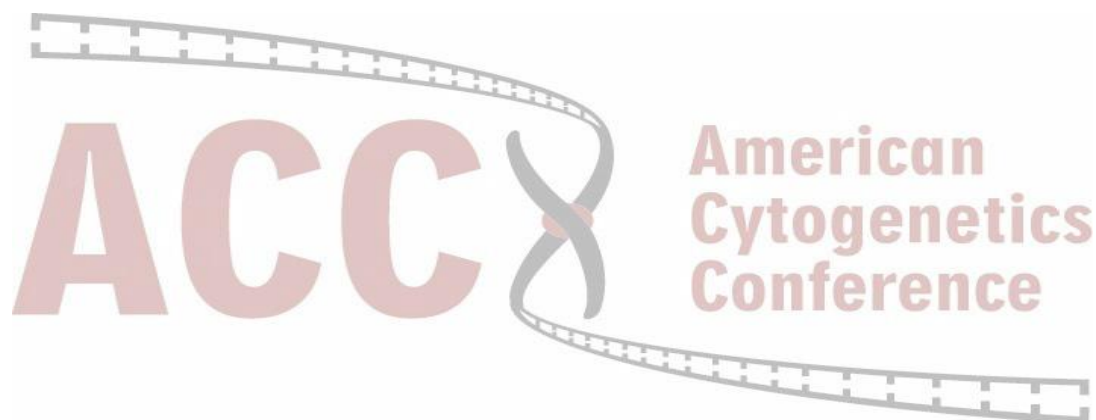


# 41<sup>th</sup> Biennial American Cytogenetics Conference



May 13-16, 2010 • Crowne Plaza Hotel • Niagara Falls, Ontario, Canada

## Preliminary Scientific Program

### Thursday, May 13

- Noon - 1:30 pm      **ACC Board Meeting – Lobby Board Room**
- 2:30 – 5:00 pm      **Exhibitor Registration and set up – Brock Room**
- 3:00 – 6:00 pm      **Conference Registration – Hotel Lobby**  
**Poster set-up – Niagara Room**
- 5:30 – 7:00 pm      **Opening Reception – Niagara and Brock Rooms**  
*Sponsored by LabCorp and PerkinElmer*

### Friday, May 14

- 7:00am – 12:00 pm    **Conference Registration - Lobby**  
**Poster set-up – Brock Room**
- 7:00 am – 6:00 pm    **Exhibitors open - Brock Room**  
Please visit their booths, especially during the continental breakfasts, breaks, lunch time and poster sessions!
- 3:00 – 6:00 pm      **Poster session – Niagara Room**
- 7:00 – 9:00 am      **Continental Breakfast – Brock Room**  
*Sponsored by Genetics Associates*

### All Scientific Sessions will take place in the - Niagara Room

- 8:00 – 8:15 am      **Welcome Address: Avirachan Tharapel - President**  
**and Susan Zneimer – Program Chair**

