Curriculum Vitae

Philip D. Cotter, PhD, FACMG, FFSc(RCPA)

Email: Philip.Cotter@ucsf.edu
pcotter@researchdx.com
Phone: (510) 332 0477
Fax: (949) 419 3488

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EDUCATION:

1990 – 1994 **Doctor of Philosophy** - Biomedical Sciences Graduate Program in Human Genetics. Mount Sinai School of Medicine of the City University of New York, New York, NY.

1990 – 1992 **Master of Philosophy** - Biomedical Sciences Graduate Program in Human Genetics. Mount Sinai School of Medicine of the City University of New York, New York, NY.

1987 – 1990 **Diploma of Management.** New Zealand Institute of Management, Wellington, New Zealand.

1988 – 1989 **Diploma of Medical Laboratory Technology** - Clinical Cytogenetics. New Zealand Board of Medical Laboratory Technology, Wellington, New Zealand.

1984 – 1986 **Master of Applied Science (Honours)** - Plant Breeding and Cytogenetics. Lincoln University, Lincoln, New Zealand.

1980 – 1983 **Bachelor of Science** - Botany and Microbiology. University of Canterbury, Christchurch, New Zealand.

PROFESSIONAL EXPERIENCE:

2009 – Laboratory Director, Pacific Diagnostics Clinical Laboratory, Irvine, CA.

2008 – Principal and Co-Founder, ResearchDx Inc., Irvine, CA.

2007 – Associate Clinical Professor, Department of Pediatrics, College of Medicine, University of California San Francisco, San Francisco, CA.

2004 – 2015 Vice President of Clinical Laboratories and Laboratory Director, Biocept Laboratories, San Diego, CA.

2010 – 2013 Chief Scientific Officer and Vice President of Operations, CymoGen Dx LLC, Irvine, CA.

2003 – 2006 Adjunct Associate Professor, Department of Pediatrics, College of Medicine, University of California San Francisco, San Francisco, CA.

2003 – 2004 Director of Advanced Molecular Diagnostics and Chief of the FISH and Molecular Genetics Laboratories, US Labs Inc., Irvine, CA.

2002 – 2004 Associate Medical Director - Cytogenetics and Molecular Genetics, US Labs Inc., Irvine, CA.

1997 – 2003 Adjunct Assistant Professor, Department of Pediatrics, College of Medicine, University of California San Francisco, San Francisco, CA.

1996 – 2002 Director, Cytogenetics Laboratory and Director, Molecular Genetics Laboratory, Division of Medical Genetics, Children's Hospital Oakland, Oakland, CA.

1995 – 1996 Assistant Director, Cytogenetics Laboratory, Department of Human Genetics, Mount Sinai School of Medicine, New York, NY.

1994 – 1996 Director, Molecular Cytogenetics, Department of Human Genetics, Mount Sinai School of Medicine, New York, NY.

1994 – 1996 Postdoctoral Fellow, American Board of Medical Genetics Training Program, Department of Human Genetics, Mount Sinai School of Medicine, New York, NY.

1987 – 1990 Clinical Cytogeneticist, Cytogenetics Department, Princess Mary Children's Hospital, Auckland, New Zealand.

TEACHING EXPERIENCE:

2015 – Continuing Education Program Director, ResearchDxU, ResearchDx Inc., Irvine, CA.

2009 – Advisory Board, Medical Laboratory Technician Training Program, Miramar College, San Diego, CA.

2007 – 2013 Molecular Genetic Pathology Fellowship Faculty, Departments of Pathology and Laboratory Medicine, University of California San Francisco, San Francisco, CA.

2012 – 2013 Training Program Director, Molecular Genetic Specialist Training Program, Illumina Clinical Services Laboratory, San Diego, CA.

2011 – 2014 Training Program Director, State of California Cytogenetics Specialist Training Program, Biocept Laboratories, San Diego, CA.

2007 – 2011 Cytogenetics Training Program Director, NeoGenomics Inc., Fort Myers, FL.

1997 – 2007 American Board of Medical Genetics Training Program in Medical Genetics, Division of Medical Genetics, Department of Pediatrics, University of California San Francisco, San Francisco, CA.
Cytogenetics Training Program Director
Molecular Genetics Training Program Director

2001 – 2002 Cytogenetics Lectures, Nursing Genetics Seminar Course, School of Nursing, University of California San Francisco, San Francisco, CA.

1999 – 2002 Cytogenetics Lectures, Medical Genetics Course, University of California San Francisco Medical School, San Francisco, CA.

1999 – 2000 Cytogenetics Module, Program in Genetic Counseling, School of Public Health, University of California Berkeley, Berkeley, CA: HMS 245A Clinical Genetics: Biology and Methodology - Introduction to Chromosomes and DNA Analysis.

1990 Clinical Cytogenetics Lecturer, National Diploma in Medical Laboratory Technology, Auckland Technical Institute, Auckland, New Zealand.

1989 – 1990 Clinical Cytogenetics Lecturer, Auckland School of Medical Laboratory Technology, Auckland Hospital, Auckland, New Zealand.

HONORS AND AWARDS:

1992 – 1994 March of Dimes Birth Defects Foundation Predoctoral Graduate Research Training Fellowship.
1992 American Society of Human Genetics Predoctoral Clinical Research Award.
1990 Auckland Area Health Board School of Medical Laboratory Technology, Cytogenetics Specialist Level Award
1989 Auckland Area Health Board School of Medical Laboratory Technology, Cytogenetics Certificate Level Award

PROFESSIONAL CERTIFICATION/LICENSES:

American Board of Medical Genetics and Genomics
Diplomate, Clinical Cytogenetics: Certified 1996; Recertified 2010
Diplomate, Clinical Molecular Genetics: Certified 1996; Recertified 2010

American College of Medical Genetics and Genomics
Fellow (FACMG): 1996 –

American Society for Clinical Pathology Board of Certification
Technologist in Cytogenetics [CG(ASCP)^{CM}]: 2009 –
Technologist in Molecular Biology [MB(ASCP)^{CM}]: 2009 –

