

The American Society of Human Genetics
57th Annual Meeting

Program Addendum

• **Special Notice** •

Due to security reasons and overcrowding in session rooms in previous years, **badges must be worn and be visible at all times**. They will be checked at all scientific sessions, including the Wednesday morning Social Issues and Education Sessions. Individuals who have not registered or who have lost or forgotten their badges will not be admitted to sessions, exhibits or poster areas. Registrants who lose badges may obtain a replacement badge for a charge of \$5.

• **Sponsor Acknowledgement** •

The American Society of Human Genetics gratefully acknowledges the following organizations for their generous support of its 57th annual meeting.

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• **Program Updates** •

• **TUESDAY, October 23**

Ancillary Events Added:

1:00 PM-4:00 PM **A-T and DNA Repair Disorders**, San Diego Marriott Hotel and Marina, Cardiff Room

• **WEDNESDAY, October 24**

Ancillary Events Added:

11:30 AM-1:00 PM **Applied Biosystems Workshop: The SOLiD System: a Technology Enabling New Applications**, Hilton San Diego Gas Lamp Quarter, Santa Rosa Room

6:30 PM-8:30 PM **Cleveland Clinic Genomic Medicine Institute Reception**, San Diego Marriott Hotel and Marina, Coronado Room (4th Floor of the South Tower)

6:30 PM-8:30 PM **Early Intervention for LSDs: What is Appropriate Care and Management of the Pre-Symptomatic Individual? "A Collaborative Discussion,"** San Diego Convention Center, Room 33 (Upper Level)

9:00 PM-12:00 midnight **UCSF-Stanford Medical & Human Genetics Reception**, San Diego Marriott Hotel and Marina, Torrance Room (4th Floor of the South Tower)

Posters Changed:

324/W Detection of Philadelphia chromosome in patients with chronic myeloid leukemia (CML) and acute lymphoblastic leukemia (ALL) by conventional and molecular techniques.

Fifth author added: F. Cuellar-Ambrosi, San Vicente de Paul Hospital, Cancer Unit, Antioquia University, Medellin, Colombia.

1886/W Analysis of a de novo balanced translocation [46,XY,t(2;9)(p13;p24)] in a patient with autism spectrum disorder reveals direct interruption of RAB11FIP5.

Added as last author: Eli Hatchwell, Stony Brook University Medical Center, Stony Brook, NY.

2672/W Representing genomic variation: a hierarchical database structure.

New e-mail address: hgnc@genenames.org

• **THURSDAY, October 25**

Ancillary Events Added:

1:00 PM-2:00PM **Applied Biosystems Genotyping Workshop: Exploring Structural Variation and Copy Number in the Human Genome**, Hilton San Diego Gas Lamp Quarter, Santa Rosa Room

6:30 PM-9:00 PM **Qiagen – RNAi and miRNA Screening: Applications in Chemical and Functional Genomics**, San Diego Marriott Hotel and Marina, Coronado Room (4th Floor of the South Tower)

Platform Information Changed:

Session 21 Cancer Genetic and Cytogenetics
9:30 AM (73) **The leukemogenic CALM/AF10 fusion protein alters the subcellular localization of the lymphoid regulator Ikaros.**

The penultimate sentence should read:

The transcriptional repressor activity of Ikaros is reduced by AF10 but not by CALM/AF10.

continued

Thursday, October 25, continued

Session 29 Feeling Left Out: What Do Deletions Really Mean?

2:30 PM (79) **Dissecting clinical heterogeneity relevant to early development in autism by copy number variations.** The following co-authors have withdrawn: S.Yoon, K. Ye, M. Wigler, J. Sebat

Poster Presenter Changed:

1199/T Reduction of genomic complexity for re-sequencing by region-specific extraction. New presenter: M. Kunkel

• **FRIDAY, October 26**

Speaker Changed:

Session 52 Submicroscopic Chromosomal Duplications and Deletions: Medical Consequences and Population Genetics
5:20 PM **Copy number variation and susceptibility to autism spectrum disorders.** New speaker: S. Scherer, Department of Genetics and Genomic Biology, Hospital for Sick Children and University of Toronto, Ontario, Canada

Posters Changed:

635/F The NIDDK Central Repository. Using legacy data and samples to address new questions. New presenter: C. Scheper

