, Lingling X3, Huimin H3, Qun F4, Junhong C4.

1Dept of Prenatal Diagnosis, The First Affiliated Hospital of Sun Yat-sen University, Guangzhou 510080, China; Key Laboratory for Major Obstetric Diseases of Guangdong Province, Key Laboratory of Reproduction and Genetics of Guangdong Higher Education Institutes, The Third Affiliated Hospital of Guangzhou Medical University, 510080, China.; 2Dept of Pediatric Intensive Care Unit, The First Affiliated Hospital, Sun Yat-sen University, Guangzhou 510080, China. Electronic address: tangwenr@21cn.com.; 3Dept of Pediatric Intensive Care Unit, The First Affiliated Hospital, Sun Yat-sen University, Guangzhou 510080, China.; 4Dept of Prenatal Diagnosis, The First Affiliated Hospital of Sun Yat-sen University, Guangzhou 510080, China.

Eur J Med Genetics, February 2015, 58(2):116-21.

… We report a 4-year-old boy with a de novo 591 kb duplication at Xq25. The duplication was first detected by a CytoScan HD array platform (Affymetrix, USA) and was confirmed by real-time quantitative PCR (qPCR) of the STAG2 gene, and by fluorescence in situ hybridization (FISH). …

148-

MMP21 is mutated in human heterotaxy and is required for normal left-right asymmetry in vertebrates

Anne Guimier, George C Gabriel, Fanny Bajolle, Michael Tsang, Hui Liu, Aaron Noll, Molly Schwartz, Rajae El Malti, Laurie D Smith, Nikolai T Klena, Gina Jimenez, Neil A Miller, Myriam Oufadem, Anne Moreau de Bellaing, Hisato Yagi, Carol J Saunders, Candice N Baker, Sylvie Di Filippo, Kevin A Peterson, Isabelle Thiffault, Christine Bole-Feysot, Linda D Cooley, Emily G Farrow, Cécile Masson, Patric Schoen, Jean-François Deleuze, Patrick Nitschké, Stanislas Lyonnet, Loic de Pontual, Stephen A Murray, Damien Bonnet, Stephen F Kingsmore, Jeanne Amiel, Patrice Bouvagnet, Cecilia W Lo & Christopher T Gordon

Nature Genetics, online 5 October 2015, 47:1260–1263.

... For validation of the deletion identified by whole-genome sequencing in family 2, genomic DNA was isolated using the Gentra PureGene kit for whole blood. Test sample DNA was hybridized to Affymetrix CytoScan HD CN+SNP microarray chips.

149-

Modeling colorectal cancer using CRISPR-Cas9–mediated engineering of human intestinal organoids

Mami Matano, Shoichi Date, Mariko Shimokawa, Ai Takano, Masayuki Fujii, Yuki Ohta, Toshiaki Watanabe, Takanori Kanai & Toshiro Sato

Nature Medicine, 2015, 21, 256–262.

… Microarray hybridization was performed on the GeneChip Prime View Human Gene Expression Array (Affymetrix) ... organoids was extracted using the QIAamp DNA Blood Mini Kit (Qiagen) and analyzed by Cytoscan HD (Affymetrix).

150-

Molecularly targeted therapy based on tumour molecular profiling versus conventional therapy for advanced cancer (SHIVA): a multicentre, open-label, proof-of-concept, randomised, controlled phase 2 trial

Dr Christophe Le Tourneau, MDa, u, Prof Jean-Pierre Delord, MDb, Prof Anthony Gonçalves, MDc, Céline Gavoille, MDd, Coraline Dubot, MDe, Nicolas Isambert, MDf, Prof Mario Campone, MDg, Olivier Trédan, MDi, Marie-Ange Massiani, MDk, Cécile Mauborgne, MScl, Sebastien Armanet, MScm, Nicolas Servant, PhDt, Ivan Bièche, PhDn, Virginie Bernard, PhDp, David Gentien, PhDo, Pascal Jezequel, MDh, Valéry Attignon, PhDj, Sandrine Boyault, PhDj, Anne Vincent-Salomon, MDr, Vincent Servois, MDs, Marie-Paule Sablin, MDq, Maud Kamal, PhDq, Xavier Paoletti, PhDl, t, for the SHIVA investigators

a Dept of Medical Oncology, Institut Curie, Paris & Saint-Cloud, France; b Dept of Medical Oncology, Institut Claudius Régaud, Toulouse, France; c Dept of Medical Oncology, Institut Paoli-Calmettes, Marseille, France; d Dept of Medical Oncology, Centre Alexis Vautrin, Nancy, France; e Dept of Medical Oncology, Institut Curie, Saint-Cloud, France; f Dept of Medical Oncology, Centre Georges-François Leclerc, Dijon, France; g Dept of Medical

Oncology, Centre René Gauducheau, Nantes, France; h Genomic Unit, Centre René Gauducheau, Nantes, France; i Dept of Medical Oncology, Centre Léon Bérard, Lyon, France; j Genomic Unit, Centre Léon Bérard, Lyon, France; k Dept of Pneumology, Hopital Foch, Suresnes, France; l Dept of Biostatistics, Institut Curie, Paris, France; m Clinical Trial Management Unit, Institut Curie, Paris, France; n Pharmacogenomics Unit, Institut Curie, Paris, France; o Translational Research Department, Institut Curie, Paris, France; p NGS Platform, Institut Curie, Paris, France; q Dept of Medical Oncoology, Institut Curie, Paris, France; r Dept of Biopathology, Institut Curie, Paris, France; s Dept of Radiology, Institut Curie, Paris, France; t Unité INSERM U900, Paris, France; u EA7285: Risques cliniques et sécurité en santé des femmes et en santé périnatale, Versailles Saint-Quentin-en Yvelines University, Montigny-le-Bretonneux, France

The Lancet Oncology, October 2015, 16(13):1324–1334

... Molecular analyses included assessment of mutations by targeted next generation sequencing (AmpliSeq cancer panel on an Ion Torrent/PGM system, ... gene copy number alterations by Cytoscan HD (Affymetrix, ...

151-

Mosaic paternal genome-wide uniparental isodisomy with down syndrome

Diana Darcy1, Paldeep Singh Atwal2, Cathy Angell3, Inder Gadi4 and Robert Wallerstein1,*

1Silicon Valley Genetics Center, Santa Clara Valley Medical Center, San Jose, CA; 2Stanford University Medical Center, Palo Alto, CA; 3Neonatology, O'Connor Hospital, San Jose, CA; 4Laboratory Corporation of America, Research Triangle Park, NC

American Journal of Medical Genetics Part A, October 2015, 167(10): 2463–2469.

... microarray results suggest that the cell line with trisomy 21 is biparentally inherited ... this patient is the only reported case that also involves trisomy 21. ... ... The sample was run using an Affymetrix Cytoscan HD microarray ...

152-

Mosaic tetrasomy 20p associated with osteoporosis and recurrent fractures

Asmaa S. Abu Maziad1 and Laurie H. Seaver1,2,*

1Dept of Pediatrics, Univ of Hawaii John A. Burns School of Medicine, Honolulu, Hawaii; 2Kapi'olani Medical Specialists, Honolulu, Hawaii

American Journal of Medical Genetics Part A, July 2015, 167(7): 1582–1586.

... The results showed a 31.62 MB mosaic terminal duplication of 20pter>q11.21 consistent with a supernumerary mosaic isodicentric chromosome 20 (arr 20p13q11.21 (61,568-31,620, 951) ×2~4 (Affymetrix Cytoscan HD based on the GRCh37/HG19 assembly, Affymetrix, Santa ...

153-

Mosaic trisomy 15 in a liveborn infant

Jacob McPadden1, Benjamin M. Helm1,2, Brooke B. Spangler1,2, Leslie P. Ross3, Debra B. Boles4 andSamantha A. Schrier Vergano1,2,*

1Dept of Pediatrics, Eastern Virginia Medical School, Norfolk, Virginia; 2Division of Medical Genetics and Metabolism, Children's Hospital of The King's Daughters, Norfolk, Virginia; 3Quest Diagnostics, Denver, Colorado; 4Quest Diagnostics, Nichols Institute, Chantilly, Virginia

American Journal of Medical Genetics Part A, April 2015, 167(4): 821–825.

... A sample of the proband's blood was analyzed using an oligo-SNP (oligonucleotide, single nucleotide polymorphism, Affymetrix CytoScan HD) assay through Quest Diagnostics (Chantilly, VA).

154- *Meeting abstract*

MSS HNPCC frequently contain CNVs in chromatin regulators

WeiXiang Chen1, Xia sheng2, and Jun Ding1

1Shanghai Gongli Hospital, Shanghai, China; 2Shanghai Putuo Hospital, Shanghai, China.

Cancer Research, 2015;75(15 Suppl):Abst #3902. [AACR 106th Annual Mtg, Apr 18-22, 2015, Philadelphia, PA]

Note: This abstract was not presented at the meeting.

… We examined genomic DNA samples from 95 primary HNPCCs and adjacent normal appearing mucosae from patients undergoing surgery. We performed the copy number variation (CNV) of the entire genome using the Cytoscan HD Array in 20 MSS and 4 MSI HNPCC tumors and their adjacent normal tissues. Extensive comparison with somatic alterations in MSI HNPCCs allowed segregation of MSS HNPCC-exclusive alterations. ...

155- *Meeting abstract*

Multicenter Total Therapy Gimema LAL 1509 Protocol for De Novo Adult Ph+ Acute Lymphoblastic Leukemia (ALL) Patients. Updated Results and Refined Genetic-Based Prognostic Stratification

Sabina Chiaretti*,1, Antonella Vitale*,1, Loredana Elia*,1, Anna Lucia Fedullo*,1, Silvana Albino*,1, Alfonso Piciocchi*,2, Paola Fazi*,2, Francesco Di Raimondo3, Antonella Fornaro*,4, Francesco Fabbiano5, Alfonso Maria D'Arco*,6, Giovanni Martinelli, MD PhD7, Francesca Ronco*,8, Lidia Edwige Santoro*,9, Nicola Cascavilla, MD*,10, Piero Galieni*,11, Alessandra Tedeschi, MD*,12, Simona Sica*,13, Nicola Di Renzo14, Angela Melpignano*,15, Angelo Michele Carella16, Felicetto Ferrara17, Marco Vignetti*,2, and Robin Foà*,1

1Hematology, Dept of Cellular Biotechnologies and Hematology, "Sapienza" University, Rome, Italy; 2GIMEMA Data Center, Rome, Italy; 3Dept of Biomedical Sciences, Division of Hematology, Ospedale Ferrarotto, University of Catania, Italy, Catania, Italy; 4Dept of Hematology, Ospedale Civile dello Spirito Santo - Pescara, Pescara, Italy; 5Ospedali Riuniti Villa Sofia-Cervello, Palermo, Italy, Palermo, Italy; 6Division of Onco-hematology - Ospedale Umberto I - Nocera Inferiore (SA), Nocera Inferiore, Italy;7Bologna University School of Medicine, Bologna, Italy; 8hematology, azienda ospedaliera bianchi-melacrino-morelli, reggio calabria, Italy; 9Div of Hematology -Azienda Ospedaliera S.G.Moscati, Avellino, Italy, Avellino, Italy; 10Hematology and BMT Unit, Casa Sollievo della Sofferenza, San Giovanni Rotondo, Italy; 11Hematology and Cellular Therapy, Ospedale Mazzoni, Ascoli Piceno, Italy; 12Div of Hematology, Niguarda Ca' Granda Hospital, Milan, Italy; 13Institute of Hematology, Catholic University, Rome, Italy; 14Hematology, Vito Fazzi Hospital, Lecce, Italy; 15Ospedale A. Perrino, Brindisi, Brindisi, Italy; 16Div of Hematology 1, IRCCS A.O.U. San Martino IST, Genova, Genova, Italy; 17Div of Hematology, Cardarelli Hospital, Napoli, Italy;

Blood, 3 December 2015, 126(23): Abstract #81.

... Mutational screening was performed in relapsed cases, based on material availability. SNP array analysis was carried out using the Cytoscan HD arrays (Affymetrix, Santa Clara, CA) to identify genomic aberrations. ... SNP array analysis, performed in 39 cases with available DNA, showed that the most frequent aberrations were deletions of IKZF1 (85%), PAX5 (38%), CDKN2A/B (33%), MLLT3 (33%), RB1 (28%) and JAK2 (28%). ...