Human Genetics Society of Australasia
Member: Molecular Genetics (MHGSA): 2008 –

Florida State Department of Health, Board of Clinical Laboratory Personnel
Cytogenetics Director (DI38910): 2004 –
Molecular Genetics Director (DI38910): 2004 –

National Certification Agency for Medical Laboratory Personnel
Clinical Laboratory Specialist in Cytogenetics [CLSp(CG)]: 1990 – 2009
Clinical Laboratory Specialist in Molecular Biology [CLSp(MB)]: 2003 – 2009

National Provider Identifier: 1346228087

New Jersey State Board of Medical Examiners
Laboratory Director, Cytogenetics (25MS00009200): 2005 – 2015
Laboratory Director, Clinical Molecular Genetics (25MS00009200): 2005 – 2015

New York State, Department of Health: Certificate of Qualification
Cytogenetics: 2002 –
Genetic Testing – Molecular: 2002 –
Oncology – Cellular Tumor Markers: 2005 –

New Zealand Medical Laboratory Science Board
Registered Medical Laboratory Scientist (30-04620): 2005 –
Annual Practicing Certificate: 2005 –

Royal College of Pathologists of Australasia
Fellow, Faculty of Science [FFSc(RCPA)]: 2010 –

State of California, Department of Health Services
Clinical Cytogeneticist, Laboratory Director (DRM1): 2001 –
Clinical Genetic Molecular Biologist, Laboratory Director (DRN1): 2001 –

Tennessee Department of Health, Medical Laboratory Board
Director, Cytogenetics (21882): 2006 –
Director, Molecular Diagnostics (21882): 2006 –

PROFESSIONAL SOCIETIES:

American College of Medical Genetics: 1993 –
American College of Obstetricians and Gynecologists, Affiliate: 2005 – 2008
American Society for the Advancement of Science: 1991 –
American Society for Reproductive Medicine: 2005 – 2008
American Society of Hematology: 2004 –
American Society of Human Genetics: 1991 –
Association of Clinical Cytogeneticists (UK): 1988 – 2000
Association of Genetic Technologists: 1988 –
Association for Molecular Pathology: 2002 –
British Society for Human Genetics: 1996 – 2000
Clinical Laboratory Management Association: 1999 –
European Society of Human Reproduction and Embryology: 2005 – 2007
The Harvey Society: 1996 – 2004
Human Genetics Society of Australasia: 1988 –
International Society for Prenatal Diagnosis: 2002 – 2008
New Zealand Genetical Society: 1984 – 1993
New Zealand Institute of Medical Laboratory Science: 2005 –

PROFESSIONAL ACTIVITIES:

Company Boards:
RiboMed BioTechnologies, Carlsbad, CA: Board Member: 2012 –

Editorial Boards:

Applied Scientific Reports: 2013 –
Austin Journal of Genetics and Genomic Research: 2014 –
Austin Journal of Pediatrics: 2014 –
Biomedical Research and Clinical Practice: 2016 –
Case Reports in Genetics: 2011 –
Cell Science and Report: 2014 –
Datasets in Medicine: 2012 –
Enliven: Journal of Genetic, Molecular and Cellular Biology: 2013 –
Frontiers in Biotechnology: 2013 – 2014
Frontiers in Pathology and Genetics: 2013 –
German Journal of Health Research: 2013 –
Global Journal of Human Genetics & Gene Therapy: 2013 –
Henry Journal of Perinatology and Pediatrics: 2015 –
Integrative Journal of Biomedical Research: 2015 –
International Journal of Chronic Diseases & Therapy: 2015 –
International Journal of Genetic Engineering: 2011 –
International Journal of Life Science and Medical Research: 2013 –
International Journal of Medical Biotechnology & Genetics: 2013 –
Journal of Advances in Molecular Biology: 2016 –
Journal of the Association of Genetic Technologists, Review Board: 1998 –
Journal of Bioinformatics & Computational Biology: 2014 –
Journal of Cell Science and Report: Open Access: 2014 –
Journal of Down Syndrome and Chromosome Abnormalities: 2015 –
Journal of Genetic Mutation Disorders: 2015 –
Journal of Genetics Study: 2013 –
Journal of Genomics and Bioinformatics: 2016 –
Journal of Life Medicine: 2014 –
Journal of Medical Statistics and Informatics: 2013 –
Journal of Metabolomics: 2014 –
Kenkyu in Stem Cell and Regenerative Medicine: 2015 –
Molecular Biology and Genetic Engineering: 2013 –
Open Access (OA) Genetics: 2012 –
Open Journal of Genetics, Associate Editor: 2011 –
Science, Technology and Development: 2015 –
Scholarena Journal of Genetics: 2016 –
SciTz Pediatrics & Therapeutics: 2016 –
SOJ Genetic Science: 2016 –
Source Journal of Genomics: 2015 –
The Open Access Journal of Science and Technology: 2016 –
The Open Conference Proceeding Journal: 2015 –
The Open Pediatrics Medicine Journal: 2009 –
WebmedCentral Lead Faculty in Genetics: 2011 –
WebmedCentral *plus* Genetics Editorial Board: 2012 –
WebmedCentral *plus* Genetics Specialty Editor: 2012 –

Medical/Scientific Advisory Boards:

Birth Defects Research Laboratory, University of Washington, Seattle, WA: 2007 –
DNA Direct, San Francisco, CA: 2005 – 2011

IGENZ Ltd, Auckland, New Zealand: 2006 –
Jerichons, LLC, Ann Arbor, MI: 2015 –
NeoGenomics Inc., Fort Myers, FL: 2005 – 2012
PGD by ART, ART Reproductive Center, Beverly Hills, CA: 2004 – 2006
RiboMed BioTechnologies, Carlsbad, CA: 2011 –
Sun Genomics, San Diego, CA: 2016 –

Committees

Association for Molecular Pathology: Whole Genome Analysis Working Group: 2010 – 2013
Clinical and Laboratory Standards Institute: Subcommittee on Proficiency Testing (External Quality Assessment) for Molecular Methods (MM14) – Member: 2010 –
Clinical and Laboratory Standards Institute: Subcommittee on Quality Management System: Laboratory Internal Audit Program (GP39) – Observer: 2010 –

Scientific Meeting Session Chair

Technology in the Service of Companion Diagnostics: Fourth Annual Next Generation Dx Summit, Washington, DC, August 21-23, 2012.
Regulatory and Compliance: BioPharma Asia Convention 2015, Singapore, March 23-25, 2015.