## **Scientific Session 1: Constitutional Cytogenetics I**

Moderators: Colleen Jackson-Cook and Kristin Deak

- 8:15 – 8:30 am      **1 Recurrent Chromosomal Translocations Mediated by Genomic Interchromosomal NAHR**  
S-H Kang, Z Ou, P Stankiewicz, AM Breman, J Wiszniewska, MLCooper, L Shao, ST South, K Coleman, PM Fernhoff, MJ Deray, S Rosengren, ER Roeder, VB Enciso, A Patel, AC Chinault, JR Lupski, SW Cheung
- 8:30 – 8:45 am      **2 Complex Rearrangements Require Complex Analysis by Array, FISH and Karyotype**  
X Li\*, LK Conlin, S Mulchandani, B Thiel, M Deardorff, E Zackai, M Falk, H Hakonsarson, N Spinner  
\*Student presentation
- 8:45 – 9:00 am      **3 All Four Common Segregants of a Familial Balanced Translocation in Liveborn Quadruplets**  
ST South, L Rowe, D LaGrave, R Vanzo, J Carey
- 9:00 – 10:00 am    **4 Comparative Cytogenetics**  
**Invited Speaker: Oliver Ryder**  
Adjunct Professor, University of California at San Diego  
Director, San Diego Zoo's Institute for Conservation Research  
*Introduced by Susan Zneimer*
- 10:00-10:30 am    **Coffee Break – Brock Room**  
*Sponsored by Blue Gnome Unlimited*
- Scientific Session 2: Constitutional Cytogenetics II**  
Moderators: Athena Cherry and Yasmine Akkari
- 10:30 – 10:45 am    **5 Prenatal Array CGH Testing: Managing “Abnormal” Results**  
DI Quigley, AC Holt, CL Beattie, PB Jacky
- 10:45 -11:00 am    **6 Detection of Homozygosity using SNP Based Microarrays: Case Reports and Lessons Learned**  
HM Kearney, C Wilson-Brackett, C Shanahan, SF Roberts, K Crandall, JB Kearney, WP Allen, E Boyd
- 11:00 – 11:15 am    **7 Delineating Chromosomal Regions with Complex Architectures using Single and Low Copy FISH Algorithms** WA Khan, JHM Knoll, PK Rogan

- 11:15 -11:30 am      **8 Analysis of Products of Conception by Microarray Analysis**  
 JL Smith, S Schwartz, J Tepperberg, PH Papenhausen
- 11:30-11:45 am      **9 Development of LOINC for Integrating Constitutional Cytogenetic Test Result Reports into Electronic Medical Record**  
 Y Heras\*, AR Brothman, MS Williams, JA Mitchell, SM Huff  
 \*Student Presentation
- 11:45-12:00 pm      **10 New ACMG Laboratory Guideline for Turner Syndrome: Positive Impact on Patient Care at MUSC**  
 DJ Wolff
- 12:00-1:30 pm      *Lunch on your own- Exhibitor area is open to all attendees during lunch*
- Scientific Session 3: Cancer Cytogenetics I**  
 Moderators: Peter Jacky and Cheryl DeScipio
- 1:30 – 1:45 pm      **11 An Act of Generosity Turned into a Quandary by Unexpected Mosaicism: Case Report of Constitutional Trisomy 8 Mosaicism in a Healthy Bone Marrow Donor**  
 N Uddin, MS Williams, ST South
- 1:45 – 2:00 pm      **12 Detection of Cytogenetic Abnormalities by Fluorescent in situ Hybridization (FISH) after Isolation of Plasma Cells**  
 H Aviv, I Maxwell, C Creese, K Vassallo
- 2:00 – 2:15 pm      **13 Apparently Novel *EWSR1* Gene Rearrangements Detected Two Astroblastomas**  
 K Craddock\*, I Sorokin, G Nie, M Zielenska, W Halliday, M Shago  
 \*Student presentation
- 2:15 – 2:30 pm      **14 Characterization of Uncommon BCR and ABL1 Breakpoints in 3 Cases of Chronic Myelogenous Leukemia**  
 FC Wheeler\*, K Kaiser-Rogers, K Muldrew, ML Gulley, KW Rao  
 \*Student presentation
- 2:30 – 2:45 pm      **15 Chromosome Kissing and Cancer**  
 BL Flynn\*, E Pizzo, J Crivello, J Brown  
 \*Student presentation
- 2:45 – 3:00 pm      **16 Stimulation of Chronic Lymphocytic Leukemia (CLL) Cells with CpG Oligodeoxynucleotides (ODN) Gives Consistent Karyotypic Results among Laboratories: a CLL Research Consortium (CRC) Study**

NA Heerema, JC Byrd, P Dal Cin, ML Dell'Aquila, P Koduru, A Aviram, S Smoley, LZ Rassenti, A Greaves, JR Brown, KR Rai, TJ Kipps, NE Kay, D van Dyke

- 3:00 – 3:30 pm **Coffee Break – Brock Room**  
*Sponsored by Genetix Corp*
- 3:00 – 6:00 pm **View posters and exhibits**
- 3:00 – 4:00 pm Designated time for authors of the **Constitutional Cytogenetics** section to be present at their posters
- 4:00 – 5:00 pm Designated time for authors of the **Cancer Cytogenetics** section to be present at their posters