156-

Mutational landscape and clonal architecture in grade II and III gliomas

Hiromichi Suzuki, Kosuke Aoki, Kenichi Chiba, Yusuke Sato, Yusuke Shiozawa, Yuichi Shiraishi, Teppei Shimamura, Atsushi Niida, Kazuya Motomura, Fumiharu Ohka, Takashi Yamamoto, Kuniaki Tanahashi, Melissa Ranjit, Toshihiko Wakabayashi, Tetsuichi Yoshizato, Keisuke Kataoka, Kenichi Yoshida, Yasunobu Nagata, Aiko Sato-Otsubo, Hiroko Tanaka, Masashi Sanada, Yutaka Kondo, Hideo Nakamura, Masahiro Mizoguchi, Tatsuya Abe, Yoshihiro Muragaki, Reiko Watanabe, Ichiro Ito, Satoru Miyano, Atsushi Natsume & Seishi Ogawa

Nature Genetics, online 13 April 2015, 47: 458–468.

… We assessed the genome-wide copy number alterations for 291 tumors using GeneChip Human Mapping 250K NspI or CytoScan HD arrays (Affymetrix) ... For GISTIC analysis, the different probe sets for the different array platforms (250K NspI, CytoScan HD and SNP Array 6.0) were adjusted by uniformly using the 250K NspI SNPs as surrogates for SNPs that were marginally located within the abnormal copy number segments on the CytoScan HD and SNP Array 6.0.

157-

MYC amplification in multiple marker chromosomes and EZH2 microdeletion in a man with acute myeloid leukemia

Zhifu Xiang, Al-Ola Abdallah, Rangaswamy Govindarajan, Paulette Mehta, Peter D. Emanuel, Peter Papenhausen, Steven A. Schichman

Cancer Genetics, March 2015, 208(3): 96–100.

… Whole genome SNP was performed using Affymetrix Genome-wide Cytoscan SNP Array (~2.7 million probes with ~700,000 SNPs) following standard protocol per the manufacturer's instructions. ...

158-

Neuroblastoma patient-derived orthotopic xenografts retain metastatic patterns and geno- and phenotypes of patient tumours

Noémie Braekeveldt1, Caroline Wigerup1, David Gisselsson2,3, Sofie Mohlin1, My Merselius1, Siv Beckman1, Tord Jonson2, Anna Börjesson4, Torbjörn Backman4, Irene Tadeo5, Ana P. Berbegall5,6, Ingrid Öra7, Samuel Navarro6, Rosa Noguera6, Sven Påhlman1, Daniel Bexell1,*

1Translational Cancer Research, Lund University, Lund, Sweden; 2Dept of Clinical Genetics, Lund University, Lund, Sweden; 3Dept of Pathology, University and Regional Laboratories, Lund, Sweden; 4Dept of Paediatric Surgery, Skåne University Hospital, Lund, Sweden; 5Medical Research Foundation INCLIVA, Hospital Clínico, Valencia, Spain; 6Dept of Pathology, Medical School, University of Valencia, Spain; 7Dept of Paediatrics, Clinical Sciences, Lund University, Lund, Sweden

International Journal of Cancer, 1 March 2015, 136(5): E252–E261.

… DNA was extracted from the cells and tissues using DNeasy Blood and Tissue Kit (Qiagen, Hilden, Germany) according to the instructions. We used the Affymetrix CytoScan HD platform for SNP array analysis of cultured SK-N-BE(2)c cells, SK-N-BE(2)c tumours, PDXs and patient tumour #3.

159- *Meeting abstract*

NEW DELETION OF JAK2 DETECTED BY SNP ARRAY: A ROLE IN OVERALL SURVIVAL IN ACUTE MYELOID LEUKEMIA PATIENTS

M.C. Fontana,1,* V. Guadagnuolo,1,* C. Papayannidis,1 I. Iacobucci,1 M. Manfrini,1 A. Padella,1 G. Simonetti,1 A. Ferrari,1 G. Marconi,1 S. Paolini,1 M.C. Abbenante,1 S. Parisi,1 C. Sartor,1 E. Ottaviani,1 M. Cavo,1 R. Kralovics,2 J. Milosevic,2 N. Them,2 D. Chen,2 G. Martinelli1

1Dept of Hematology and Medical Sciences "L. and A. Seràgnoli", Bologna University, Bologna, Italy; 2Research Center for Molecular Medicine of the Austrian Academy of Sciences, Wien, Austria; *These authors contribute equally to this work

Haematologica, 2015, 100(Sup3) : Abstr #C080, p.18. [45° Congress of the Italian Society of Hematology

Florence, Italy, October 4-7, 2015

… Our objective is to evaluate the prognostic impact of these genetic alterations on clinical outcome. Materials and Methods: We analyzed 272 AML patients (pts): 229 performed by SNP Array 6.0 (Affymetrix), 6 by CytoScan HD Array (Affymetrix) and 37 performed by Cytoscan HD Array ... Conclusions: By SNP arrays we have identified CNAs involving important cancer genes in AML and we showed that a new deletion in JAK2 may play a role in the overall survival. …

160-

Next Generation Diagnostics: Gene Panel, Exome or Whole Genome?

Yu Sun1,3, Claudia A.L. Ruivenkamp1, Mariëtte J.V. Hoffer1, Terry Vrijenhoek2, Marjolein Kriek1, Christi J. van Asperen1, Johan T. den Dunnen1 andGijs W.E. Santen1,*

1Dept of Clinical Genetics, Leiden University Medical Center, Leiden, The Netherlands; 2Centre for Genome Diagnostics, Department of Medical Genetics, Universitair Medisch Centrum Utrecht, Utrecht, The Netherlands; 3Shanghai Institute for Pediatric Research, Xin Hua Hospital affiliated to Shanghai Jiaotong University School of Medicine, Shanghai, China

Human Mutation, June 2015, 36(6): 648–655.

... Twelve of these 13 samples were also analyzed using Affymetrix Cytoscan HD arrays ... An additional 5/21 (24%) variants would probably have been identified by the Affymetrix Cytoscan HD array, FISH studies or by CNV-calling on exome data …

161-

NFkB is activated by multiple mechanisms in hairy cell leukemia

Stefan Nagel*, Stefan Ehrentraut, Corinna Meyer, Maren Kaufmann, Hans G. Drexler, Roderick A.F. MacLeod

Dept of Human and Animal Cell Lines, Leibniz-Institute DSMZ – German Collection of Microorganisms and Cell Cultures, Braunschweig, Germany

Genes, Chromosomes and Cancer, July 2015, 54(7): 418–432.

… Labeling, hybridization, and washing were performed using the recommended kits and CytoScan HD arrays according to the manufacturers protocols (Affymetrix, High Wycombe, UK).

162- *Meeting abstract*

Novel germline copy number variations in patients with hereditary colorectal carcinoma with no mutation in the mismatch repair genes

Rolando André Rios Villacis1, Érika Maria Monteiro Santos2, Dirce Maria Carraro1, Benedito Mauro Rossi3, and Silvia Regina Rogatto4

1CIPE - Research Center, AC Camargo Cancer Center, São Paulo, Brazil; 2Oncology Center, Sírio-Libanês Hospital, São Paulo, Brazil; 3Dept of Oncogenetics, Barretos Cancer Hospital, São Paulo, Brazil; 4CIPE - Research Center, AC Camargo Cancer Center/Dept of Urology, Faculty of Medicine, UNESP, São Paulo/Botucatu, Brazil.

Cancer Research, 2015;75(15 Suppl):Abst #2754. [AACR 106th Annual Mtg, Apr 18-22, 2015, Philadelphia, PA]

…100 healthy Brazilian individuals (evaluated with 180K Agilent platform) and 1038 phenotypically healthy individuals (Affymetrix database). It was identified 263 CNVs: 86 were classified as rare (≤ 1% of reference databases) and 10 (in different chromosomes and cases) were completely new. Eleven out of 58 cases were re-evaluated using a higher-density microarray platform (Affymetrix CytoScan HD, 2.6 million probes), being the data analyzed with the Chromosome Analysis Suite v2.1 (Affymetrix) software. We identified more CNVs per case with the Affymetrix platform compared to the Agilent platform (179 to 68). ... By using Affymetrix CytoScan HD platform, two unrelated cases presented a rare duplication of 82kb on chromosome 2p22.3 encompassing two genes and one microRNA, that were previously associated with CRC. ...

163-
Novel homozygous deletion of segmental KAL1 and entire STS cause Kallmann syndrome and X-linked ichthyosis in a Chinese family
H. Xu1, Z. Li2, T. Wang1, S. Wang1, J. Liu1* and D. W. Wang2*
1Dept of Urology, Tongji Hospital, Tongji Medical College, Huazhong University of Science and Technology, Wuhan, China; 2Dept of Internal Medicine and Gene Therapy Center, Tongji Hospital, Tongji Medical College, Huazhong University of Science and Technology, Wuhan, China
Andrologia, December 2015, 47(10) : 1160–1165.
… A genomic hybridisation was performed with the CytoScan™ HD array (Affymetrix, Santa Clara, CA, USA) in accordance with the manufacturer's instruction in patient III-2. The array is characterised by more than 2 600 000 copy number variation markers including 750 000 SNP ...

164-
NUP98/11p15 translocations affect CD34+ cells in myeloid and T lymphoid leukemias
Barbara Crescenzi a, Valeria Nofrini a, Gianluca Barba a, Caterina Matteucci a, Danika Di Giacomo a, Paolo Gorello a, Berna Beverloo b, Antonella Vitale c, Iwona Wlodarska d, Peter Vandenberghe d, Roberta La Starza a, Cristina Mecucci a
a Laboratory of Molecular Medicine, CREO, University of Perugia and A.O. Perugia, 06132 Perugia, Italy; b Dept of Clinical Genetics, Erasmus MC, 3000 CB Rotterdam, The Netherlands; c Hematology, Dept of Cellular Biotechnologies and Hematology, La Sapienza University, Via Benevento 6, 06161 Rome, Italy; d Center for Human Genetics, K.U. Leuven, Gasthuisberg, Herestraat 49, Box 602, B-3000 Leuven, Belgium
Leukemia Research, July 2015, 39(7): 769–772.
… Single Nucleotide Polymorphism-Array (SNPa) was performed with CytoScan HD Affymetrix platform, ... high quality genomic DNA obtained from unsorted bone marrow cells of patients ...

165-
Panventriculomegaly with a wide foramen of Magendie and large cisterna magna
Hiroshi Kageyama, MD1,2,3, Masakazu Miyajima, MD, PhD1, Ikuko Ogino, BPHARM1, Madoka Nakajima, MD, PhD1, Kazuaki Shimoji, MD, PhD1, Ryoko Fukai, MD, PhD2, Noriko Miyake, MD, PhD2, Kenichi Nishiyama, MD, PhD4, Naomichi Matsumoto, MD, PhD2, and Hajime Arai, MD, PhD1
1Dept of Neurosurgery, Graduate School of Medicine, Juntendo University, Tokyo; 2Dept of Human Genetics, Yokohama City University Graduate School of Medicine, Yokohama; 3Dept of Neurosurgery, Kuki General Hospital, Kuki, Saitama; and 4Brain Research Institute, University of Niigata, Japan
Journal of Neurosurgery, published online December 4, 2015. DOI: 10.3171/2015.6.JNS15162.
... the first clinical, radiological, and genetic studies of panventriculomegaly defined by a wide foramen of Magendie and large cisterna magna. ... copy number analysis was performed using a Cytoscan HD Array (Affymetrix) ...

166-
PAPSS2 deficiency causes androgen excess via impaired DHEA sulfation--in vitro and in vivo studies in a family harboring two novel PAPSS2 mutations.
Oostdijk W, Idkowiak J, Mueller JW, House PJ, Taylor AE, O'Reilly MW, Hughes BA, de Vries MC, Kant SG, Santen GW, Verkerk AJ, Uitterlinden AG, Wit JM, Losekoot M, Arlt W.
J Clin Endocrinol Metab. 2015 Apr;100(4):E672-80.
... Cytogenetic microarray analysis was performed using the CytoScan HD Array (Affymetrix) according to the manufacturer's protocol. Copy number was assessed using Chromosome Analysis Suite software (Affymetrix).