Scientific Meeting Panels

Incorporating Companion Diagnostics Considerations Early in Product Development: Fifth Annual Next Generation Dx Summit, Washington DC, August 20-22, 2013.
Implementing CLIA in Clinical Operations: BioPharma Asia Convention 2015, Singapore, March 23-25, 2015.

Grant Reviewer:

Intramural Research Fund Grant Reviewer, King Fahad Medical City, Riyadh, Kingdom of Saudi Arabia: 2016.

Consultant:

21st Century Oncology, Fort Myers, FL: Clinical Consultant: 2009 – 2012
Acupath Laboratories, New Hyde Park, NY: Director of Cytogenetics: 2004
BaseHealth, Redwood City, CA: Clinical Consultant: 2015 –
Biocept Inc., San Diego, CA: Laboratory Consultant: 2004, 2014 – 2015
Biological Dynamics, Inc., San Diego, CA: Laboratory Director: 2013 – 2015
Cancer Genetics, Inc., Rutherford, NJ: Consultant Cytogeneticist: 2009 – 2010
Children's Hospital and Research Center at Oakland, Department of Pathology, Oakland, CA:
Director of Cytogenetics: 2002 – 2013
Director of Molecular Genetics: 2002 – 2014
CombiMatrix Molecular Diagnostics, Inc., Irvine, CA:
Laboratory Director/Clinical Consultant: 2006 – 2010
Coulter Pharmaceuticals, South San Francisco, CA: Consultant Cytogeneticist: 1999 – 2000
Diagnomics, San Diego, CA: Laboratory Director: 2015 –
Elim Biopharmaceuticals, Inc., Hayward, CA: Technical Supervisor: 2015 –
IGENZ Ltd., Auckland, New Zealand: Clinical and Scientific Director of Genetics: 2006 – 2013
Illumina Clinical Services Laboratory, Illumina Inc., San Diego, CA:
Laboratory Director: 2008 –
Molecular Pathology Laboratory Network, Maryville, TN: Clinical Cytogeneticist: 2013, 2015 –
MyGenetx Laboratory, Franklin, TN: Clinical Consultant: 2016 –

NeoGenomics Inc.
NeoGenomics Inc., Irvine, CA: Laboratory Director/Clinical Cytogeneticist: 2006 – 2012
NeoGenomics Inc., Fort Myers, FL: Clinical Cytogeneticist: 2004 – 2012
NeoGenomics Inc., Nashville, TN: Clinical Consultant: 2008 – 2012
Parabase Genomics, Boston, MA: Clinical Consultant: 2015 –
Personalis, Inc., Menlo Park, CA: Clinical Consultant: 2012 – 2013
Pfizer Oncology Clinical Research Laboratory, La Jolla, CA: Laboratory Director: 2011 – 2012
Prognosys Biosciences, La Jolla, CA: Laboratory Director: 2008 – 2012
Questgenomics, Nanjing, China: Clinical Consultant: 2015 –
Quest Diagnostics, San Juan Capistrano, CA: Clinical Cytogeneticist: 2004
RiboMed BioTechnologies, Carlsbad, CA: Clinical Consultant: 2010 – 2013
Rieback Medical-Legal Consultants, Fort Lauderdale, FL: Consultant Geneticist: 2002 –
Riken Genesis, Tokyo, Japan: Clinical Consultant: 2014 –
Sequenta, South San Francisco, CA: Clinical Consultant: 2013 – 2015
UCSF Benioff Children's Hospital Oakland, Oakland, CA:
Director of Molecular Genetics: 2014 –
Vantage Point Laboratories, San Diego, CA: Clinical Cytogeneticist: 2012 –
Verinata Health, Inc., San Carlos, CA: Cytogenetics Consultant: 2010 – 2013
WuXi AppTec Central Laboratory, Shanghai, China: Laboratory Director: 2015 –
WuXi AppTec Genomics Clinical Laboratory, Shanghai, China: Laboratory Director: 2012 – 2015
WuXi NextCODE Genomics, Shanghai, China: Laboratory Director: 2015 –

***Ad Hoc* Reviewer for:**

Advances in Genomics and Genetics
American Journal of Medical Genetics
American Journal of Pathology
Application of Clinical Genetics
Austin J. of Genetics and Genomic Research
Austin Journal of Pediatrics
Biomedical Research and Clinical Practice
Birth Defects Research
Cancer Research Journal
Case Reports in Genetics
Cleft Palate-Craniofacial Journal
Clinica Chemica Acta
Clinical Chemistry
Clinical and Experimental Dermatology
Clinical Genetics
Clinical Investigation
Clinical and Laboratory Haematology
Clinical Medicine Insights
Congenital Anomalies
Cytogenetic and Genome Research
Doody Online Publishing - Genetics
European Journal of Human Genetics
European Journal of Medical Genetics
European Journal of Pediatrics
Genes, Chromosomes and Cancer
International Journal of Genetics and Genomics
International Journal of Laboratory Hematology
International J. of Life Science & Medical Research
J. Association of Genetic Technologists
Journal of Genetic Counseling
Journal of Genetics Study
Journal of Genetic Syndromes & Gene Therapy
Journal of Life Medicine
Journal of Medical Genetics
Journal of Metabolomics
Journal of Molecular Diagnostics
Journal of Personalized Medicine
Journal of Visualized Experiments
Lancet
Medical Principles and Practice
Nature Biotechnology
Open Conference Proceedings Journal
Open Journal of Genetics
Open Pediatrics Medicine Journal
Pediatrics
Pediatrics International
Personalized Medicine
Pharmacogenomics
Prenatal Diagnosis
Public Health Frontier

Genetic Testing and Molecular Biomarkers
Genetics in Medicine
Global J. Human Genetics & Gene Therapy
Human Genetics
International Journal of Fertility and Sterility
SciTz Pediatrics & Therapeutics
Source Journal of Genetics
Source Journal of Genomics
Tohoku Journal of Experimental Medicine