**Distinguished Cytogenetics Award Dinner takes place in Niagara Room**

- 7:00 – 10:00 pm **ACC 2008 Distinguished Cytogeneticist Award Dinner**  
Award Recipient: Dr. Gordon Dewald  
*Sponsored by Abbott Molecular*

**Saturday, May 15**

- 7:00 – 8:30 am **Continental Breakfast – Brock Room**  
*Sponsored by Cytocell/Rainbow Scientific, Inc.*
- 7:00 – 12:30 pm **Conference Registration - Lobby**
- 10:00 – 11:00 am **Poster session – Niagara Room**
- Scientific Session 4: Cancer Cytogenetics II**  
Moderators: Charleen Moore and Laura Conlin
- 8:00 – 8:15 am **17 Heterogeneous MLL Abnormalities in 509 consecutively Newly Diagnosed Pediatric Acute Leukemias**  
X-Y Lu, M Folsom, M Azim, J Ringrose, S Plon, C Lau, C Steuber, J Margolin, K Rabin, P Rao
- 8:15 – 8:30 am **18 Molecular Study of Complex Karyotypes in Bone Marrow Cells of Patients with Myelodysplastic Syndromes (MDS)**  
Z Zemanova, K Michalova, J Brezinova, L Lizcova, S Izakova, D Bystricka, I Sarova, M Siskova, O Cerna, J Cermak

- 8:30 – 8:45 am      **19 Emerging Data using a Targeted Oligo-Based Microarray in the Myelodysplastic Syndromes Reveal Provocative Complexity and New Recurrent Genetic Aberrations**  
ML Slovak, T Brown, J-Y Han, LJ Campbell, M Wall, LG Shaffer, B Ballif, R Schultz
- 8:45 - 9:00 am      **20 Implementation of a High Density SNP Array with Cytogenetic and Molecular Techniques for the Characterization of Genetic Aberrations in a Patient with CML and Resistance to Imatinib Mesylate**  
R Toydemir, L Rowe, M Hibbard, M Salama, S Shetty
- 9:00 – 10:00 am      **21 Defining the Molecular Cytogenetic Landscape of Cancer Using Array Based Technologies**  
**Invited Speaker: Norma Nowak**  
Director, DNA Microarray and Genomics Facility  
Roswell Park Cancer Institute  
*Introduced by Ann Marie Block*  
*Sponsored by Illumina, Inc.*
- 10:00 – 11:00      **Coffee Break – Brock Room**  
*Sponsored by Roche Applied Sciences*
- 10:00 – 11:00      **View posters and exhibits**
- Scientific Session 5: Cancer Cytogenetics III**  
Moderators: Debra Saxe and Stephen Moore
- 11:00- 11:15 am      **22 Genomic Alterations in Breast Cancer Cell Lines and Mouse Xenograft Models from Fresh Human Tumors**  
M Herrler, W Ke, J Stanchfield, H Zhang, G Deng, SS Jeffrey
- 11:15 – 11:30 am      **23 Implementation of High Resolution SNP Array Analysis in the Clinical Cancer Cytogenetics Laboratory**  
J Biegel, D Wilmoth, L Wainwright, L Tooke, M Dougherty
- 11:30 – 11:45 am      **24 High Resolution Genome-wide Chromosomal Microarray Analyses of Matched Tumor and Germline DNA in Multiple Myeloma**  
S Kulkarni, V Huchtagowder, M Evenson, C Mullins, M Fiala, M Tomasson, R Vij
- 11:45 - 12:00 pm      **25 From Chromosomes to FISH to aCGH: Genomic Approaches to Study Chronic Lymphocytic Leukemia**  
YJ Liu