167-
Parkinson's Disease in Saudi Patients: A Genetic Study
Bashayer R. Al-Mubarak, Saeed A. Bohlega, Thamer S. Alkhairallah, Amna I. Magrashi, Maha I. AlTurki, Dania S. Khalil, Basma S. AlAbdulaziz, Hussam Abou Al-Shaar, Abeer E. Mustafa, Eman A. Alyemni, Bashayer A. Alsaffar, Asma I. Tahir, Nada A. Al Tassan
PlosONE, 14 August 2015, 10(8): e0135950.
... Out of the 98 samples, 25 representative samples were screened for dosage alterations of both common and associated-PD genes using the Cyto Scan HD array (Affymetrix, Santa Clara, CA,USA) which contains 2.6 million markers for genome coverage. The data was analyzed using the Chromosome Analysis Suite version Cyto 3.0 using GRC 38/hg19 of the UCSC Genome Browser.

168-
Partial USH2A deletions contribute to Usher syndrome in Denmark
Shzeena D1, Rendtorff ND2, Kann E1, Albrechtsen A3, Mehrjouy MM4, Bak M4, Tommerup N4, Tranebjærg L2, Rosenberg T5, Jensen H5, Møller LB1.
1Clinical Genetics Clinic, The Kennedy Center, Rigshospitalet, University of Copenhagen, Glostrup, Denmark.; 21] Dept of Cellular and Molecular Medicine, Univ of Copenhagen, Copenhagen, Denmark [2] Dept of

Otorhinolaryngology, Head and Neck Surgery and Audiology, Bispebjerg Hospital/Rigshospitalet, Copenhagen, Denmark.; 3Dept of Biology, Computational and RNA Biology, University of Copenhagen, Copenhagen, Denmark.; 4Dept of Cellular and Molecular Medicine, Univ of Copenhagen, Copenhagen, Denmark.; 5Dept of Ophthalmology, The National Eye Clinic, Copenhagen University Hospital, The Kennedy Center, Glostrup, Denmark.
European Journal of Human Genetics, December 2015, 23(12):1646-1651.
… The Affymetrix CytoScan HD array (AROS Applied Biotechnology AS, Aarhus, Denmark) was used to confirm ...

169-
PAX5-KIAA1549L: a novel fusion gene in a case of pediatric B-cell precursor acute lymphoblastic leukemia
Stefanie Anderl1, Margit König1, Andishe Attarbaschi2, 3, Sabine Strehl1*
1 CCRI, Children's Cancer Research Institute, St. Anna Kinderkrebsforschung e.V., Vienna, Austria ; 2 Dept of Pediatrics, St. Anna Children's Hospital, Vienna, Austria ; 3 Medical University of Vienna, Vienna, Austria
Molecular Cytogenetics, 8 July 2015, 8:48
... Our report underlines the high diversity of PAX5 fusion partners in BCP-ALL and we describe the second involvement of KIAA1549L in a genetic rearrangement in acute leukemia. ... Genome-wide CNAs were determined using the CytoScan HD array platform (Affymetrix, USA).

170-
Persistence of müllerian duct structures in a genetic male with distal monosomy 10q
Mustafa Tosur1,*, Cara A. Geary1, Reuben Matalon1, Ravi S. Radhakrishnan1,2, Leonard E. Swischuk3, William F. Tarry2, Jianli Dong4 andPhillip D. K. Lee1
1Dept of Pediatrics, University of Texas Medical Branch, Galveston, TX; 2Dept of Surgery, Univ of Texas Medical Branch, Galveston, TX; 3Dept of Radiology, University of Texas Medical Branch, Galveston, TX; 4Dept of Pathology, Univ of Texas Medical Branch, Galveston, TX
American Journal of Medical Genetics Part A, April 2015, 167(4):791–796.
… Chromosome karyotyping analysis showed 46,XY,add(10)(q25.2). Cytogenomic SNP microarray (Cytoscan HD Array, Affymetrix, Santa Clara, CA) revealed an 8.2 MB interstitial deletion of 10q25.3q26.13 (117,012,878-125,217,066) x1, indicating monosomy of this region.

171-
Positional mapping of PRKD1, NRP1 and PRDM1 as novel candidate disease genes in truncus arteriosus
Ranad Shaheen1, Amal Al Hashem2,3, Mohammed H Alghamdi4, Mohammed Zain Seidahmad5, Salma M Wakil1, Khalid Dagriri6, Bernard Keavney7, Judith Goodship8, Saad Alyousif5, Fahad M Al-Habshan9, Khalid Alhussein5, Agaadir Almoisheer1, Niema Ibrahim1, Fowzan S Alkuraya1,3
1Dept of Genetics, King Faisal Specialist Hospital and Research Center, Riyadh, Saudi Arabia ; 2Dept of Pediatrics, Prince Sultan Military Medical City, Riyadh, Saudi Arabia ; 3Dept of Anatomy and Cell Biology, College of Medicine, Alfaisal University, Riyadh, Saudi Arabia ; 4Dept of Cardiac Sciences, College of Medicine, King Saud University, Riyadh, Saudi Arabia ; 5Dept of Pediatrics, Security Forces Hospital, Riyadh, Saudi Arabia ; 6Dept of Pediatric Cardiology, Prince Sultan Cardiac Center, Riyadh, Saudi Arabia ; 7Institute of Cardiovascular Sciences, University of Manchester, Manchester, UK ; 8Institute of Genetic Medicine, Newcastle University, Newcastle upon Tyne, UK ; 9Dept of Cardiology, King Saud bin Abdulaziz University for Health Sciences, Riyadh, Saudi Arabia
Journal of Medical Geneics, 2015, 52:322-329.
… Molecular karyotyping Detection of CNVs at the DNA level (molecular karyotyping) was performed using the CytoScan HD Array platform according to the manufacturer's instructions.

172-
Prader–Willi syndrome and Tay–Sachs disease in association with mixed maternal uniparental isodisomy and heterodisomy 15 in a girl who also had isochromosome Xq
Susan Zeesman1, Elizabeth McCready2,3, Bekim Sadikovic2,3 and Małgorzata JM Nowaczyk1,2,3,*
1Dept of Pediatrics, McMaster University, Hamilton, Ontario, Canada; 2Dept of Pathology and Molecular Medicine, McMaster University, Hamilton, Ontario, Canada; 3Hamilton Regional Laboratory Medicine Program, Hamilton Health Sciences and St. Joseph's Healthcare, Hamilton, Ontario, Canada
American Journal of Medical Genetics Part A, January 2015, 167(1):180–184.
… To further investigate this finding, SNP oligonucleotide microarray (Cytoscan HD microarray, Affymetrix, Santa Carla, USA) was performed. ... Figure 1. Characterization of chromosome 15 isodisomy segment by SNP oligonucleotide array. DNA extracted from peripheral blood was analyzed using the Cytoscan HD genomic microarray (Affymetrix ...

173-

Prader-Willi Syndrome With a Long-Contiguous Stretch of Homozygosity Not Covering the Critical Region

Xie Yingjun, MD1, Zhou Yi, MD1, Wu Jianzhu, BD1, Sun Yunxia, MD2, Chen Yongzhen, BD1, Zhong Liangying, MD3, Jing Xiangyi, PhD4, Fang Qun, MD1
1Fetal Medicine Center, the First Affiliated Hospital of Sun Yat-sen University, Guangzhou, China ; 2Dept of Neonatology, Guangdong General Hospital, Guangdong Academy of Medical Sciences, Guangzhou, China ; 3Dept of Laboratory Medicine, the First Affiliated Hospital of Sun Yat-sen University, Guangzhou, China; 4Dept of Medical Genetics, Zhongshan School of Medicine and Center for Genome Research, Sun Yat-Sen University, Guangzhou, China

J Child Neurol, Mar 2015, 30:371-377.

… DNA was amplified, labeled, and hybridized to the CytoScan HD array (Affymetrix) platform according to the manufacturer's... CEL files obtained by scanning CytoScan arrays were analyzed with Chromosome Analysis Suite Software...

174-

Prenatal diagnosis of chromothripsis, with nine breaks characterized by karyotyping, FISH, microarray and whole-genome sequencing†

M. J. Macera1, A. Sobrino1, B. Levy2, V. Jobanputra2, V. Aggarwal2, A. Mills1, C. Esteves1, C. Hanscom4, S. Pereira5, V. Pillalamarri4, Z. Ordulu, C. Morton5,6, M. Talkowski4 andD. Warburton3,*
1New York-Presbyterian Hospital, Columbia Univ Medical Center, New York, NY; 2Dept of Pathology, Columbia Univ, New York, NY; 3Depts of Genetics and Development and Pediatrics, Columbia Univ, New York, NY; 4Center for Human Genetic Research and Dept of Neurology, Massachusetts General Hospital and Harvard Medical School, Boston, MA; 5Depts of Obstetrics, Gynecology and Reproductive Biology, Harvard Medical School, Boston, MA; 6Pathology, Brigham and Women's Hospital, Harvard Medical School, Boston, MA

Prenatal Diagnosis, March 2015, 35(3):299–301.

… SNP oligonucleotide microarray analysis (Affymetrix Cytoscan HD) on fetal DNA showed no loss or gain of chromosomal material at any of the breakpoints.

175-

Prognostic significance of acquired copy-neutral loss of heterozygosity in acute myeloid leukemia

Christine M. Gronseth1, Scott E. McElhone1, Barry E. Storer PhD2, Kathleen A. Kroeger1, Vicky Sandhu MD3, Matthew L. Fero MD3,4,6, Frederick R. Appelbaum MD3,4,6, Elihu H. Estey MD3,4,6, Min Fang MD, PhD3,5,6,*
1Cytogenetics Dept, Seattle Cancer Care Alliance, Seattle, WA; 2Clinical Statistics Dept, Fred Hutchinson Cancer Research Center, Seattle, WA; 3Clinical Research Div, Fred Hutchinson Cancer Research Center, Seattle, WA; 4Dept of Medicine, University of Washington, Seattle, WA; 5Dept of Pathology, University of Washington, Seattle, WA; 6Seattle Cancer Care Alliance, Seattle, WA

Cancer, September 1, 2015, 121(17): 2900–2908.

... At the time of diagnosis or disease recurrence, CGAT was performed on DNA from bone marrow or blood specimens using CytoScanHD (Affymetrix ...

176-

Quantitative analysis of genomic DNA degradation in whole blood under various storage conditions for molecular diagnostic testing

Jessalyn Permenter a, Arjun Ishwar a, Angie Rounsavall a, Maddie Smith a, Jennifer Faske a, Charles J. Sailey a, b, 1, Maria P. Alfaro a, b
a Dept of Molecular Genetic Pathology, Arkansas Children's Hospital, 1 Children's Way, Little Rock, AR 72212, USA; b University of Arkansas for Medical Sciences, Little Rock, AR 72212, USA

Molecular and Cellular Probes, December 2015, 29(6): 449–453.

... Affymetrix Cytogenomic SNP Array as a high complexity test--Approximately 250 ng of gDNA was hybridized onto the CytoScan® HD array ... quality of SNP array data (Cytoscan HD, ... stored at room temperature was considered inadequate after 30 days.

177-

RAG1 Deficiency May Present Clinically as Selective IgA Deficiency

Tamaki Kato(1), Elena Crestani(2), Chikako Kamae(1), Kenichi Honma(1), Tomoko Yokosuka(3)
Takeshi Ikegawa(4), Naonori Nishida(5), Hirokazu Kanegane(5)(6), Taizo Wada(7), Akihiro Yachie(7), Osamu Ohara(8)(9), Tomohiro Morio(6), Luigi D. Notarangelo(2), Kohsuke Imai(1)(6), Shigeaki Nonoyama(1)
1. Dept of Pediatrics, National Defense Medical College, Tokorozawa, Saitama, Japan; 2. Div of Immunology, Children's Hospital Boston, Harvard Medical School, Boston, MA, USA; 3. Dept of Hemato-oncology/Regeneration Medicine, Kanagawa Children's Medical Center, Yokohama, Kanagawa, Japan; 4. Dept of Pediatrics, Yokohama Rosai Hospital, Yokohama, Kanagawa, Japan; 5. Dept of Pediatrics, University of Toyama, Toyama, Japan; 6. Dept of Pediatrics, Tokyo Medical and Dental University (TMDU), 1-5-45, Yushima, Bunkyo-ku, Tokyo, 113-8519, Japan; 7. Dept of Pediatrics, Kanazawa University, Kanazawa, Ishikawa, Japan; 8. Laboratory for Integrative

Genomics, RIKEN Center for Integrative Medical Sciences, Yokohama, Kanagawa, Japan; 9. Dept of Technology Development, Kazusa DNA Research Institute, Kisarazu, Chiba, Japan
Journal of Clinical Immunology, April 2015, 35(3):280-288.
... A copy number and loss of heterozygosity (LOH) analysis in the patient was performed using a CytoScan HD Array (Affymetrix, Santa Clara, California, USA), according to the manufacturer's protocol.