Other:

American Association for Laboratory Accreditation
Accreditation Council: 2010 –
ISO 15189 Assessor: 2010 –
CLIA Assessor: 2014 –
Association of Genetic Technologists, Student Research Award Abstract Review Committee: 2010 –
California Department of Health Services, Laboratory Field Services
Clinical Laboratory Director Licensure Examiner: 2001 –
Board of Examiners, PhD Dissertation
Cairo University Faculty of Science, Giza, Egypt: 2005
University of Madras, India: 2011, 2015
Children's Oncology Group, Associate Member: 2000 – 2004
College of American Pathologists, Laboratory Inspector, Inspector ID# 1021745: 1998 –
UCLA ABMG Training Program Committee - Tina Hambuch PhD: 2011 – 2013

PUBLICATIONS:

1. Cotter PD, Stewart NL: Partial trisomy 17q and monosomy 9p due to a familial translocation. *Ann. Génét.* 33: 231-233, 1990.

2. Cotter PD: Chromosome preparations from direct and overnight cultures of colonic adenomatous polyps. *N.Z. J. Med. Lab. Sci.* 45: 16-17, 1991.

3. Cotter PD, Willard HF, Gorski JL, Bishop DF: Assignment of human erythroid δ-aminolevulinate synthase (ALAS2) to a distal subregion of band Xp11.21 by PCR analysis of somatic cell hybrids containing X;autosome translocations. *Genomics* 13: 211-212, 1992.

4. Cotter PD, Baumann M, Bishop DF: Enzymatic defect in "X-linked" sideroblastic anemia: molecular evidence for erythroid δ-aminolevulinate synthase deficiency. *Proc. Natl. Acad. Sci. USA* 89: 4028-4032, 1992.

5. Cotter PD, Tumewu P, Browett PJ: Deletion of the long arm of chromosome 20 in a patient with small cell lymphocytic lymphoma. *Cancer Genet. Cytogenet.* 70: 142-143, 1993.

6. Jardine PE, Cotter PD, Johnson SA, Fitzsimons EJ, Tyfield L, Lunt PW, Bishop DF: Pyridoxine refractory congenital sideroblastic anaemia with evidence for autosomal inheritance: exclusion of linkage to ALAS2 at Xp11.21 by polymorphism analysis. *J. Med. Genet.* 31: 213-218, 1994.

7. Cotter PD, Rucknagel DL, Bishop DF: X-linked sideroblastic anemia: identification of the mutation in the erythroid-specific δ-aminolevulinate synthase (ALAS2) gene in the original family described by Cooley. *Blood* 84: 3915-3924, 1994.

8. Cotter PD, Drabkin HA, Varkony T, Smith DI, Bishop DF: Assignment of the human housekeeping δ-aminolevulinate synthase gene (ALAS1) to chromosome band 3p21.1 by PCR analysis of somatic cell hybrids. *Cytogenet. Cell Genet.* 69: 207-208, 1995.

9. Bottomley SS, May BK, Cox TC, Cotter PD, Bishop DF: Molecular defects of erythroid 5-aminolevulinate synthase in X-linked sideroblastic anemia. *J. Bioenerg. Biomembr.* 27: 161-168, 1995.

10. Cotter PD, May A, Fitzsimons EJ, Houston T, Woodcock B, Al-Sabah AI, Wong L, Bishop DF: Late onset X-linked sideroblastic anemia: missense mutations in the erythroid δ-aminolevulinate synthase (ALAS2) gene in two pyridoxine-responsive patients initially diagnosed with acquired refractory anemia and ringed sideroblasts. *J. Clin. Invest.* 96: 2090-2096, 1995.

11. Cotter PD, Caggana M, Willner JP, Babu A, Desnick RJ: Prenatal diagnosis of a fetus with two balanced de novo chromosome rearrangements. *Am. J. Med. Genet.* 66: 197-199, 1996.

12. Levy B, Gershin IF, Desnick RJ, Babu A, Gelb BD, Hirschhorn K, Cotter PD: Characterization of a de novo unbalanced chromosome rearrangement by comparative genomic hybridization and fluorescence in situ hybridization. *Cytogenet. Cell Genet.* 76: 68-71, 1997.

13. Wright TJ, Ricke DO, Denison K, Abmayr S, Cotter PD, Hirschhorn K, Keinänen M, McDonald-McGinn D, Somer M, Spinner N, Yang-Feng T, Zackai E, Altherr MR: A transcript map of the newly defined 165 kb Wolf Hirschhorn syndrome critical region. *Hum. Mol. Genet.* 6: 317-324, 1997.

14. Gelb BD, Zhang J, Cotter PD, Gershin IF, Desnick RJ: Physical mapping of the human connexin 40 (GJA5), flavin-containing monooxygenase 5, and natriuretic peptide receptor A genes on 1q21. *Genomics* 39: 409-411, 1997.

15. Iglesias A, McCurdy LD, Glass IA, Cotter PD, Illueca M, Perenyi A, Sansaricq C: Mosaic trisomy 14 with hepatic involvement. *Ann. Génét.* 40: 104-108, 1997.

16. Davies JP, Cotter PD, Ioannou YA: Cloning and mapping of human Rab7 and Rab9 cDNA sequences and identification of a Rab9 pseudogene. *Genomics* 41: 131-134, 1997.

17. Abad MM, Cotter PD, Fodor FH, Larsen S, Ginsberg-Fellner F, Desnick RJ, Abdenur JE: Screening for the mitochondrial DNA A3243G mutation in children with insulin-dependent diabetes mellitus. *Metabolism* 46: 445-449, 1997.

18. Cotter PD, Kaffe S, McCurdy LD, Jhaveri M, Willner JP, Hirschhorn K: Paternal heterodisomy for chromosome 14: a case report and review. *Am. J. Med. Genet.* 70: 74-79, 1997.

19. Kang JS, Gao M, Feinleib JL, Cotter PD, Guadagno SN, Krauss RS: CDO: an oncogene-, serum- and anchorage-regulated member of the Ig/fibronectin type III repeat family. *J. Cell Biol.* 138: 203-213, 1997.