926/F Identification of a ciliary protein as a novel contributor to sporadic heterotaxia.
Authors added: H. Olbrich, Univ. Hospital Freiburg, Germany; E. Greiner, Univ. Hospital Freiburg, Germany; V. Mohr, Univ. Hospital Freiburg, Germany; T. Severin, Univ. Hospital Freiburg, Germany; S. Dittrich, Univ. Hospital Freiburg, Germany; D. Kececioglu, Ruhr Univ. of Bochum, Germany; J. Riethmüller, Univ. Children's Hospital Tübingen, Germany; W. Knirsch, Univ. Children's Hospital Zurich, Switzerland; P. Bouvagnet, Univ. C. Bernard Lyon, France; M. Splitt, International Centre for Life, Newcastle upon Tyne, United Kingdom; R. Reinhardt, Max-Planck Inst. for Molecular Genetics, Berlin, Germany.

1417/F Gene-centric Association Mapping of Chromosome 3p implicates potential role of GPX1 in Crohn Disease. New presenter: P. Goyette

2630/F The correct title is: Detecting Loss-of-Heterozygosity and Amplification events from Illumina SNP genotyping data in the presence of stromal contamination and intra-tumor heterogeneity.
New presenter: J. Ragoussis

2640/F Development of strategies for efficient use of revolutionary sequencing technology for detecting human sequence variation. Author added: Z. Xuan

• **SATURDAY, October 27**

Award Update:

Session 64 C.W. Cotterman Award
Monetary awards of \$500 each and commemorative plaques will be presented to the first authors of the best papers published in *The American Journal of Human Genetics* during the previous year on which the first author is either a pre- or postdoctoral trainee and an ASHG member.

Each September, the editorial board of the Journal selects the articles that best represent outstanding contributions to the field of genetics.

Two awards are presented annually. This year the award winners are:

Min Shi, Ph.D., National Institute of Environmental Health Sciences, Biostatistics Branch. Winning paper: **Orofacial Cleft Risk Is Increased with Maternal Smoking and Specific Detoxification-Gene Variants**

Saima Riazuddin, Ph.D., National Institute on Deafness and Other Communication Disorders/ National Institutes of Health, Laboratory of Molecular Genetics. Winning paper: **Tricellulin Is a Tight-Junction Protein Necessary for Hearing**

Platform Information Changed:

Session 56 Getting Under the Skin
9:15 AM (242) **Evidence for T(Brachyury) as a Candidate Gene for Vertebral Malformations.**
New speaker: R.D. Blank. Second author added: N. Ghebranious, Marshfield Clinic, Marshfield, WI

Session 59 Therapy for Genetic Disorders
8:30 AM (269) **Sapropterin dihydrochloride (sapropterin) increases phenylalanine (Phe) tolerance in children with phenylketonuria (PKU) maintained on a Phe-restricted diet.** New presenter: B. Burton

continued

**• Exhibitors Added After
Program Guide Publication •**

• Accredo Nova Factor 132

1640 Century Center Parkway
Suite 105
Memphis, TN 38134
Tel: (901) 381-7400/(800) 235-8498
Fax: (901) 385-3666
Email: anitra.arcenaux@accredotx.com
URL: <http://www.accredonovafactor.com>
Accredo Nova Factor offers specialty pharmaceutical services customized for lysosomal storage disorder therapies. Our LSD-dedicated team will guide you and your patients through the therapy process from start to finish.

• AutoGenomics, Inc. 844

2251 Rutherford Road
Carlsbad, CA 92008
Tel: (760) 804-7378
URL: <http://www.AutoGenomics.com>
AutoGenomics, Inc., a privately held company based in Carlsbad, California, has developed an automated microarray based multiplexing molecular diagnostic platform. The INFINITI™ platform can be used to assess disease signatures with novel genomic and proteomic markers in areas of genetic disorders, infectious diseases, cancer, and pharmacogenetics.

**• Basal Cell Carcinoma Nevus Syndrome
Life Support Network 240**

PO Box 321
Burton, OH 44021
Tel: (440) 635-0078/(866) 834-1895
Fax: (440) 635-0267
Email: info@bccns.org
URL: <http://www.bccns.org>
BCCNS Life Support Network is dedicated to providing counseling & support services to children and adults manifesting inherited, metabolic or genetic disorders and diseases most commonly known as BCCNS, NBCCS, and/or Gorlin Syndrome, working to educate medical professionals on the importance of early diagnosis and treatment of patients with BCCNS.