178-
Rare genomic rearrangement in a boy with Williams–Beuren syndrome associated to XYY syndrome and intriguing behavior
Roberta L. Dutra1,2,*, Flavia B. Piazzon2, Évelin A. Zanardo2, Thais Virginia Moura Machado Costa2, Marília M. Montenegro1,2, Gil M. Novo-Filho1,2, Alexandre T. Dias2, Amom M. Nascimento1,2, Chong Ae Kim1 and Leslie D. Kulikowski2,3
1Genetics Unit, Instituto da Criança, Faculdade de Medicina da Universidade de São Paulo (USP), São Paulo, Brazil; 2Dept of Pathology, Cytogenomics Lab - LIM03, Faculdade de Medicina da Universidade de São Paulo (USP), São Paulo, Brazil; 3Dept of Collective Health - Human Reproduction and Genetics Center, Faculdade de Medicina do ABC, Santo André, São Paulo, Brazil
American Journal of Medical Genetics Part A, December 2015, 167(12): 3197–3203.
... (SNP-array) confirmed the alterations found by MLPA and revealed ... pathogenic CNVs, in the chromosomes 7 and X. ... demonstrates an association not yet described ... between Williams–Beuren syndrome and 47,XYY. ... performed with the CytoScan™ HD Array (Affymetrix ...

179-
rCGH: a comprehensive array-based genomic profile platform for precision medicine
Frederic Commo1,2,*, Justin Guinney2, Charles Ferté1,2, Brian Bot2, Celine Lefebvre1, Jean-Charles Soria1,3 and Fabrice André1,3
1INSERM U981, Gustave Roussy, University Paris-Sud, Villejuif 94805, France, 2Sage Bionetworks, Seattle, WA 98109; 3Dept of Medical Oncology, Gustave Roussy, Villejuif 94805, France
Bioinformatics, online 26 Dec 2015; 10.1093/bioinformatics/btv718.
… As input rCGH supports Agilent Human CGH data, from 44K to 400K, and Affymetrix, SNP6 and cytoScanHD. All are provided in text format by platform-specific softwares: … Affymetrix cychp.txt, cnchp.txt or probeset.txt files are obtained by processing Affymetrix CEL files through ChAS or Affymetrix Power Tools (APT) softwares …

180-
RCSD1-ABL1 Translocation Associated with IKZF1 Gene Deletion in B-Cell Acute Lymphoblastic Leukemia
Shawana Kamran, 1 , * Gordana Raca, 2 and Kamran Nazir 3
1Shifa International Hospital, Islamabad 44000, Pakistan; 2University of Chicago, Chicago, IL 60637; 3Combined Military Hospital, Kharian 50090, Pakistan
Case Reports in Hematology, online 27 October 2015; 2015: 353247.
... SNP array testing was performed using the Affymetrix CytoScan HD arrays ...

181-
RCSD1–ABL2 fusion resulting from a complex chromosomal rearrangement in high-risk B-cell acute lymphoblastic leukemia
Gordana Raca, Sandeep Gurbuxani, Zhiyu Zhang, Zejuan Li, Madina Sukhanova, Jennifer McNeer,
Wendy Stock
1Dept of Medicine, University of Chicago, Chicago, IL; 2Dept of Pathology, Univ of Chicago, Chicago, IL; 3Dept of Pediatrics, University of Chicago, Chicago, IL
Leukemia & Lymphoma, 2015 Apr;56(4):1145-7.
… (CMA) was performed using the CytoScan® HD arrays (Affymetrix Inc., Santa Clara CA), to look for submicroscopic copy number alterations and/or extended regions of loss of heterozygosity. ...

182-
Recessive ITPA mutations cause an early-infantile encephalopathy
Sietske H. Kevelam1,2, Jörgen Bierau PhD3, Ramona Salvarinova MD, FRCPC, FCCMG4, Shakti Agrawal MRCP, MRCPCH5, Tomas Honzik MD, PhD6, Dennis Visser3, Marjan M. Weiss PhD7, Gajja S. Salomons PhD2,8, Truus E. M. Abbink PhD1,2, Quinten Waisfisz PhD7 and Marjo S. van der Knaap MD, PhD1,2,9,*
1Dept of Child Neurology, VU University Medical Center, Amsterdam, the Netherlands; 2Neuroscience Campus Amsterdam, VU University, Amsterdam, the Netherlands; 3Dept of Clinical Genetics, Maastricht University Medical Center, Maastricht, the Netherlands; 4Div of Biochemical Diseases, Dept of Pediatrics, University of British Columbia, British Columbia Children's Hospital, Vancouver, British Columbia, Canada; 5Dept of Pediatric Neurology, Birmingham Children's Hospital, Birmingham, UK; 6Dept of Pediatrics, First Faculty of Medicine, Charles University in Prague and General University Hospital in Prague, Prague, Czech Republic; 7Dept of Clinical Genetics, VU University Medical Center, Amsterdam, the Netherlands; 8Dept of Clinical Chemistry, Metabolic Unit,

VU University Medical Center, Amsterdam, the Netherlands; 9Dept of Functional Genomics, Center for Neurogenomics and Cognitive Research, VU University, Amsterdam, the Netherlands
Annals of Neurology, October 2015, 78(4): 649–658.
... SNP array revealed one overlapping homozygous region on chromosome 20 in the consanguineous families. ... the second consanguineous family were subjected to SNP-array analysis (Affymetrix CytoScan HD array) ...

183-
Recessive Mutations in the α3 (VI) Collagen Gene COL6A3 Cause Early-Onset Isolated Dystonia
Michael Zech1, 2, 3, 10, Daniel D. Lam2, 10, Ludmila Francescatto4, 10, Barbara Schormair1, 3, 5, Aaro V. Salminen3, Angela Jochim1, Thomas Wieland3, Peter Lichtner3, 5, Annette Peters6, Christian Gieger7, Hanns Lochmüller8, Tim M. Strom3, 5, Bernhard Haslinger1, 11, Nicholas Katsanis4, 11, Juliane Winkelmann1, 2, 3, 9
1 Neurologische Klinik und Poliklinik, Klinikum rechts der Isar, Technische Universität München, 81675 Munich, Germany; 2 Dept of Neurology and Neurological Sciences and Center for Sleep Sciences and Medicine, Stanford University School of Medicine, Palo Alto, CA 94304, USA; 3 Institut für Humangenetik, Helmholtz Zentrum München, 85764 Munich, Germany; 4 Center for Human Disease Modeling, Dept of Cell Biology, Duke University, Durham, NC 27710, USA; 5 Institut für Humangenetik, Technische Universität München, 81675 Munich, Germany; 6 Institute of Epidemiology II, Helmholtz Zentrum München, 85764 Munich, Germany; 7 Institute of Genetic Epidemiology, Helmholtz Zentrum München, 85764 Munich, Germany; 8 John Walton Centre for Muscular Dystrophy Research, MRC Centre for Neuromuscular Diseases, Institute of Genetic Medicine, Newcastle University, Newcastle upon Tyne NE1 3BZ, UK; 9 Munich Cluster for Systems Neurology, SyNergy, 81377 Munich, Germany
American Journal of Human Genetics, Volume 96, Issue 6, 4 June 2015, Pages 883–893
... screening for larger CNVs was done with the CytoScan HD Array from Affymetrix ...

184-
Recurrent CDKN1B (p27) mutations in hairy cell leukemia
Sascha Dietrich1, Jennifer Hüllein2, Stanley Chun-Wei Lee3, Barbara Hutter4, David Gonzalez5, Sandrine Jayne6, Martin J.S. Dyer6, Małgorzata Oleś7, Monica Else8, Xiyang Liu2, Mikołaj Słabicki2, Bian Wu2, Xavier Troussard9, Jan Dürig10, Mindaugas Andrulis11, Claire Dearden12, Christof von Kalle13, Martin Granzow14, Anna Jauch14, Stefan Fröhling2, Wolfgang Huber7, Manja Meggendorfer15, Torsten Haferlach15, Anthony D. Ho1, Daniela Richter2, Benedikt Brors4, Hanno Glimm2, Estella Matutes12, Omar Abdel Wahab3, Thorsten Zenz1,*
1 Dept of Medicine V, University Hospital Heidelberg, Heidelberg, Germany; 2 Dept of Translational Oncology, National Center for Tumor Diseases (NCT) and German Cancer Research Center (dkfz), Heidelberg, Germany; 3 Human Oncology and Pathogenesis Program, Leukemia Service, Dept. of Medicine, Memorial Sloan Kettering Cancer Center, New York, NY, United States; 4 Div. Applied Bioinformatics, German Cancer Research Center (DKFZ), Heidelberg, Germany; 5 Centre for Molecular Pathology, Royal Marsden NHS FTl, London, United Kingdom; 6 Ernest and Helen Scott Hematological Research Institute, University of Leicester, Leicester, United Kingdom; 7 Genome Biology Unit, European Molecular Biology Laboratory, Heidelberg, Germany; 8 Div of Molecular Pathology, The Institute of Cancer Research, London, United Kingdom; 9 Laboratoire d'Hematologie, CHU Cote de Nacre, Caen, France; 10 Dept of Hematology, University Hospital, Essen, Germany; 11 Dept of General Pathology, Institute of Pathology, University of Heidelberg, Heidelberg, Germany; 12 Royal Marsden Hospital, London, United Kingdom; 13 Heidelberg Center for Personalized Oncology, DKFZ-HIPO, DKFZ, Heidelberg, Germany; 14 Institute of Human Genetics, University Hospital Heidelberg, Germany; 15 MLL Munich Leukemia Laboratory, Munich, Germany
Blood, Aug 2015; 126: 1005 - 1008.
... This revealed loss of chromosome 21 in all three patients (supplemental material). Although these findings were validated by Affymetrix Cytoscan HD Arrays, CNV analysis of 7 additional patients failed to reveal chromosome 21 abnormalities ...

185-
Recurrent chromosomal gains and heterogeneous driver mutations characterise papillary renal cancer evolution
Michal Kovac, Carolina Navas, Stuart Horswell, Max Salm, Chiara Bardella, Andrew Rowan, Mark Stares, Francesc Castro-Giner, Rosalie Fisher, Elza C. de Bruin, Monika Kovacova, Maggie Gorman, Seiko Makino, Jennet Williams, Emma Jaeger, Angela Jones, Kimberley Howarth, James Larkin, Lisa Pickering, Martin Gore, David L. Nicol, Steven Hazell, Gordon Stamp, Tim O'Brien, Ben Challacombe, Nik Matthews, Benjamin Phillimore, Sharmin Begum, Adam Rabinowitz, Ignacio Varela, Ashish Chandra, Catherine Horsfield, Alexander Polson, Maxine Tran, Rupesh Bhatt, Luigi Terracciano, Serenella Eppenberger-Castori, Andrew Protheroe, Eamonn Maher, Mona El Bahrawy, Stewart Fleming, Peter Ratcliffe, Karl Heinimann, Charles Swanton & Ian Tomlinson
Nature Communications, 19 March 2015, 6:6336.
… Twenty-three tumours were genotyped using the Affymetrix 2.7 M Cytogenetics array or the CytoScan HD array.

186-
Recurrent deletions and duplications of chromosome 2q11.2 and 2q13 are associated with variable outcomes
Kacie N. Riley1,2, Lisa M. Catalano1, John A. Bernat3, Stacie D. Adams3, Donna M. Martin3,4, Seema R. Lalani5, Ankita Patel5, Rachel D. Burnside2, Jeffrey W. Innis3,4 andM. Katharine Rudd1,*
American Journal of Medical Genetics Part A, November 2015, 167(11): 2664–2673.
... Subjects 4–7 were tested using the Affymetrix CytoScan HD array and Subjects 11 and 14 were tested using the Illumina CytoSNP-850K BeadChip, both based on the GRCh37/hg19 genome assembly.

187-
Recurrent inactivating RASA2 mutations in melanoma
Rand Arafeh, Nouar Qutob, Rafi Emmanuel, Alona Keren-Paz, Jason Madore, Abdel Elkahloun, James S Wilmott, Jared J Gartner, Antonella Di Pizio, Sabina Winograd-Katz, Sivasish Sindiri, Ron Rotkopf, Ken Dutton-Regester, Peter Johansson, Antonia L Pritchard, Nicola Waddell, Victoria K Hill, Jimmy C Lin, Yael Hevroni, Steven A Rosenberg, Javed Khan, Shifra Ben-Dor, Masha Y Niv, Igor Ulitsky, Graham J Mann, Richard Scolyer, Nicholas K Hayward & Yardena Samuels
Nature Genetics, online 26 October 2015, 47:1408–1410.
... We profiled the copy number landscape of 22 samples using the CytoScan High-Definition array (Affymetrix) and found three focal deletions (13.6%). ...