20. Cotter PD, McCurdy LD, Gershin IF, Babu A, Willner JP, Desnick RJ: Prenatal detection and molecular characterization of a de novo duplication of the distal long arm of chromosome 19. *Am. J. Med. Genet.* 71: 325-328, 1997.

21. Cotter PD, Babu A, McCurdy LD, Caggana M, Willner JP, Desnick RJ: Homozygosity for pericentric inversions of chromosome 9: prenatal diagnosis of two cases. *Ann. Génét.* 40: 222-226, 1997.

22. Ashton-Prolla P, Gershin IF, Babu A, Neu RL, Zinberg RE, Willner JP, Desnick RJ, Cotter PD: Prenatal diagnosis of a familial interchromosomal insertion of Y chromosome heterochromatin. *Am. J. Med. Genet.* 73: 470-473, 1997.

23. Manea SR, Gershin IF, Babu A, Willner JP, Desnick RJ, Cotter PD: Mosaicism for a small supernumerary ring X chromosome in a dysmorphic, growth retarded male: mos47,XXY/48,XXY,+r(X). *Clin. Genet.* 52: 432-435, 1997.

24. Cotter PD, Hirschhorn K: Chimerism detected by an unbalanced chromosome translocation: an alternative hypothesis. *Clin. Genet.* 53: 230, 1998.

25. Glass IA, Stormer P, Oei PTSP, Hacking E, Cotter PD: Trisomy 2q11.2→q21.1 resulting from an unbalanced insertion in two generations. *J. Med. Genet.* 35: 319-322, 1998.

26. Cotter PD, Babu A, Willner JP, Desnick RJ: Prenatal diagnosis and outcome of mosaicism for a *de novo* unbalanced translocation detected in amniocytes. *Prenat. Diagn.* 18: 857-861, 1998.

27. Cotter PD, May A, Li L, Al-Sabah AI, Fitzsimons EJ, Cazzola M, Bishop DF: Four new mutations in the erythroid–specific 5-aminolevulinate synthase (*ALAS2*) gene causing X-linked sideroblastic anemia: increased pyridoxine responsiveness after removal of iron overload by phlebotomy and coinheritance of hereditary hemochromatosis. *Blood* 93: 1757-1769, 1999.

28. Bigler SA, Bigler LR, Kaelbling M, Cotter PD: Characterization of two distinct kidney tumors in a patient with end-stage renal disease. *J. Urol. Pathol.* 11: 133-142, 1999.

29. Cotter PD, Ledesma CT, Dietz LG, Pusso S, Wohlferd MM, Goldberg JD: Prenatal diagnosis of supernumerary marker 15 chromosomes and exclusion of uniparental disomy for chromosome 15. *Prenat. Diagn.* 19: 721-726, 1999.

30. McGhee EM, Cohen NR, Wolf JL, Ledesma CT, Cotter PD: Monosomy 16 as the sole abnormality in myeloid malignancies. *Cancer Genet. Cytogenet.* 118: 163-166, 2000.

31. Glass IA, Trenholme A, Mildenhall L, Bailey RJ, Cotter PD: Mild phenotype in two siblings with distal monosomy 12p13.31→pter. *Clin. Genet.* 57: 401-405, 2000.

32. Rauen KA, Cotter PD, Bitts SM, Cox VA, Golabi M: Cardio-facio-cutaneous syndrome in an individual with an interstitial deletion of 12q: identification of a candidate region for CFC syndrome. *Am. J. Med. Genet.* 93: 219-222, 2000.

33. McGhee EM, Klump CJ, Bitts SM, Cotter PD, Lammer EJ: Candidate region for Coffin-Siris syndrome at 7q32→34. *Am. J. Med. Genet.* 93: 241-243, 2000.

34. Zneimer SM, Cotter PD, Stewart SD: Telomere-telomere (end to end) fusion of chromosomes 7 and 22 with an interstitial deletion of chromosome 7p11.2→p15.1: phenotypic consequences and possible mechanisms. *Clin. Genet.* 58: 129-133, 2000.

35. Rowe AG, Abrams L, Qu Y, Chen E, Cotter PD: Tetrasomy 15q25→qter: cytogenetic and molecular characterization of an analphoid supernumerary marker chromosome. *Am. J. Med. Genet.* 93: 393-398, 2000.

36. Govberg IJ, Wolf JL, Cotter PD: Acute myeloid leukemia with trisomy 4 and double minutes: further evidence that double minutes can occur as the primary cytogenetic abnormality. *Cancer Genet. Cytogenet.* 121: 212-215, 2000.

37. Cotter PD, Ko E, Larabell SK, Rademaker AW, Martin RH: Segregation of a supernumerary del(15) marker chromosome in sperm. *Clin. Genet.* 58: 488-492, 2000.

38. Xuan WC, Han B, Zhang X, Li X, Sun L, Guo C, Cotter PD: Study of complicated chromosomal minute structure aberration by FISH. *Chinese Journal of Birth Health and Heredity* 2000: 4-5, 2000.

39. Cotter PD, Musci TJ: Interphase FISH with chromosome-specific protelomere probes for rapid prenatal diagnosis in a reciprocal translocation carrier. *Prenat. Diagn.* 21: 171-175, 2001.

40. McGhee EM, Qu Y, Wohlferd MM, Goldberg JD, Norton ME, Cotter PD: Prenatal diagnosis and characterization of a whole arm translocation resulting in monosomy for 18p. *Clin. Genet.* 59: 274-278, 2001.

41. Lammer EJ, Punglia DR, Fuchs AE, Rowe AG, Cotter PD: Inherited duplication of Xq27.2→qter: phenocopy of infantile Prader-Willi syndrome. *Clin. Dysmorphol.* 10: 141-144, 2001.

42. Rauen KA, Cotter PD: Candidate region for cardio-facio-cutaneous syndrome. *Am. J. Med. Genet.* 101: 173, 2001.

43. Chen E, Cotter PD, Cohen RA, Lachman RS: Characterization of a long-term survivor with Stüve-Wiedemann syndrome and mosaicism of a supernumerary marker chromosome. *Am. J. Med. Genet.* 101: 240-245, 2001.