- 12:00 – 12:30 pm      **Discussion on Cancer Microarrays**
- Afternoon on your own**
- 1:00 pm                    **Departure for optional activities (box lunch pickup) – Brock Room**
- 6:30 – 10:00 pm        **Dinner and Entertainment – Niagara Room**
- Sunday, May 16**
- 7:00 – 9:00 am         **Continental Breakfast – Brock Room**  
*Sponsored by Baylor College of Medicine Medical Genetics Labs*
- 8:00 – 8:30 am         **ACC Business Meeting - Niagara Room**  
 With student award presentations
- Scientific Session 6: General Microarray Analysis**  
 Moderators: Barbara DuPont and Jillene Kogan
- 8:30 – 8:45 am         **26 Interpretation of Microarray Results: A Point System to Discriminate Benign, Unknown Significance and Copy Number Changes**  
 DAS Batista, E Wohler, S Morsey, J Biscoe, L Wo, N Jinawath
- 8:45 – 9:00 am         **27 Leveraging Genotype Information from SNP Microarrays for Cytogenetic Purposes**  
 V Aggarwal\*, O Nahum, E Carron, D Warburton, V Jobanputra, B Levy  
 \*Student presentation
- 9:00 – 9:15 am         **28 CNV and Regions of Homozygosity Identified by SNP Array: Difficulties in Elucidating Clinical Significance.**  
 E Wohler\*, N Jinawath, A Hamosh, J Hoover-Fong, A Elias, D Valle, D Batista  
 \* Student presentation
- 9:15 – 9:30 am         **29 Subtelomeric CNV: Mechanisms of Chromosome Repair**  
 K Rudd, Y Luo, K Hermetz
- 9:30 – 9:45 am         **30 Towards a CNV Atlas for Human Development**  
 CL Martin, EB Kaminsky, B Bunke, D Kunig, V Kaul, K Huang, D Saul, DL Pickering, DM Golden, E Aston, TJ Gliem, T Ackley, S Huang, JC Barber, JA Crolla, R Iyer, EC Thorland, AR Brothman, W Sanger, MR Rossi, MK Rudd, S Aradhya, DH Ledbetter
- 9:45 – 10:00 am       **31 Next Generation Sequencing (NGS) in Mendelian Cytogenetics**  
 N Tommerup, SC Minocherhomji, L Klitten, L Nazaryan, R Buonincontri, RS Møller, H Hjalgrim, KF Henriksen, HM Nielsen, I Bache, A Silahatoglu, C Hansen, M Bak

- 10:00 -11:00 am      **32 Human Genome Structural Variation: From Cytogenetics to Sequence**  
**Invited Speaker: Evan Eichler**  
Principal Investigator and Professor,  
Department of Genome Sciences and Howard Hughes Medical Institute, University of  
Washington, Seattle  
*Introduced by Urvashi Surti*
- 11:00 – 11:30 pm      **Coffee Break – Brock room**  
*Sponsored by Roche Applied Sciences*
- Scientific Session 7: Cytogenetics III**  
Moderators: John Wiley and Gordana Raca
- 11:30 – 11:45 am      **33 Mosaic Trisomy 13: Understanding the Origin using SNP**  
**Array**  
N Jinawath\*, R Zambrano, E Wohler, D Batista  
\* Student presentation
- 11:45 – 12:00 pm      **34 Increasing the Yield and Accuracy of Karyotypes from**  
**Spontaneous Abortion Specimens**  
V Jobanputra, C Esteves, A Sobrino, S Brown, J Kline, D Warburton
- 12:00 – 12:15 pm      **35 Variable Mechanisms of Chimera Formation Identified by SNP Array Analysis**  
LK Conlin, BD Thiel, S Mulchandani, DN Hartung, L Medne, E Zackai, CG  
Bonnemann, BK Burton, MA Deardorff, HH Akonarson, NB Spinner
- 12:15 – 12:30 pm      **36 SNP Microarray Analysis offers New Insights into the Accuracy of FISH**  
**Diagnostics in Preimplantation Embryos**  
B Levy, LE Northrop, NR Treff, RT Scott Jr.
- 12:30 – 12:45 pm      **37 SNP Microarray Detects Mosaic Segmental Uniparental Isodisomy in Three**  
**Cases**  
TA Smolarek, K Lipscomb Sund, HM Saal, R Hopkin, L Bao, SL  
Zimmerman
- 12:45 – 1:00 pm      **38 Molecular Characterization of Chromosome Aberrations in 146 Patients with**  
**Phelan-McDermid Syndrome by Array-CGH**  
BR DuPont, A Dwivedi, L Boccuto, RC Rogers, K Phelan
- 1:00 – 1:15 pm      **ACC 2012 and Adjournment**