188-
Relapse after Allogeneic Hematopoietic Cell Transplantation for Myelodysplastic Syndromes: Analysis of Late Relapse Using Comparative Karyotype and Chromosome Genome Array Testing
Cecilia Yeung1, 2, 3, Aaron T. Gerds4, Min Fang1, 2, 3, Bart L. Scott1, 2, 3, Mary E.D. Flowers1, 2, 3, Ted Gooley1, 2, H. Joachim Deeg1, 2, 3
1 Fred Hutchinson Cancer Research Center, Seattle, WA; 2 University of Washington, Seattle, WA; 3 Seattle Cancer Care Alliance, Seattle, WA; 4 Cleveland Clinic, Cleveland, OH
Biology of Blood and Bone Marrow Transplantation, September 2015, 21(9):1565–1575.
… CGAT, a combination of comparative genome hybridization (CGH) and single nucleotide polymorphism (SNP)-array, was used for the detection of DNA copy number aberration (CNA) or SNP using CytoScan HD (Affymetrix, ...

189-
Repressed BMP signaling reactivates NKL homeobox gene MSX1 in a T-ALL subset
Stefan Nagel, Stefan Ehrentraut, Corinna Meyer, Maren Kaufmann, Hans G. Drexler, and Roderick A. F. MacLeod
Department of Human and Animal Cell Lines, Leibniz-Institute DSMZ – German Collection of Microorganisms and Cell Cultures, Braunschweig, Germany
Leukemia & Lymphoma, February 2015, 56(2): 480-491.
... Labeling, hybridization and washing were performed using the recommended kits and CytoScan HD arrays according to the manufacturer's protocols (Affymetrix). The data were subsequently analyzed using Chromosome Analysis Suite software version 2.0.1.2 (Affymetrix). ...

190-
Reversing chromatin accessibility differences that distinguish homologous mitotic metaphase chromosomes
Wahab A. Khan1, Peter K. Rogan23* and Joan H. M. Knoll13*
1 Dept of Pathology and Laboratory Medicine, Univ of Western Ontario, London N6A 5C1, ON, Canada; 2 Depts of Biochemistry, Computer Science, and Oncology, University of Western Ontario, London N6A 5C1, ON, Canada; 3 Cytognomix, Inc., London N6G 4X8, ON, Canada
Molecular Cytogenetics, 13 August 2015, 8:65
... Genomic locations of single copy probes were evaluated relative to locations of common CNVs ... samples have been tested with high resolution Affymetrix Cytoscan HD ... CNVs in cell line GM06326, initially characterized using Affymetrix Genome-Wide Human SNP Array 6.0 ...

191-
ROBO1 deletion as a novel germline alteration in breast and colorectal cancer patients

Rolando A. R. Villacis1, Francine B. Abreu1, Priscila M. Miranda1, Maria A. C. Domingues2, Dirce M. Carraro1, Erika M. M. Santos3, Victor P. Andrade4, Benedito M. Rossi3, Maria I. Achatz5, Silvia R. Rogatto1,6

1. Intl Research Center (CIPE), A.C. Camargo Cancer Center, Rua Taguá 440, São Paulo, CEP: 01508-010, SP, Brazil; 2. Dept of Pathology, Faculty of Medicine, University of São Paulo State (UNESP), Botucatu, SP, Brazil; 3. Oncology Center, Sírio-Libanês Hospital, São Paulo, SP, Brazil; 4. Dept of Pathology, A.C. Camargo Cancer Center, São Paulo, SP, Brazil; 5. Dept of Oncogenetics, A.C. Camargo Cancer Center, São Paulo, SP, Brazil; 6. Dept of Urology, Faculty of Medicine, University of São Paulo State (UNESP), CEP: 18618-970, Botucatu, SP, Brazil

Tumor Biology, first online 1 October 2015.

... identical germline deep intronic deletion of ROBO1 was identified in three index patients using ... (Agilent 4x180K and Affymetrix CytoScan HD). ... Transcriptomic analysis (HTA 2.0, Affymetrix) in two breast carcinomas from a single patient revealed ROBO1 down-expression ...

192-

Role of additional chromosomal changes in the prognostic value of t(4;14) and del(17p) in multiple myeloma: the IFM experience.

Hebraud B1, Magrangeas F2, Cleynen A1, Lauwers-Cances V3, Chretien ML4, Hulin C5, Leleu X6, Yon E3, Marit G7, Karlin L8, Roussel M9, Stoppa AM10, Belhadj K11, Voillat L12, Garderet L13, Macro M14, Caillot D4, Mohty M13, Facon T6, Moreau P15, Attal M9, Munshi N16, Corre J1, Minvielle S2, Avet-Loiseau H17.

1Laboratory for Genomics in Myeloma, Institut Universitaire du Cancer and University Hospital; and CRCT INSERM U1037, Toulouse, France; 2Unite Mixte de Genomique du Cancer, University Hospital; and INSERM U892, Nantes, France; 3Service d'Epidemiologie, University Hospital, Toulouse, France; 4Hematology Dept, Univ Hospital, Dijon, France; 5Hematology Department, Univ Hospital, Nancy, France; 6Hematology Dept, University Hospital, Lille, France; 7Hematology Dept, University Hospital, Bordeaux, France; 8Hematology Dept, University Hospital, Lyon, France; 9Hematology Dept, Univ Hospital, Toulouse, France; 10Hematology Dept, Institut Paoli-Calmettes, Marseille, France; 11Hematology Dept, Henri Mondor University Hospital, Creteil, France; 12Hematology Dept, General Hospital, Chalon sur Saone, France; 13Hematology Dept, Saint-Antoine University Hospital, Paris, France; 14Hematology Dept, University Hospital, Caen, France; 15Hematology Dept, Univ Hospital, Nantes, France; 16Lebow Inst of Myeloma Therapeutics and Jerome Lipper Multiple Myeloma Center, Dana-Farber Cancer Institute, Harvard Medical School, Boston, MA, USA; 17Laboratory for Genomics in Myeloma, Institut Universitaire du Cancer and University Hospital; and CRCT INSERM U1037, Toulouse, France; avet-loiseau.h@chu-toulouse.fr.

Blood, 2015; 125(13):2095-2100.

… DNA was extracted, labeled and hybridized on Affymetrix chips ... We used either the SNP 6.0 or the CytoScan chips, depending on the date of hybridization.

193-

Simultaneous Detection of Both Single Nucleotide Variations and Copy Number Alterations by Next-Generation Sequencing in Gorlin Syndrome

Kei-ichi Morita , Takuya Naruto, Kousuke Tanimoto, Chisato Yasukawa, Yu Oikawa, Kiyoshi Masuda, Issei Imoto, Johji Inazawa, Ken Omura, Hiroyuki Harada

PLoS ONE, 6 November 2015, 10(11): e0140480.

... CNA validation and detailed mapping of altered regions were performed using an Affymetrix CytoScan HD chromosome microarray platform ...

194- *Meeting abstract*

Single Cell Genotyping of Inv(16) AML in CBL Mutated Clonal Hematopoiesis Characterizes Clonal Architecture and Evolution of Exome Sequencing-Identified Mutations in the Protein Tyrosine Phosphatase Ptprt and Other Genes

Christoph Niemöller, Sabine Bleul, Nadja Blagitko-Dorfs, Christine Greil, Kenichi Yoshida, Rainer Claus, Dietmar Pfeifer, Seishi Ogawa, Michael Lübbert, and Heiko Becker

Blood, 3 Dec 2015; 126(23): Abstr #3834.

...WGA can lead to allele dropout (ADO), we also sequenced single nucleotide polymorphisms (SNPs), that were identified by CytoScan HD array (Affymetrix) to be heterozygous in the sample and that were located nearby the respective mutation loci.

195-

SIPA1L3 identified by linkage analysis and whole-exome sequencing as a novel gene for autosomal recessive congenital cataract.

Evers C1, Paramasivam N2, Hinderhofer K1, Fischer C1, Granzow M1, Schmidt-Bacher A3, Eils R4, Steinbeisser H1, Schlesner M2, Moog U1.

1Institute of Human Genetics, Heidelberg University, Heidelberg, Germany.; 2Div of Theoretical Bioinformatics, German Cancer Research Center (DKFZ), Heidelberg, Germany; 3Dept of Ophthalmology, St Vincentius-Kliniken gAG Karlsruhe, Karlsruhe, Germany; 41] Division of Theoretical Bioinformatics, German Cancer Research Center (DKFZ), Heidelberg, Germany [2] Dept for Bioinformatics and Functional Genomics, Institute for Pharmacy and Molecular Biotechnology (IPMB) and BioQuant, Heidelberg University, Heidelberg, Germany.

European Journal of Human Genetics, December 2015, 23(12):1627-1633.
… Array analysis/SNP genotyping Affymetrix CytoScan HD Oligo/SNP-Array was performed ...

196- *Meeting abstract*
SIRPB1: BIOMARKER OF RESPONSE TO 5-AZACITIDINE TREATMENT IN MDS AND AML PATIENTS
V. Guadagnuolo,1 M.C. Fontana,1 C. Papayannidis,1 I. Iacobucci,1 A. Padella,1 G. Simonetti,1 E. Imbrogno,1 S. Paolini,1 M. Abbenante,1 S. Parisi,1 C. Sartor,1 E. Ottaviani,1 A. Ferrari,1 M. Delledonne,2 M. Malagola,3 C. Filì ,3 D. Russo,3 M. Cavo,1 G. Martinelli1
1L. and A. Seràgnoli, Bologna University, Bologna; 2Dept of Biotechnology, Verona University, Verona; 3University of Brescia, AO Spedali Civili, Brescia, Italy
Haematologica, 2015, 100(Sup3) : Abstr #C106, p.18. [45° Congress of the Italian Society of Hematology
Florence, Italy, October 4-7, 2015
… To identify the genes mostly predictive of treatment response we use SNP arrays. Materials and Methods: SNPs array was performed by CytoScan HD Array (Affymetrix, Inc) …

197-
Small bowel malrotation in distal 15q duplication: evidence for a rare association.
McLaughlin, Brooke M.; Hufnagel, Robert B.; Saal, Howard M.
Clinical Dysmorphology, April 2015, 24(2): 65–67.
… SNP microarray analysis was performed using the hybridized Affymetrix Cytoscan HD GeneChip ...

198- *Meeting abstract*
SNP array reveals a new deletion of JAK2 in AML patients
Viviana Guadagnuolo1, Maria Chiara Fontana1, Antonella Padella1, Ilaria Iacobucci1, Cristina Papayannidis1, Giorgia Simonetti1, Anna Ferrari1, Giovanni Marconi1, Stefania Paolini1, Maria Chiara Abbenante1, Sarah Parisi1, Francesca Volpato1, Chiara Sartor1, Emanuela Ottaviani1, Massimo Delledonne2, Michele Cavo1, Guido Biasco3, and Giovanni Martinelli1
1L. & A. Seragnoli, Bologna, Italy; 2Dept of Biotechnology, Verona, Italy; 3Dept of Specialized, Experimental & Diagnostic Medicine, Bologna, Italy.
Cancer Research, 2015;75(15 Suppl):Abst #4848. [AACR 106th Annual Mtg, Apr 18-22, 2015, Philadelphia, PA]
Introduction: In Acute Myeloid Leukemia (AML) there is a strong need to develop new diagnostic and therapeutic options: to identify the genes mostly predictive of treatment response, we use Single Nucleotide Polymorphism (SNP) Arrays and Whole Exome Sequencing (WES) in AML patients with heterogeneous karyotypes and different subgroups. Materials and Methods: SNP arrays (CytoScan HD Array, Affymetrix Inc.) was positively done in 58 AML samples … Conclusion: By SNP arrays we have identified Copy Number Alterations involving important cancer genes AML and we showed that a new deletion in JAK2 may have a role in overall survival rate. ...