44. Cotter PD, Kaffe S, Li L, Gershin IF, Hirschhorn K: Loss of subtelomeric sequence associated with a terminal inversion duplication of the short arm of chromosome 4. *Am. J. Med. Genet.* 102: 76-80, 2001.

45. Skinner JL, Govberg IJ, DePalma RT, Cotter PD: Heteromorphisms of chromosome 18 can obscure detection of fetal aneuploidy by interphase FISH. *Prenat. Diagn*. 21: 702-704, 2001.

46. Tung G, Covert SM, Malabed KL, Wohlferd MM, Beckerman KP, Goldberg JD, Cotter PD: Minute supernumerary marker chromosomes identified in two patients with a larger, related pseudodicentric chromosome. *Am. J. Med. Genet.* 103: 193-197, 2001.

47. Rauen KA, Bitts SM, Li L, Golabi M, Cotter PD: Tandem duplication mosaicism: characterization of a mosaic dup(5q) and review. *Clin. Genet.* 60: 367-371, 2001.

48. Rauen KA, Albertson DG, Pinkel D, Cotter PD: Additional patient with del(12)(q21.2q22): further evidence for a candidate region for cardio-facio-cutaneous syndrome? *Am. J. Med. Genet.* 110: 51-56, 2002.

49. Kostiner DR, Nguyen H, Cox VA, Cotter PD: Stabilization of a terminal inversion duplication of 8p by telomere capture of 18q. *Cytogenet. Genome Res.* 98: 9-12, 2002.

50. Rauen KA, Golabi M, Cotter PD: Fertility in a female with mosaic trisomy 8. *Fertil. Steril.* 79: 206-208, 2003.

51. Rauen KA, Cotter PD: Cardio-facio-cutaneous syndrome phenotype and del(12q). *Am. J. Med. Genet.* 116A: 411-412, 2003.

52. Sheen VL, Wheless JW, Bodell A, Braverman E, Cotter PD, Rauen KA, Glenn O, Weisiger K, Packman S, Walsh CA, Sherr EH: Periventricular heteropia associated with chromosome 5p anomalies. *Neurology* 60: 1033-1036, 2003.

53. Dietz LG, Wylie AA, Rauen KA, Murphy SK, Jirtle RL, Cotter PD: Exclusion of maternal uniparental disomy of chromosome 14 in patients referred for Prader-Willi syndrome using a multiplex methylation polymerase chain reaction assay. *J. Med. Genet*. 40: e46, 2003.

54. Tartaglia M, Cotter PD, Zampino G, Gelb BD, Rauen KA: Exclusion of *PTPN11* mutations in Costello syndrome: further evidence for distinct genetic etiologies for Noonan, cardio-facio-cutaneous, and Costello syndromes. *Clin. Genet*. 63: 423-426, 2003.

55. Moore MW, Dietz LG, Tirtorahardjo B, Cotter PD: A multiplex methylation PCR assay for identification of uniparental disomy of chromosome 7. *Hum. Mutat*. 21: 645-648, 2003.

56. Bekri S, May A, Cotter PD, Al-Sabah AI, Guo XJ, Masters GS, Bishop DF: A promoter mutation in the erythroid-specific 5-aminolevulinate synthase (*ALAS2*) gene causes X-linked sideroblastic anemia. *Blood* 102: 698-704, 2003.

57. Murphy SK, Wylie AA, Coveler KJ, Cotter PD, Papenhausen PR, Sutton VR, Shaffer LG, Jirtle RL: Epigenetic detection of human chromosome 14 uniparental disomy. *Hum. Mutat.* 22: 92-97, 2003.

58. Ki A, Rauen KA, Black LD, Kostiner DR, Sandberg PL, Pinkel D, Albertson DG, Norton ME, Cotter PD: Ring 21 chromosome and a satellited 1p in the same patient: novel origin for an ectopic NOR. *Am. J. Med. Genet.* 120A: 365-369, 2003.

59. Cotter PD, Musci TM, Norton ME: Rapid prenatal diagnosis in translocation carriers by interphase FISH with chromosome-specific subtelomere probes. *Am. J. Med. Genet.* 122A: 1-5, 2003.

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OTHER PUBLICATIONS:

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INVITED PRESENTATIONS:

1. Clinical cytogenetic referrals as a research resource. 58th Annual Meeting of the California Association for Medical Laboratory Technology, Santa Clara, CA, October 3-7, 1997.

2. Prenatal diagnosis and rare chromosomal anomalies: an approach to risk assessment. Grand Rounds Presentation, Department of Obstetrics, Gynecology and Reproductive Sciences, University of San Francisco, San Francisco, CA, October 24, 1997.

3. The application of new technologies in prenatal diagnosis: successes and frustrations. Third Annual Conference of the Pacific Southwestern Regional Genetics Network, San Jose, CA, March 24, 2001.

4. Prenatal diagnosis of minute supernumerary marker chromosomes. Third Meeting on Prenatal Diagnosis in China, Qingdao, Shandong Province, China, October 31-November 2, 2001.

5. Beyond the karyotype: the application of new technologies to prenatal diagnosis. Third Meeting on Prenatal Diagnosis in China, Qingdao, Shandong Province, China, October 31-November 2, 2001.

6. Clinical applications of gene expression profiling. 29th Annual Meeting of the Association of Genetic Technologists, Anaheim, CA, June 17-20, 2004.

7. Non-invasive prenatal diagnosis: are we there yet? 30th Annual Meeting of the Association of Genetic Technologists, Kansas City, MO, June 16-19, 2005.

8. Array comparative genomic hybridization: applications in genetic medicine. XXXII Congreso Nacional de Genética Humana, Asociación Mexicana de Génetica Humana, Oaxaca, Oaxaca, Mexico, November 7-11, 2007.

9. New technologies in genetic testing. Grand Rounds Presentation, Department of Pediatrics, California Pacific Medical Center, San Francisco, CA, December 7, 2007.

10. Clinical applications of chromosomal microarray analysis. Grand Rounds Presentation, Childrens' Hospital and Research Center at Oakland, Oakland, CA, February 16, 2010.