## Poster Presentations

Poster set up times are Thursday 3:00 – 6:00 pm  
and Friday 7:00 am – Noon

Designated times for authors to be at their posters are:  
Friday 3:00 – 4:00 pm – Constitutional Cytogenetics posters  
Friday 4:00 – 5:00 pm – Cancer Cytogenetics posters

### Constitutional Cytogenetics

#### **39 Array Comparative Genomic Hybridization Identifies New Candidate Genes for Idiopathic Mental Retardation**

A Bewell, C Jackson, G Rice, G Raca, J Laffin

#### **40 Cytogenetics of Mental Retardation**

D Chandel, VR Hydrabadi, Rao M V

#### **41 Prenatal Diagnosis of De Novo Partial Trisomy 2q**

AC Kuskucu, K Yasarbas, G Yildirim, O Sahin, A Gul, Y Ceylan

#### **42 Origin and Phenotypic Consequences of Mosaic 45,X / 47,X,dup(Y)(q11.2q12),+i(18)(p10) Karyotype**

S Moore, R Cooper, H Lawce, S Olson

#### **43 SNP-based Genomic Microarray in the Workup of Mental Retardation**

J Kogan, S Zimmerman, S Mikulich, T Smolarek

#### **44 A Case of Bardet-Beidl Syndrome Associated with Homozygosity and Identified by Chromosome Microarray Analysis**

J E Wiley, H Kearney, B Ozturk, A Meyer

#### **45 Uncovering Single Gene Disorders with aCGH: An Example of a family with Branchiootorenal Spectrum Disorder**

MA Manning, J Kaplan, J Bernstein, KM Chen, AM Cherry

#### **46 Hybridization of a Centromere Probe to a Non-specific Chromosome in Interphase FISH. Y-P Essig, B Dickey, M Prentice, SA Berend**

#### **47 Implementation of a Global Platform-Independent CNV Repository**

C Parman, S Verma, Z Che, R Keshavan, S Shams

#### **48 Prenatal Diagnosis of Complete Monosomy 21 Confirmed by FISH and array-CGH**

AS Kulharya, VS Tonk, K King, P Miller, C Lovell, L Devoe, D Flannery

#### **49 Uncovering Hidden Cytogenetic Aberrations by Integration of Karyotyping, FISH and Chromosomal Microarray**

J Liu, BA Kozel, M Simon, JY Bauer, S Kulkarni

**50 Subjectivity in Chromosome Band Assessment**

K Geiersbach, M Sederberg, N Uddin, S Shetty

**51 Small Genomic Abnormalities Observed by Array CGH can be Detected using Oligonucleotide FISH Probes**

LS Rector, NA Yamada, MC Sederberg, ME Aston, RA Ach, P Tsang, E Carr, A Scheffer-Wong, N Sampas, B Peter, L Bruhn, AR Brothman

**52 Institutional Clinical Experience Using the Oligo- SNP Chromosomal Microarray**

F Quintero-Rivera, W Grody, E Baldwin, S Cederbaum, B Crandall, K Dipple, M Fox, JA Martinez-Agosto, J Peredo, E Vilain, X Li, PN Rao

**Cancer Cytogenetics**

**53 Whole Genomic Microarray Analysis of Pediatric Acute Leukemia Cases**

LS Jenkins, X Li, V Kiley

**54 Cancer Predisposition Syndromes and Array Comparative Genomic Hybridization: When One Answer does not Tell the Whole Story**