199-
Somatic copy number losses on chromosome 9q21.33q22.33 encompassing the PTCH1 loci associated with cardiac fibroma
Qianqian Zhang a, 1, Tongjian Wang a, 1, Dong Wang a, Jinxiu Liu a, Wenqian Yu a, Xiangju Liu b, Xiaoli Xiang c, Kai Dong a, Feng You a, Guichun Zhang a, Jifeng Ju a, Meng Zhu a, Wenyuan Duan a, , 2, Bin Qiao a, 2
a Institute of Cardiovascular Disease, General Hospital of Ji'nan Military Region, Ji'nan, China; b Genetics Diagnostic Lab, Tai'an Health Center for Women and Children, Tai'an, China; c Medical Information Institute of Sichuan Province, Chengdu, China
Cancer Genetics, December 2915, 208(12): 615–620.
... peripheral blood and tumor tissues of the patient were both investigated by Affymetrix CytoScan HD Chip for copy number analysis. It demonstrated copy number losses on chromosome 9q21.33q22.33 in cardiac fibroma tissue that didn't exist in peripheral blood ...

200-
Somatic loss of polycystic disease genes contributes to the formation of isolated and polycystic liver cysts
Manoe J Janssen1, Jody Salomon1, Wybrich R Cnossen1, Carsten Bergmann2,3, Rolph Pfundt4, Joost P H
1Dept of Gastroenterology & Hepatology, Radboudumc, Institute for Genetic and Metabolic Diseases, Nijmegen, The Netherlands; 2Dept of Bioscientia, Center for Human Genetics, Ingelheim, Germany; 3Renal Div, Dept of

Medicine, University Freiburg Medical Center, Freiburg, Germany; 4Dept of Human Genetics, Radboudumc, Institute for Genetic and Metabolic Diseases, Nijmegen, The Netherlands
Gut, 2015, 64:688-690.
…copy-number variations and loss of heterozygosity (LOH) regions through genome-wide high resolution cytogenetic array analysis (CytoScan HD, Affymetrix) and searched for intragenic mutations by sequencing.6 In the patients with autosomal dominant polycystic...

201-
Split Hand/Foot Malformation Associated with 7q21.3 Microdeletion: A Case Report
Sivasankaran A.a · Srikanth A.c · Kulshreshtha P.S.c · Anuradha D.b · Kadandale J.S.c · Samuel C.R.a
aDepartment of Genetics, Dr. ALM PG Institute of Basic Medical Sciences, University of Madras, and bDepartment of Medical Genetics, Institute of Obstetrics and Gynecology, Government Hospital for Women and Children, Madras Medical College, Chennai, and cCenter for Human Genetics, Biotech Park, Electronic City Phase I, Bangalore, India
Molecular Syndromology, 2015, 6:287-296.
... Whole-genome array-CGH analysis was performed using Affymetrix CytoScan® 750K microarray (Affymetrix Inc., Santa Clara, Calif., USA) consisting of 750,000 oligonucleotide probes across the genome (including 550K unique non-polymorphic probes and 200K SNP probes ...

202-
Streamlining the OncoScan® Array Procedure for Use in a Clinical Laboratory.
Paxton CN1, Rowe LR1, South ST1.
1ARUP Institute for Clinical and Experimental Pathology®, Salt Lake City, UT.
J Assoc Genet Technol. 2015;41(2):61-5.
... The objective of this study was to identify steps in the OncoScan procedure that could be modified to make the process more efficient and technician-friendly in the clinical laboratory setting. ... an adjustment to the overnight hybridization temperature to allow for simultaneous hybridization of OncoScan and CytoScan® arrays. ...
Related abstract by the same authors, Validation of a Modified OncoScan Protocol for Use in a Clinical Laboratory, Cancer Genetics, June 2015, 208(6): 361.

203-
Susceptibility allele-specific loss of miR-1324-mediated silencing of the INO80B chromatin-assembly complex gene in pre-eclampsia
Cees B.M. Oudejans1,4,*, Omar J. Michel1, Rob Janssen2,4, Rob Habets1, Ankie Poutsma1, Erik A. Sistermans3, Marjan M. Weiss3, Danny Incarnato5,6, Salvatore Oliviero5,6, Gunilla Kleiverda7, Marie Van Dijk1,4 and Reynir Arngrímsson8
1Dept of Clinical Chemistry, 2Dept of Physiology, 3Dept of Clinical Genetics and 4Institute for Cardiovascular Research, VU University Medical Center, De Boelelaan 1117, 1081 HV Amsterdam, The Netherlands, 5Human Genetics Foundation (HuGeF), Via Nizza 52, 10126 Torino, Italy, 6Dipartimento di Biotecnologie, Chimica e Farmacia, Università degli Studi di Siena, Via Fiorentina 1, 53100 Siena, Italy, 7Depa of Gynecology, Flevo Hospital, Hospitaalweg 1, 1315 RA Almere, The Netherlands and 8Biomedical Center, University of Iceland, Vatnsmýrarvegi 16, 101 Reykjavík, Iceland
Human Molecular Genetics, 2015, 24(1): 118-127.
… The array used, CytoScan HD, has greater than 99% sensitivity and can reliably detect ...

204-
Syndromic craniosynostosis associated with microdeletion of chromosome 19p13.12–19p13.2
Sarah M. Lyon a, Darrel Waggoner b, Sara Halbach b, Erik C. Thorland c, Leila Khorasani d, Russell R. Reid d
a Pritzker School of Medicine, University of Chicago, IL; b Dept of Human Genetics and Pediatrics, University of Chicago, 5841 S. Maryland Ave, M/C 0077, Chicago, IL 60637; c Lab Medicine & Pathology, 200 First St SW, Hilton 970, Rochester, MN 55905-0001; d Dept of Surgery, University of Chicago, 5841 S. Maryland Ave, M/C 0077, Chicago, IL 60637
Genes & Diseases, December 2015, 2(4): 347–352.
... Cytogenomic array analysis was performed using either a custom Agilent oligonucleotide microarray ... analyzed on human genome build hg18 or the Affymetrix CytoScan HD array including 750,000 SNP probes and 1.9 million non-polymorphic probes analyzed on hg19.

205-
Syndromic X-linked intellectual disability segregating with a missense variant in RLIM

Elin Tønne1, Rita Holdhus2, Christine Stansberg2, Asbjørg Stray-Pedersen1, Kjell Petersen3, Han G Brunner4, Christian Gilissen2,4, Alexander Hoischen2,4, Trine Prescott1, Vidar M Steen2,5 and Torunn Fiskerstrand2,5

1Dept of Medical Genetics, Oslo University Hospital, Oslo, Norway; 2Dept of Clinical Science, University of Bergen, Bergen, Norway; 3Computational Biology Unit, University of Bergen, Bergen, Norway; 4Dept of Human Genetics, Radboud University Medical Center and Radboud Institute for Molecular Life Sciences (RIMLS), Nijmegen, The Netherlands; 5Center for Medical Genetics and Molecular Medicine, Haukeland University Hospital, Bergen, Norway

European Journal of Human Genetics, 2015, 23: 1652–1656.

... Affymetrix Cytoscan HD containing 2.7 million oligonucleotides was performed ...

206- *Meeting abstract*

Targeted Detection of Copy Number Variants Using a Myeloid Malignancy Next Generation Sequencing Mutation Panel Allows Comprehensive Genetic Analysis Using a Single Testing Method

Wei Shen, PhD*,1, Philippe Szankasi, PhD2, Maria Sederberg, MS*,2, Jonathan Schumacher, MS*,2, Kimberly Frizzell, PhD*,1, Elaine P Gee, PhD*,3, Jay L Patel, MD*,4,2, Sarah T. South, PhD*,4,5, Xinjie Xu, PhD*,4,2, and Todd W Kelley, MD4,2

1Genomics, ARUP Laboratories, Salt Lake City, UT; 2ARUP Institute for Clinical and Experimental Pathology®, Salt Lake City, UT; 3Clinical Laboratory Informatics and Integration, ARUP Laboratories, Salt Lake City, UT; 4Dept of Pathology, University of Utah, Salt Lake City, UT; 5ARUP Institute for Clinical and Experimental Pathology, Salt Lake City, UT

Blood, 3 December 2015; 126: 2887.

... comprehensive genetic profiling of myeloid malignancies requires multiple testing strategies at high cost. In an effort to provide more efficient genetic profiling of these disorders, we designed and tested an algorithm to evaluate for CNVs using sequence coverage data derived from a NGS-based 53-gene myeloid mutation panel … CNVs were detected using a circular binary segmentation algorithm. In a subset of cases (n=43) CNVs detected using NGS data were validated by comparing to the results obtained by SNP microarray (CytoScan HD Array, Affymetrix) testing, the current gold standard, and analyzed by CHAS 2.0 (Affymetrix) and Nexus 7.5 (Biodiscovery). ...

207-

Thalassemia Intermedia Caused by 16p13.3 Sectional Duplication in a β-Thalassemia Heterozygous Child

Sha Liu, Hua Jiang, Man-Yu Wu, Yong-Ling Zhang, and Dong-Zhi Li

Dept of Hematology/Oncology, Guangzhou Women & Children Medical Center affiliated to Guangzhou Medical University, Guangzhou, Guangdong, China; Prenatal Diagnostic Center, Guangzhou Women & Children Medical Center affiliated to Guangzhou Medical University, Guangzhou, Guangdong, China

Pediatric Hematology-Oncology, 2015, 32(5): 349-353.

... Sample preparation, array hybridization, and analysis of the results were performed according to the manufacturer's instructions (Affymetrix CytoScan High-Density Array; ...

208- *Meeting abstract*

THE CODING GENOME OF NODAL MARGINAL ZONE LYMPHOMA REVEALS RECURRENT
MOLECULAR ALTERATIONS OF PTPRD AND OTHER JAK/STAT SIGNALING GENES

V. Spina,1 A. Bruscaggin,1 H. Khiabanian,2 L. Martuscelli,1 E. Spaccarotella,1 M. Messina,3 S. Monti,1 A.B. Holmes,4 S. Chiaretti,3 L. Arcaini,5 M. Lucioni,6 F. Tabbò,7 S. Rasi,1 S. Zairis,2 A. Ramponi,8 R. Marasca,9 M. Ponzoni,10 E. Tiacci,11 B. Falini,11 F. Bertoni,12 L. Pasqualucci,4 R. Foà,3 G. Inghirami,7 R. Rabadan,2 G. Gaidano,1 D. Rossi1

1Div of Hematology, Dept of Translational Medicine, Amedeo Avogadro University of Eastern Piedmont, Novara, Italy; 2Dept of Biomedical Informatics and Center for Computational Biology and Bioinformatics Columbia University, New York, USA; 3Div of Hematology, Dept of Cellular Biotechnologies and Hematology, Sapienza University, Roma, Italy; 4Institute for Cancer Genetics and the Herbert Irving Comprehensive Cancer Center, Columbia University, New York; 5Dept of Hematology-Oncology, Fondazione IRCCS Policlinico San Matteo and Dept of Molecular Medicine, University of Pavia, Pavia, Italy; 6Division of Pathology, Fondazione IRCCS Policlinico San Matteo, University of Pavia, Pavia, Italy; 7Dept of Pathology and Laboratory Medicine, Weill Cornell Medical College, New York; 8Div of Pathology, Dept of Health Science, Amedeo Avogadro University of Eastern Piedmont, Novara, Italy; 9Div of Hematology, Dept of Oncology and Hematology, University of Modena and Reggio Emilia, Modena, Italy; 10Unit of Lymphoid Malignancies, San Raffaele H. Scientific Institute, Milan, Italy; 11Institute of Hematology, Ospedale S. Maria della Misericordia, Univ of Perugia, Perugia, Italy; 12Lymphoma and Genomics Research Program, IOR Institute of Oncology Research and Oncology Institute of Southern Switzerland, Bellinzona, Switzerland

Haematologica, 2015, 100(Sup3) : Abstr #C028, p.18. [45° Congress of the Italian Society of Hematology
Florence, Italy, October 4-7, 2015

Nodal marginal zone lymphoma (NMZL) is one of the few B-cell tumors still remaining orphan of cancer gene lesions. Here we aim at disclosing the pathways that are molecularly deregulated in this lymphoma. The study was based on 35 NMZL with a diagnosis confirmed by pathological revision and

lack of clinico-radiological evidence of extranodal or splenic disease. WES (HiSeq 2500, Illumina) and high density SNP array (Cytoscan HD, Affymetrix) of tumor/normal DNA pairs from 18 discovery NMZL …

209- *Meeting abstract*

THE CLINICAL UTILITY OF SNP ARRAY ANALYSIS IN MYELODYSPLASTIC SYNDROME PATIENTS: RESULTS FROM A UK DIAGNOSTIC GENETIC LABORATORY USING THE AFFYMETRIX CYTOSCAN HD 2.6M SNP ARRAY

N. Trim, E. Huxley, M. Griffiths, S. Jeffries

1West Midlands Regional Genetic Laboratory, Birmingham Women's Hopsital, Birmingham, United Kingdom

Leukemia Research, April 2015, 39(Sup 1):S80-S81, Abst #157. [13th Intl Symp Myelodysplastic Syndromes]

… The aim of the study is: to establish the benefits of using SNP arrays in a haematooncology setting; to determine the utility of SNP array as the primary test for MDS patients to replace metaphase chromosomes; to determine whether blood may be used instead of marrow; and to collate the additional genomic/genetic abnormalities that are detected. Analysis is being performed using the Affymetrix Cytoscan HD SNP array containing 2.6 million markers for copy number analysis including approximately 750,000 SNPs. We present some of the preliminary findings from the first 100 samples including abnormality rates, failure rates and types of abnormalities detected with focus on individual cases to demonstrate the clinical utility of SNP arrays above that of metaphase chromosome analysis.