11. Whole genome sequencing in the clinical laboratory. Annual Conference of the Pacific Southwestern Regional Genetics Network, Duarte, CA, April 9, 2011.

12. A new paradigm for personalized medicine and companion diagnostics: the Contract Diagnostics Organization. 4th International Conference on Drug Discovery and Therapy, Dubai, UAE, 12-15 February, 2012.

13. A new paradigm for advancing personalized medicine: The Contract Diagnostics Organization. Molecular Med TriCon Meeting, San Francisco, CA, February 21-23, 2012.

14. Personalized medicine, companion diagnostics and lifestyle genetics. Annual Conference of the Pacific Southwestern Regional Genetics Network, Aliso Viejo, CA, March 17, 2012.

15. Quality management in the clinical laboratory. 2nd Russian Congress on the Molecular Basis of Clinical Medicine, St. Petersburg, Russian Federation, June 18-20, 2012.

16. Personalized molecular medicine and companion diagnostics. 2nd Russian Congress on the Molecular Basis of Clinical Medicine, St. Petersburg, Russian Federation, June 18-20, 2012.

17. A new paradigm for advancing personalized medicine: The Contract Diagnostics Organization. Fourth Annual Next Generation Dx Summit, Washington DC, August 21-23, 2012.

18. Outsourcing companion diagnostics development: the role of the Contract Diagnostics Organization. World CDx Summit, Boston, MA, November 13-15, 2012.

19. Solutions for the outsourcing of companion diagnostics development. Molecular Med TriCon Meeting, San Francisco, CA, February 11-15, 2013.

20. Outsourcing companion diagnostics development: the role of the Contract Diagnostics Organization. BioPharma Asia Convention, Singapore, March 18-21, 2013.

21. The pathway to creating an in-vitro diagnostic. LEAD San Diego, San Diego, CA, August 7, 2013.

22. Outsourcing the development of companion diagnostics. Fifth Annual Next Generation Dx Summit, Washington DC, August 20-22, 2013.

23. Outsourcing companion diagnostics development: the role of the Contract Diagnostics Organization. BioPharma India Convention, Mumbai, India, November 25-26, 2013.

24. Outsourcing companion diagnostics development: the role of the Contract Diagnostics Organization. BioPharma Asia Convention, Singapore, March 10-13, 2014.

25. Regulatory considerations for Next-Gen sequencing in the clinical laboratory. Clinical Genomics Seminar, Centre for Translational Medicine, Yong Loo Lin School of Medicine, National University of Singapore, Singapore, May 20, 2014.

26. CLIA for international clinical laboratories. EOne Laboratories / EOne-Diagnomics Genome Center, Incheon, Korea, May 4, 2015.

27. Regulatory considerations for Next-Gen sequencing in the clinical laboratory. Center for Genomic Medicine, Riken Genesis, Yokohama, Japan, June 2, 2015.

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29. Regulatory Considerations for the Validation of NGS Assays in the Clinical Laboratory. Illumina's Next Generation Sequencing in the Clinic, Long Beach, CA, August 26, 2015.

INTERVIEWS:

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DIGITAL MEDIA:

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ABSTRACTS / MEETING PRESENTATIONS:

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13. Bishop DF, Cotter PD, May A, Hoffbrand AV, Hofferbert S: A novel mutation in exon 9 of the erythroid 5-aminolevulinate synthase gene shows phenotypic variability leading to X-linked sideroblastic anemia in two unrelated male children but to the late-onset form of this disorder in an unrelated female. 46th Annual Meeting of the American Society of Human Genetics, San Francisco, CA, October 29-November 2, 1996. *Am. J. Hum. Genet.* 59: A248, 1996.

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34. McGhee EM, Qu Y, Cotter PD: Prenatal diagnosis and characterization of an unbalanced whole arm translocation, der(18;22)(q10;q10), resulting in monosomy for 18p. 50th Annual Meeting of the American Society of Human Genetics, Philadelphia, PA, October 3-7, 2000. *Am. J. Hum. Genet.* 67: A161, 2000.

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36. Rauen KA, Albertson DG, Pinkel D, McCormick F, Cotter PD: The search for submicroscopic deletions in patients with CFC syndrome using high resolution CGH microarrays. XXII David W. Smith Workshop on Malformations and Morphogenesis. Lake Arrowhead, CA, September 7 - 12, 2001.

37. Rauen KA, Albertson DG, Cotter PD: An additional patient with del(12)(q21.2q22): Further evidence for a candidate region for cardio-facio-cutaneous syndrome? 51st Annual Meeting of the American Society of Human Genetics, San Diego, CA, October 12-16, 2001. *Am. J. Hum. Genet.* 69: A295, 2001.

38. Wylie AA, Cotter PD, Wiliford B, Papenhausen PR, Jirtle RL: Methylation analysis of maternal and paternal uniparental disomy of human chromosome 14. 51st Annual Meeting of the American Society of Human Genetics, San Diego, CA, October 12-16, 2001. *Am. J. Hum. Genet.* 69: A587, 2001.

39. Friedman BD, Lester SM, Dietz LG, Cotter PD: Prader-Willi Syndrome (PWS) in two siblings with initially disparate FISH results: Implications for diagnosis and recurrence risk counseling. American College of Medical Genetics and March of Dimes Birth Defects Foundation Annual Clinical Genetics Meeting, New Orleans, LA, March 14-17, 2002. *Genet. Med.* 4: A191, 2002.

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European Human Genetics Conference, Prague, Czech Republic, May 7-10, 2005.

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101. Shankar SP, Aras M, Cotter PD, Slavotinek AM: Partial chromosome 22q duplication resulting in congenital diaphragmatic hernia and multiple congenital anomalies. American College of Medical Genetics 15th Annual Clinical Genetics Meeting, Phoenix, AZ, March 12-16, 2008.

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103. Iglesias, A, Macera M, Cotter PD, Cohen F, Breshin J, Babu A: Molecular cytogenetic and microarray characterization of a de novo der(Y)t(X;Y)(q27.1;q12) in a male with mental retardation, hypotonia, intractable seizure disorder and dysmorphic features. 58th Annual Meeting of the American Society of Human Genetics, Philadelphia, PA, November 11-15, 2008.