RD Clark, S Ramanathan, D Michelson

**55 Chromosome Analysis Detects two Cases of Donor Cell Disease Following Allogeneic Bone Marrow Transplant**

J Malik, C Miller, R Blumer, E Johnson, K Thompson, J Northup, J Laffin, G Raca

**56 Recurrent Interstitial Genomic Deletion of the Human *PTEN* Gene are Facilitated by Flanking Microhomologies**

JA Squire, M Yoshimoto, A Evans, K Sircar, T Bismar, J Williams, P Nuin

**57 Tetrasomy 13 as the Sole Chromosomal Abnormality in Acute Myeloid Leukemia**

CM Moore, GVN Velagaleti, S Vemulapalli

**58 Variant Philadelphia Translocations in Chronic Myeloid Leukemia: Genesis and Prognosis**

MM Brahmhatt\*, PS Patel, PJ Trivedi, SN Shukla, PM Shah, SR Bakshi

\*Student presentation

**59 Infantile Mixed Phenotype Acute Leukemia (Bilineal and Biphenotypic) with t(10;11)(p12;q23);MLL-MLLT10**

Z Lou1, CC Zhang, CA Tirado, T Slone, J Zheng, CM Zaremba1, D Oliver, W Chen

**60 Tumor Metastasis of *Peromyscus leucopus* Harderian gland tumors**

EE Mlynarski, CJ Obergfell, MJ O'Neill, RJ O'Neill

**61 Expanded FISH Assay Panel for Lymphoid Neoplasms Resulted Negative by Cytogenetics and Current FISH Panels but Positive by Hematopathology as a Reflex Test Rather than as a Part of Standard FISH Panels**

NS Mitter, S Lanno, J Blackson, M Donskoy, R Ehrenpreis

**62 *CIZ* Gene Rearrangements in Pediatric CD10-Negative Acute Lymphoblastic Leukemia**

M Shago, G Maire, O Abl, J Hitzler, S Weitzman, M Abdelhaleem

**63 Detection of a Minimally Deleted Region of 6q21 in Chronic Lymphocytic Leukemia: Efficacy of BAC Array Data in Development of a Targeted 6q21 Fluorescence *In Situ* Hybridization (FISH) Probe**

JL Murata-Collins, V Bedell, DB Estrine, SY Kim, Y-H Hsu

**64 Chronic Lymphocytic Leukemia with t(14;19)(q32;q13) is Characterized by Atypical Morphologic, Immunophenotypic, and Genetic Features, and Stereotypy of the B-Cell Antigen Receptor**

LV Abruzzo, CD Schweighofer, R Luthra, R Sargent, RP Ketterling, RA Knudson, LL Barron, MJ Keating, LJ Medeiros, YO Huh

**65 Validation of FISH Studies on Chronic Lymphocytic Leukemia (CLL) by Members of the CLL Research Consortium**

SA Smoley, DL Van Dyke, NE Kay, NA Heerema, ML dell' Aquila, PD Cin, P Koduru, A Aviram, L Rassenti, JC Byrd, KR Rai, JR Brown, AW Greaves, TJ Kipps, J Eckel-Passow, GW Dewald

**66 Chromosomal Aberrations in 48 Patients with Multiple Myeloma: Three-year Study from a Single Institution in Singapore**

L-C Lau, P Lim, G-Y Lee, L-E Loo, P-S F Liaw, Y-C J Lim, L-M Teng, H-S K See, T-H Lim, S-T A Lim, D Tan, S-L Tien

**67 A Female Patient with *FIP1L1-PGDFRA*-positive HES/CEL after Receiving R-CHOP Chemotherapy for Large Cell Lymphoma.**

S Kantarci, M Zhang, M Gorman, G Pihan

**68 A Method for Achieving a 4-hour Turnaround Time for FISH**

JM Cowan, KA Krajewski, SC Tillson, NA Kalgren, KJ Connor

**69 Use of Microarray Technology (aCGH) in Chronic Lymphocytic Leukemia (CLL): Evaluation of Both Copy Number Changes and Relevant IGH Translocations**

R Schultz, LG Shaffer, H Greisman, R Tubbs, K Theil, J Cook, T Brown, B Ballif, ML Slovak