• BioTechniques 231

52 Vanderbilt Avenue
7th Floor
New York, NY 10017
Tel: (212) 520-2714
Fax: (212) 561-6629
Email: christine.briglia@informausa.com
URL: <http://www.biotechniques.com>
BioTechniques, the international journal of life science methods, provides open access to first-quality, peer-reviewed papers on laboratory

techniques and protocols. Now in its 42nd volume, *BioTechniques* has over 80,000 print subscribers worldwide. The journal augments its peer-reviewed content with feature articles and topic-specific supplements. Visit www.biotechniques.com for more details.

• Cryo Bio System 239

420 Lexington Ave., Suite 300
New York, NY 10170
Tel: (212) 297-6156
Fax: (212) 986-1952
Email: contact@cryobiosystem-imv.com
URL: <http://www.cryobiosystem.com>
Cryo Bio System: State of the art cryopreservation technology for genomic biobanks and biorepositories.

• Embi Tec 446

7738 Arjons Drive
San Diego, CA 92126
Tel: (858) 684-3190/(800) 255-1777
Fax: (858) 684-3195
Email: inquiry@embitec.com
URL: <http://www.embitec.com>
Embi Tec® makes the RunOne™ DNA Electrophoresis System with Power Supply. Power Supply comes with or without timer feature. Pre-cast agarose/PAG gels. New product is the LightOne™ Illuminator, a compact, bench top pipetting aid for enhanced visualization of 96 well plates. It provides illumination of specific wells during dispensing.

• febit inc 207

200 Boston Ave
Suite 1850
Medford, MA 02155
Tel: (781) 391-4360
Fax: (781) 391-4362
Email: info@febit.com
URL: <http://www.febit.com>
About febit and Geniom: With the DNA analysis device Geniom, the German-American biotech company febit provides a whole world of novel experimental opportunities: it enables users to design, synthesize and apply a customized biochip right on their own laboratory bench. Geniom is an easy-to-use, compact benchtop device.

• National Disease Research Interchange 130

1628 JFK Boulevard
8 Penn Center, 8th Floor
Philadelphia, PA 19103
Tel: (215) 557-7361/(800) 222-6374
Fax: (215) 557-7154
Email: research@ndriresource.org
URL: <http://www.ndriresource.org>

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*Exhibitors Added After Program Guide Publication
(continued)*

**• NICHD Brain and Tissue Bank for
Developmental Disorders 140**

1550 NW 10th Avenue
R-5 PAP BLDG, Room 415
Miami, FL 33136
Tel: (305) 243-6834/(800) 592-7246
Fax: (305) 243-6970
Email: btbcoord@med.miami.edu
URL: <http://www.braindonation.org>
The NICHD Brain and Tissue Bank for Developmental Disorders at the University of Miami was established to collect samples from patients with a variety of genetic and developmental disorders, patients with related disorders and age-matched controls. The material is used by investigators studying diseases that affect normal brain development.

• Parallabs, Inc. 136

67 Millbrook Street, #522
Worcester, MA 01606
Tel: (508) 459-7727
Fax: (508) 459-7548
Email: office@parallabs.com
URL: <http://www.parallabs.com>
Parallabs presents: * **Chemagen™** automated extraction of DNA and RNA from a wide range of blood volumes and sample types. Advantages are fast processing, volumes from 10 µl to 10 ml of blood and robust chemistry. * **Nanodrop™** low volume, high precision dispensing systems.

• Rare Disease Therapeutics, Inc. 235

1101 Kermit Drive
Suite 608
Nashville, TN 37217
Tel: (615) 399-0700
Fax: (615) 399-1217
Email: tpodgorski@raretx.com
URL: <http://www.raretx.com>
Rare Disease Therapeutics, Inc., America's first biopharmaceutical company committed to developing and marketing drugs for rare disorders. Founded in 1991, Rare Disease Therapeutics is dedicated to patients with unmet medical needs that have been overlooked by other pharmaceutical companies.

• Rutgers University Cell & DNA Repository 846

Nelson Biological Laboratories
604 Allison Road - C Wing
Piscataway, NJ 08854
Tel: (732) 445-1498
Fax: (732) 445-1149
URL: <http://www.rucdr.org>

• Zymo Research Corporation 126

625 W Katella Avenue
Suite 30
Orange, CA 92867
Tel: (714) 288-9682/(888) 882-9682
Fax: (714) 288-9643
Email: ssanchez@zymoresearch.com
URL: <http://www.zymoresearch.com>
Specializing in the development of technologies for DNA/RNA purification and analysis, our goal is to provide the simplest, most reliable tools for DNA, RNA, and epigenetics research.