210-

The future of laboratory medicine — A 2014 perspective

Larry J. Kricka a, Tracey G. Polsky a, b, Jason Y. Park c, Paolo Fortina d, e

aDept of Pathology and Laboratory Medicine, University of Pennsylvania School of Medicine, 7.103 Founders Pavilion, 3400 Spruce Street, Philadelphia, PA 19104; bDept of Pathology and Laboratory Medicine, Children's Hospital of Philadelphia, Philadelphia, PA 19104; cDept of Pathology and the Eugene McDermott Center for Human Growth and Development, Univ of Texas Southwestern Medical Center, Children's Medical Center, 1935 Medical District Dr, Dallas, TX 75235; dCancer Genomics Laboratory, Kimmel Cancer Center, Dept of Cancer Biology, Sidney Kimmel Medical College, Thomas Jefferson Univ, Philadelphia, PA; eDept of Molecular Med, Universita' La Sapienza, Rome, Italy

Clinica Chimica Acta, 1 January 2015, 438: 284–303. doi:10.1016/j.cca.2014.09.005

… This review surveys the many predictions, beginning in 1887, about the future of laboratory medicine and its sub-specialties such as clinical chemistry and molecular pathology. It provides a commentary on the accuracy of the predictions and offers opinions on emerging technologies, economic factors and social developments that may play a role in shaping the future of laboratory medicine. … In 2014, the FDA cleared the Cytoscan HD high density SNP array (Affymetrix) for use in the diagnosis of developmental delay, intellectual disability and congenital anomalies [141].

211-

The genomic landscape of phaeochromocytoma

Aidan Flynn, Diana Benn, Roderick Clifton-Bligh, Bruce Robinson, Alison H Trainer, Paul James, Annette Hogg, Kelly Waldeck, Joshy George, Jason Li, Stephen B Fox, Anthony J Gill, Grant McArthur, Rodney J Hicks, Richard W Tothill

The Journal of Pathology, May 2015, 236(1): 78–89.

Phaeochromocytomas (PCC) and paragangliomas (PGL) are rare neural crest derived tumours originating from adrenal chomaffin cells or extra-adrenal sympathetic and parasympathetic tissues. ... To explore the genomic landscape of PCC/PGL we applied exome-sequencing, high-density SNP-array analysis and RNA-sequencing to 36 PCC and four functional PGL tumours. ... Affymetrix Cytoscan HD array data is made available through NCBI GEO (http://www.ncbi.nlm.nih.gov/geo/, GEO Accession ID: GSE61594 ...

212-

The Human Glioblastoma Cell Culture Resource: Validated Cell Models Representing All Molecular Subtypes

Yuan Xie a, 1, Tobias Bergström a, 1, Yiwen Jiang a, 1, Patrik Johansson a, 1, Voichita Dana Marinescu a, Nanna Lindberg b, Anna Segerman a, Grzegorz Wicher a, Mia Niklasson a, Sathishkumar Baskaran a, Smitha Sreedharan a, Isabelle Everlien a, Marianne Kastemar a, Annika Hermansson a, Lioudmila Elfineh a, Sylwia Libard a, Eric Charles Holland b, Göran Hesselager c, Irina Alafuzoff a, Bengt Westermark a, 2, Sven Nelander a, 2, Karin Forsberg-Nilsson a, 2, Lene Uhrbom a, 2

a Dept of Immunology, Genetics and Pathology, Science for Life Laboratory, Rudbeck Laboratory, Uppsala University, 751 85 Uppsala, Sweden; b Fred Hutchinson Cancer Research Center, 1100 Fairview Ave N., PO Box

19024, Seattle, WA 98109; c Dept of Neuroscience, Uppsala University, Uppsala University Hospital, SE-751 85 Uppsala, Sweden
EbioMedicine, October 2015, 2(10): 1351–1363.
... Total RNA extracted from 48 GC lines ... labeled and hybridized on Affymetrix GeneChip Human Exon 1.0 ST arrays. ... copy number aberration in all of our 48 GC lines employing Affymetrix Cytoscan HD arrays ... RNA was prepared from cells and tumor tissue and analyzed on the Affymetrix HTA 2.0 platform.

213-
The phenotype spectrum of X-linked ichthyosis identified by chromosomal microarray
Jennifer L. Hand, MDa, b, c, Cassandra K. Runke, CGCd, Jennelle C. Hodge, PhDb, d, e
a Dept of Dermatology, Mayo Clinic, Rochester, MN; b Dept of Medical Genetics, Mayo Clinic, Rochester, MN; c Dept of Pediatric and Adolescent Medicine, Mayo Clinic, Rochester, MN; d Dept of Laboratory Medicine and Pathology, Mayo Clinic, Rochester, MN; e Dept of Pathology and Laboratory Medicine, Cedars-Sinai Medical Center, Los Angeles, CA
Dermatology, April 2015, 72(4): 617–627.
... submitted to the Mayo Clinic Cytogenetics Laboratory from March 4, 2008 to September 3, 2013 for CMA analysis using either a 4x44K or 4x180K oligonucleotide–based whole-genome cytogenomic microarray (Agilent Technologies, ... or Cytoscan HD array ...

214-
The Protein Tyrosine Phosphatase Rptpζ Suppresses Osteosarcoma Development in Trp53-Heterozygous Mice
Christina Baldauf , Anke Jeschke , Vincent Kanbach, Philip Catala-Lehnen, Daniel Baumhoer, Helwe Gerull, Sophia Buhs, Michael Amling, Peter Nollau, Sheila Harroch, Thorsten Schinke
PLoS ONE, 11 Sept 2015, 10(9): e0137745.
... We additionally isolated RNA from CD11b-negative osteoblast cultures at day 5 and day 12 of differentiation to perform genome-wide expression analysis using Affymetrix Gene Chips (Affymetrix MG 430 2.0). ... To address the question, if Rptpζ would also act as a tumor suppressor in human OS, we first analyzed copy number data sets (CytoScan Arrays) from 160 human OS samples for chromosomal alterations around the PTPRZ1 locus on chromosome 7q31.3, yet we did not detect any specific changes supporting a contribution of PTPRZ1 loss to OS development or severity.

215-
The role of candidate-gene CNTNAP2 in childhood apraxia of speech and specific language impairment
TM Centanni1,2, JN Sanmann3, JR Green1, J Iuzzini-Seigel1,4, C Bartlett5, WG Sanger3,† TP Hogan1,*
1MGH Institute of Health Professions, Boston, MA; 2Massachusetts Institute of Technology, Cambridge, MA; 3University of Nebraska Medical Center, Nebraska Medical Center, Omaha, NE; 4Marquette University, Milwaukee, MI; 5The Ohio State University, Columbus, OH
American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, October 2015,168(7): 536–543.
... High-resolution genome-wide analysis was performed on genomic DNA using the CytoScanHD™ array (Affymetrix, Santa Clara, CA) according to manufacturer's instruction. **...**

216-
The senescent microenvironment promotes the emergence of heterogeneous cancer stem-like cells
Luis Jaime Castro-Vega1,2,*, Karina Jouravleva1,2,*, Paola Ortiz-Montero3, Win-Yan Liu1,2, Jorge Luis Galeano3, Martha Romero4, Tatiana Popova5, Silvia Bacchetti6, Jean Paul Vernot3, Arturo Londoño-Vallejo1,2
1UMR3244, Telomeres and Cancer Laboratory, “Labellisé Ligue”, Institut Curie, Paris 75248, France; 2UPMC University - Paris 06, Paris 75005, France; 3Cellular and Molecular Physiology Group, Instituto de Investigaciones Biomédicas, Faculty of Medicine, Universidad Nacional de Colombia, Bogotá D.C, Colombia; 4Dept of Pathology, Hospital Universitario-Fundación Santa Fe de Bogotá, Bogotá, Colombia
; 5U830 Genetics and Cancer Biology, Institut Curie, Paris 75248, France; 6Dept of Experimental Oncology, Istituto Regina Elena, Rome 00158, Italy
Carcinogenesis, 2015, 36(10): 1180-1192.
... Here we show that post-crisis cells are not able to form tumors unless a senescent microenvironment is provided. ... Affymetrix Cytoscan HD arrays were hybridized with labeled DNA from PC1, PC2 and the four explanted cell lines.

217-
TP53 mutation analysis in chronic lymphocytic leukemia: comparison of different detection methods.
Kantorova B, Malcikova J, Smardova J, Pavlova S, Trbusek M, Tom N, Plevova K, Tichy B, Truong S, Diviskova E, Kotaskova J, Oltova A, Patten N, Brychtova Y, Doubek M, Mayer J, Pospisilova S.
Tumour Biol. 2015 May;36(5):3371-80.
... In 15 of the TP53-mutated patients, TP53 gene abnormalities or copy-neutral loss of heterozygozity (cn-LOH) were analyzed using Cytogenetics Whole-Genome 2.7 M Array (n=9; Affymetrix) and CytoScan® High Density Array (n=6; Affymetrix).

218-
Typical and Atypical Associated Findings in a Group of 346 Patients with Mayer-Rokitansky-Kuester-Hauser Syndrome
Katharina Rall, MD1, Simone Eisenbeis, PhD1, Verena Henninger, MD1, Melanie Henes, MD1, Diethelm Wallwiener, MD1, Michael Bonin, PhD2, Sara Brucker, MD1
1 University Hospital Tuebingen, Dept of Obstetrics and Gynecology, Tuebingen, Germany; 2 University Hospital Tuebingen, Dept of Medical Genetics, Microarray Facility, Tuebingen, Germany
Journal of Pediatric and Adolescent Gynecology, October 2015, 28(5): 362–368.
... The Mayer-Rokitansky-Kuester-Hauser (MRKH) syndrome is characterized by vaginal and uterine aplasia in a 46,XX individual. ... analysis using Human Mapping CytoScan HD Array (Affymetrix, Santa Clara, CA) detected 4 times euchromatin from the region 22q11.1-22q11.21.

219-
Understanding the role of hyperdiploidy in myeloma prognosis: which trisomies really matter?
Marie-Lorraine Chretien1,*, Jill Corre2,*, Valerie Lauwers-Cances3, Florence Magrangeas4, Alice Cleynen2, Edwige Yon3, Cyrille Hulin5, Xavier Leleu6, Frederique Orsini-Piocelle7, Jean-Sebastien Blade8, Claudine Sohn9, Lionel Karlin10, Xavier Delbrel11, Benjamin Hebraud12, Murielle Roussel12, Gerald Marit13, Laurent Garderet14, Mohamad Mohty14, Philippe Rodon15, Laurent Voillat16, Bruno Royer17, Arnaud Jaccard18, Karim Belhadj19, Jean Fontan20, Denis Caillot1, Anne-Marie Stoppa21, Michel Attal12, Thierry Facon6, Philippe Moreau22, Stephane Minvielle4, and Hervé Avet-Loiseau2
1Dept of Hematology, Centre Hospitalier Universitaire, Dijon, France; 2Unit for Genomics in Myeloma, Institut Universitaire du Cancer-Oncopole, and Centre de Recherches sur le Cancer de Toulouse INSERM 1037, Toulouse, France; 3Service d'Epidemiologie, Centre Hospitalier Universitaire, Toulouse, France; 4Unité Mixte de Génomique du Cancer, Centre Hospitalier Universitaire, and INSERM U892, Nantes, France; 5Dept of Hematology, Centre Hospitalier Universitaire, Nancy, France; 6Dept of Hematology, Centre Hospitalier Universitaire, Lille, France; 7Dept of Hematology, Centre Hospitalier, Annecy, France; 8Dept of Hematology, Centre Hospitalier Universitaire, Nancy, France; 9Dept of Hematology, Hopital Inter-Armées, Toulon, France; 10Dept of Hematology, Centre Hospitalier, Toulon, France; 11Dept of Hematology, Centre Hospitalier, Pau, France; 12Dept of Hematology, Institut Universitaire du Cancer, Toulouse, France; 13Dept of Hematology, Centre Hospitalier Universitaire, Bordeaux, France; 14Dept of Hematology, Centre Hospitalier Universitaire St-Antoine, Paris, France; 15Dept of Hematology, Centre Hospitalier, Perigueux, France; 16Dept of Hematology, Centre Hospitalier, Chalon sur Saone, France; 17Dept of Hematology, Centre Hospitalier Universitaire, Amiens, France; 18Dept of Hematology, Centre Hospitalier Universitaire, Limoges, France; 19Dept of Hematology, Centre Hospitalier Universitaire, Créteil, France; 20Dept of Hematology, Centre Hospitalier Universitaire, Besançon, France; 21Dept of Hematology, Institut Paoli Calmettes, Marseille, France; 22Dept of Hematology, Centre Hospitalier Universitaire, Nantes, France
Blood, 17 December 2015, 126(25): 2713 – 2719.
... For the first time ... we show that not all trisomies display the same prognostic impact. ... We analyzed by SNP array (Affymetrix, ... using either the SNP6.0 array or the Cytoscan array ...