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June 16-18, 2009.

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116. McCarthy D, Cotter PD, Hanna MM: Abscription based, bisulfite-free detection of CpG methylation: application to imprinting disorders. Cambridge Healthtech Institute's Second Annual X-Gen Congress & Expo, San Diego, CA, March 14-18, 2011.

117. Hambuch TM, Laurent M, Sickler B, Liao A, Cotter PD, Jain S, Lyan Y, Bernd J, O'Daniel JM, Bentley DR: Analyses of quality and quantity for whole genome sequencing in a clinical laboratory. American College of Medical Genetics 18th Annual Clinical Genetics Meeting, Vancouver, British Columbia, Canada, March 16-20, 2011.

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119. Bischoff FZ, Pircher TJ, Pham T, Wong K, Mikolajczyk S, Cotter PD, Mayer JA: Redefining CTCs: Detection of additional circulating tumor cells using an antibody capture cocktail and HER2 FISH. American Society of Clinical Oncology, Chicago, IL, June 3-7, 2011.

120. Mayer JA, Pham T, Wong KL, Scoggin EJ, Sales EV, Clarin T, Pircher TJ, Mikolajczyk SD, Cotter PD, Bischoff FZ: Efficiency of a laboratory developed HER2 FISH test on circulating tumor cells. San Antonio Breast Cancer Symposium, San Antonio, TX, December 6-10, 2011.

121. Houtchens K, ten Bosch J, Graham K, Morin S, Vinson M, Cotter PD, Gibbons R, Hoppe C: A complex phenotypic picture in a complex genetic background – results from a multi-pronged approach. Association for Molecular Pathology Annual Meeting, Long Beach, CA, October 25-27, 2012.

122. Verma S, Cotter PD, Moore MW: Optimization of a q-RTPCR assay for identification of multiple bcr-abl1 transcripts from degraded RNA specimens. Association for Molecular Pathology Annual Meeting, Long Beach, CA, October 25-27, 2012.

123. Ajay SS, Hambuch T, Lee H, Deignan J, Jain S, Lyan Y, Hurless, J Cotter PD: Ensuring the quality of genomic sequencing

in the clinic: a model for inter-laboratory proficiency testing. American College of Medical Genetics Annual Clinical Genetics Meeting, Phoenix, AZ, March 19-23, 2013.

124. Volchek I, Cotter PD, Petrov A, Moore MW: Business analysis and market analysis and market potential of drug screening technology for preventive and personalized therapy. European Association for Predictive, Preventive and Personalized Medicine World Congress 2013, Brussels, Belgium, September 20-21, 2013.

125. Phin S, Moore MW, Cotter PD: *PTEN* Del-Tect™ four color FISH assay shows improved specificity and sensitivity for the detection of genomic *PTEN* deletion over the standard dual probe. Association for Molecular Pathology Annual Meeting, Phoenix, AZ, November 14-16, 2013.

126. Verma S, Cotter PD, Moore MW: Detection of multiple BCR-abl1 transcripts from degraded RNA samples. Association for Molecular Pathology Annual Meeting, Phoenix, AZ, November 14-16, 2013.

127. Kovaleva NV, Cotter PD: Gonadal mosaicism for structural autosomal rearrangements with non-centromeric breakpoints: data from 76 carriers suggests male-specific selection against abnormal cell lines and association of clinical manifestation with a high proportion of abnormal cells. European Human Genetics Conference, Milan, Italy, May 31 – June 3, 2014.

128. Ta M, Cotter PD, Moore MW: An approach for detecting structural variations from NGS paired end reads using Split Reads, Discordant Read Pairs and Local Alignment. Galaxy Community Conference, Baltimore, MD, June 30-July 2, 2014.

129. Moore, MW, Verma S, Cotter PD: A novel q-RTPCR assay for identification of multiple bcr-abl1 transcripts from degraded RNA specimens. The 3rd World Congress on Controversies in Hematology, Istanbul, Turkey, September 11-13, 2014.

130. Gerber A, Clarin TC, Bhandari A, Sero V, Bolognesi C, Medoro G, Manaresi N, Moore MW, Cotter PD, Bischoff: Isolation and analysis of pure intact tumor cell populations from FFPE: implications for more precise HER2 FISH testing in breast cancer. San Antonio Breast Cancer Symposium, San Antonio, TX, December 8-12, 2015.

131. Kovaleva N, Cotter PD: Mosaicism for structural non-centromeric autosomal rearrangements in ill-defined carriers: sex differences in the rearrangements profile and evidence for maternal age effect on post-zygotic stability of female genomes. European Human Genetics Conference 2016, Barcelona, Spain, May 21-24, 2016.

132. Gerber A, Clarin TC, Bhandari A, Medoro G, Manaresi N, Moore M, Cotter PD, Bolognesi C, Bishoff F: Image- based collection allows isolation and analysis of pure intact tumor cell populations from FFPE: implications for more precise HER2 FISH analysis. American Society of Clinical Oncology, Chicago, IL, June 3-7, 2016.

133. Sims CL, Govender S, Khurana AK, Morales N, Moore MW, Cotter PDC, Gunn S: DNA-based microarray CGH as a high-resolution technique for HER2 equivocal breast cancer. Association of Genetic Technologists 41st Annual Meeting, Garden Grove, CA, June 23-25, 2016.

134. Khurana A, Verma S, Cotter PD, and Moore MW: Novel four-color FISH probe set for simultaneous detection of RET break-apart and PDGFRα amplification. Association of Genetic Technologists 41st Annual Meeting, Garden Grove, CA, June 23-25, 2016.

135. Ta M, Khurana A, Cotter PD, Moore MW: Utilizing Galaxy for clinical NGS sequence interpretation. Association of Genetic Technologists 41st Annual Meeting, Garden Grove, CA, June 23-25, 2016.

136. Ta M, Cotter PD, Moore MW: A Galaxy workflow for the generation of synthetic FASTQ samples. Galaxy Community Conference, Bloomington, IN, June 25-29, 2016.