220-
Uniparental Trisomy of a Mutated HRAS Proto-Oncogene in Embryonal Rhabdomyosarcoma of a Patient With Costello Syndrome
Jan Menke, Silke Pauli, Matthias Sigler, Ingrid Kühnle, Moneef Shoukier, Barbara Zoll, Christina Ganster, Gabriela Salinas-Riester and Inga-Marie Schaefer
University Medical Center Goettingen, Goettingen, Germany
Journal of Clinical Oncology, May 1, 2015, 33(13):e62-e65.
... In the tumor, fluorescent in situ hybridization and single nucleotide polymorphism (SNP) microarray analysis (CytoScan HD; Affymetrix, Santa Clara, CA) indicated trisomy of chromosome 11.

221- *Meeting abstract*

Unique pattern of copy number changes including chromothripsis in pulmonary adenocarcinoma with EML4-ALK fusion

Hironori Ninomiya1, Motohiro Kato2, Seishi Ogawa2, Noriko Motoi1, Kengo Takeuchi1, Tatsushi Kodama3, Hiroshi Sakamoto3, Nobuya Ishii3, Mutsunori Fujiwara4, and

1Japanese Foundation for Cancer Research, Tokyo, Japan; 2Cancer Genomics Project, The University of Tokyo, Tokyo, Japan; 3Chugai Pharmaceutical Co., Ltd., Kamakura, Japan; 4Japanese Red Cross Medical Center, Tokyo, Japan.

Cancer Research, 2015;75(15 Suppl):Abst #3853. [AACR 106th Annual Mtg, Apr 18-22, 2015, Philadelphia, PA]

… To elucidate the underlying mechanisms resulting in gene fusion, two cancer cell lines positive for EML4-ALK fusion and surgically resected frozen specimens (33 ALK fusion-positive and 95 ALK fusion-negative adenocarcinomas) were analyzed using Affymetrix CytoScan HD Array with Chromosome Analysis Suite (ChAS) software for cell lines (JFCR-LC649 and JFCR-LC654) and Affymetrix GeneChip Mapping 250K arrays for surgical specimens. ...

222-

Unusual Genetic Aberrations Including a Deletion of KLF6 Tumor Suppressor Gene Revealed by Integrated Cytogenetic Approaches in a Pediatric Ewing Sarcoma

White, Jason C. DO1*; Halligan, Gregory E. MD*; de Chadarévian, Jean-Pierre MD†; Pascasio, Judy M. MD†; Punnet, Hope H. PhD†; Liu, Jinglan PhD†

1*Dept of Pediatrics, Section of Oncology †Dept of Pathology and Laboratory Medicine, Drexel University College of Medicine and St. Christopher's Hospital for Children, Philadelphia, PA.

Journal of Pediatric Hematology/Oncology, Jan 2015, 37(1):e51–e54.

… SNP microarray assay using Affymetrix CytoScan HD SNP array on the frozen tumor specimen showed an interstitial hemizygous deletion from nucleotide 3,032,920 to 6,609,423 (NCBI build 37.1/hg19) on the short arm of chromosome 10 at 10p15.2p14, spanning ...

223-

Unusual Presentation of Pelizaeus-Merzbacher Disease: Female Patient with Deletion of the Proteolipid Protein 1 Gene

Teva Brender, Donna Wallerstein, John Sum, and Robert Wallerstein

Silicon Valley Genetics Center, Santa Clara Valley Medical Center, San Jose, CA 95128, USA

Case Reports in Genetics, accepted 10 Feb 2015, 2015:Article #453105.

… Genetic testing performed by chromosome microarray using the Affymetrix Cytoscan HD platform with 743,000 SNP probes showed a 712KB interstitial deletion on the X chromosome at position Xq22.2.

224-

Use of Affymetrix arrays in the diagnosis of gene copy-number variation

Farah R. Zahir and Marco A. Marra

1Canada's Michael Smith Genome Sciences Centre, BC Cancer Agency, Vancouver, British Columbia, Canada; 2Univ of British Columbia, Dept of Medical Genetics, Vancouver, British Columbia, Canada

Current Protocols in Human Genetics, Unit 8.13, online 1 April 2015.

... Here we discuss the application of the CytoScan high-density (HD) platform for the detection of genomic imbalance. We provide an overview of the sequence of computational analyses involved in identifying pathogenic CNVs and highlight important parameters for consideration in assessing the pathogenicity of a detected CNV.

225-

Using extended pedigrees to identify novel autism spectrum disorder (ASD) candidate genes

Marc Woodbury-Smith1,2, Andrew D. Paterson2,6, Bhooma Thiruvahindrapduram2, Anath C. Lionel2, Christian R. Marshall2, Daniele Merico2, Bridget A. Fernandez3, Eric Duku1, James S. Sutcliffe8, Irene O'Conner1, Christina Chrysler1, Ann Thompson1, Barbara Kellam2, Kristiina Tammimies2, Susan Walker2, Ryan K. C. Yuen2, Mohammed Uddin2, Jennifer L. Howe2, Morgan Parlier5, Kathy Whitten3, Peter Szatmari7, Veronica J. Vieland4, Joseph Piven5, Stephen W. Scherer2

1. Dept of Psychiatry and Behavioural Neurosciences, McMaster University, Chedoke Campus (Evel Building, Rm 457), Hamilton, ON, Canada; 2. Program in Genetics and Genome Biology, The Centre for Applied Genomics, The Hospital for Sick Children, Toronto, ON, Canada; 3. Provincial Medical Genetics Program, Memorial Hospital, St John's, Newfoundland, Canada; 4. Battelle Center for Mathematical Medicine, Nationwide Children's Hospital, Columbus, OH, USA; 5. Carolina Institute for Developmental Disabilities, University of North Carolina, Wilmington, NC; 6. Dalla Lana School of Public Health, University of Toronto, Toronto, ON, Canada; 7. Centre for Addiction and Mental Health, The Hospital for Sick Children and University of Toronto, Toronto, ON, Canada; 8. Vanderbilt University Medical Center, Vanderbilt University, Nashville, TN

Human Genetics, February 2015, 134(2):191-201

... additional controls included 4,768 individuals from the SAGE and HABC datasets typed on Illumina1 M, 2,357 OHI and POPGEN controls typed on the Affymetrix SNP 6.0 platform, and 863 OPGP controls typed on Affymetrix CytoscanHD.

226- *Meeting abstract*

Very poor outcome and chemoresistance of acute myeloid leukemia patients with TP53 mutations: Correlation with complex karyotype and clinical outcome.

Cristina Papayannidis, Anna Ferrari, Stefania Paolini, Carmen Baldazzi, Chiara Sartor, Maria Chiara Abbenante, Sarah Parisi, Francesca Volpato, Ilaria Iacobucci, Antonella Padella, Viviana Guadagnuolo, Margherita Perricone, Valentina Robustelli, Claudia Venturi, Giorgia Simonetti, Elisa Zuffa, Eugenia Franchini, Emanuela Ottaviani, Nicoletta Testoni, and Giovanni Martinelli

"Seràgnoli" Institute of Hematology, Bologna University School of Medicine, Bologna, Italy.

Clinical Cancer Research, Sep 2015; 21: Abstract #B03. [AACR Special Conference on Hematologic Malignancies: Translating Discoveries to Novel Therapies; September 20-23, 2014; Philadelphia, PA]

... we focused on a subgroup of 172/886 AML pts treated at the Seràgnoli Institute of Bologna between 2002 and 2013. ... 40 samples were genotyped with Genome-Wide Human SNP 6.0 arrays or with CytoScan HD Array (Affymetrix) and analysed by Nexus Copy Number™ v7.5 (BioDiscovery). ... Copy Number Alterations (CNAs) analysis performed on 40 cases by Affymetrix SNP arrays showed the presence of several CNAs in all cases ... We recommend TP53 mutation screening at least in AML pts carrying either complex karyotype or chr. 8 gain.

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When Cri du chat syndrome meets Edwards syndrome

Yingjun Xie, Yi Zhou, Jianzhu Wu, Yunxia Sun, Yongzhen Chen, Baojiang Chen

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Molecular Medicine Reports, 15 March 2015, 11(3): 1933-1938.
doi:10.3892/mmr.2014.2920

… CEL files obtained by scanning CytoScan arrays were analyzed with the Chromosome Analysis Suite software (Affymetrix), employing genome annotation data (version GRCH37, hg19).

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Whole-exome sequencing of breast cancer, malignant peripheral nerve sheath tumor and neurofibroma from a patient with neurofibromatosis type 1

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Cancer Medicine, December, 4(12): 1871–1878,.

... For higher resolution, the blood, dermal neurofibroma and primary breast tumor samples were assayed using the Affymetrix CytoScan HD platform ...

229-

Whole-genome sequencing of quartet families with autism spectrum disorder

Ryan K C Yuen,Bhooma Thiruvahindrapuram,Daniele Merico,Susan Walker,Kristiina Tammimies,Ny Hoang,Christina Chrysler,Thomas Nalpathamkalam,Giovanna Pellecchia,Yi Liu,Matthew J Gazzellone,Lia D'Abate,Eric Deneault,Jennifer L Howe,Richard S C Liu,Ann Thompson,Mehdi Zarrei,Mohammed Uddin,Christian R Marshall,Robert H Ring,Lonnie

Zwaigenbaum,Peter N Ray,Rosanna Weksberg,Melissa T Carter,Bridget A Fernandez,Wendy Roberts,Peter Szatmari, Stephen W Scherer
Nature Medicine, 2015, 21: 185–191.
… 40 of the 85 quartet families were run in parallel using the CytoScan HD Array (Affymetrix, Santa Clara, CA).

230-
Wiskott-Aldrich Syndrome in a Girl Caused by Heterozygous WASP Mutation and Extremely Skewed X-Chromosome Inactivation: A Novel Association with Maternal Uniparental Isodisomy 6
Takimoto T.a· Takada H.b· Ishimura M.a· Kirino M.a· Hata K.d· Ohara O.c· Morio T.e· Hara T.a
aDepts of Pediatrics and bPerinatal and Pediatric Medicine, Graduate School of Medical Sciences, Kyushu University, Fukuoka, cDept of Human Genome Technology, Kazusa DNA Research Institute, Kisarazu, dDept of Maternal-Fetal Biology, National Research Institute for Child Health and Development, eDept of Pediatrics and Developmental Biology, Graduate School of Medicine, Tokyo Medical and Dental University, Tokyo, Japan
Neonatology, 2015, 107: 185-190.
… SNP microarray analysis was performed using the Affymetrix Cytoscan HD platform [17] which uses over 743,000 SNP probes and 1,953,000 NPCN probes with a median spacing of 0.88 kb.

231- *Meeting abstract*
Y Chromosome Microdeletion Detection by Cytoscan HD Microarray Platform
Maria C. Sederberg, Leslie R. Rowe, Allen N. Lamb
ARUP Laboratories, Department of Pathology, University of Utah, Salt Lake City, UT, USA
Cancer Genetics, June 2015, 208(6): 359. Available online 22 July 2015
... Many laboratories do not report Y microdeletions in studies of patients with DD/ID, autism, or dysmorphism. It is important to understand how well the Affymetrix Cytoscan HD detects AZF deletions, to recognize when mosaicism exist, and when a rearranged Y is present. ...