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Volume 3, Number 1

Welcome to the April 2011 PreventionGenetics Newsletter. In this issue, we present new DNA sequencing tests for 40 disorders. In addition, we introduce two new geneticists to our staff. In the President's Corner, Dr. Jim Weber discusses recent progress at PreventionGenetics.

New Tests at PreventionGenetics Please follow the gene links for the corresponding test description.

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- Achondrogenesis ([SLC26A2](#))
- Achondrogenesis Type II-Hypochondrogenesis ([COL2A1](#))
- Amyotrophic Lateral Sclerosis and Primary Open-Angle Glaucoma ([OPTN](#))
- Atelosteogenesis ([SLC26A2](#))
- Camurati-Engelmann Disease ([TGFB1](#))
- Cartilage-hair Hypoplasia and Related Disorders ([RMRP](#))
- Chediak-Higashi Syndrome ([LYST](#))
- Chondrodysplasia Punctata, X-Linked Dominant ([EBP](#))
- Cleidocranial Dysplasia ([RUNX2](#))
- Cranioectodermal Dysplasia 1 ([IFT122](#))
- Diastrophic Dysplasia ([SLC26A2](#))
- Dilated Cardiomyopathy and Limb-Girdle Muscular Dystrophy Type 2F ([SGCD](#))
- Dentinogenesis Imperfecta ([DSPP](#))
- Ellis-van Creveld Syndrome ([EVC](#), [EVC2](#))
- Emery-Dreifuss Muscular Dystrophy-1 ([EMD](#))
- Fanconi Anemia ([FANCL](#))
- Hennekam Lymphangiectasia-Lymphedema ([CCBE1](#))
- Hereditary Breast Cancer ([CHEK2](#))
- Hermansky Pudlak Syndrome ([HPS1](#), [HPS2/AP3B1](#), [HPS3](#), [HPS4](#), [HPS5](#), [HPS6](#), [HPS7/DTNBP1](#), [HPS8/BLOC1S3](#))
- Hirschsprung Disease ([RET](#))
- Holt-Oram Syndrome ([TBX5](#))
- Kneist Dysplasia ([COL2A1](#))
- Lynch Syndrome ([PMS2](#))
- Menkes Disease and X-Hereditary Motor Neuropathy ([ATP7A](#))
- Multiple Endocrine Neoplasia Type 1 ([MEN1](#), [CDKN1B](#))
- Multiple Endocrine Neoplasia Type 2 ([RET](#))
- Multiple Epiphyseal Dysplasia ([SLC26A2](#))
- Nephronophthisis-Like Nephropathy-1 ([XPNPEP3](#))
- Neutropenia ([G6PC3](#))

Ovarian Dysgenesis Type 1 and Ovarian Hyperstimulation Syndrome ([FSHR](#))
Papillon-Lefevre Syndrome (Periodontitis) ([CTSC](#))
PLP1-Related Disorders - Pelizaeus-Merzbacher Disease and Spastic Paraplegia 2 ([PLP1](#))
Pulmonary Arterial Hypertension ([BMPR2](#))
Primary Ciliary Dyskinesia ([DNAH5](#), [DNAH11](#), [DNAI1](#), [DNAI2](#), [RSPH4A](#), [RSPH9](#), [KTU](#), [TXNDC3](#), [LRRC50](#))
Trimethylaminuria ([FMO3](#))
Usher Syndrome Type 2 ([USH2A](#))
Usher Syndrome Type 3 ([CLRN1](#))
X-linked Retinitis Pigmentosa and X-linked Primary Ciliary Dyskinesia ([RPGR](#))

Prevention Genetics proudly announces the addition of two new geneticists to our staff.

Introducing Margaret Chen, PhD, FACMG, CGC



Dr. Chen has been working in the field of genetics for more than 15 years and has held positions in academia, industry, and the non-profit sector. Her experience includes positions with Michigan State University, Athena Diagnostics, and the Alliance of Genetic Support Groups.

Dr. Chen is board-certified in both clinical molecular genetics (FACMG) and genetic counseling (CGC).

She received her master's degree in 1998 through the Genetic Counseling program at the University of Colorado Health Sciences Center in Denver and her doctoral degree from the Molecular, Cellular, Developmental Biology, and Genetics graduate program at the University of Minnesota, Twin Cities, in 2005. After finishing her doctoral degree, Dr. Chen completed a clinical molecular fellowship at the University of Alabama at Birmingham.

Dr. Chen has clinical experience in pediatric, adult, and prenatal genetics and served as an assistant lab director at Athena Diagnostics from 2008 to 2010.

Her portfolio at PreventionGenetics will focus on metabolic disorders.

Introducing Ying Wang, MD, PhD

Dr. Wang joined PreventionGenetics in January, 2011. She earned her MD from Zhengzhou University, China in 2003 and her PhD in Human Genetics from the University of Maryland, Baltimore in 2008.

She completed her Clinical Molecular Genetics fellowship at Johns Hopkins University Mckusick-Nathans Institute of

Genetic Medicine at the end of 2010 and is Board-Eligible for the ACMG. Dr. Wang's lifetime career goal is to apply the exciting new discoveries in genetics to the development of diagnostics and prevention for human diseases.

Her portfolio at PreventionGenetics will focus on skeletal dysplasia. There are at least 350 different skeletal disorders known to date.

PRESIDENT'S CORNER

Jim Weber, PhD
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Progress at PreventionGenetics

In March, we began offering our 400th gene sequencing test. This represents quite a change from the original 3 gene tests we first began offering in 2005. PreventionGenetics now has one of the largest gene sequencing menu of any lab in North America.

Over the last 12 months, we recruited five new PhDs to PreventionGenetics. We now have 11 doctorate-level scientists and physicians. Together, we comprise a powerful team of knowledge, accreditation and experience. Each of our doctorate staff is responsible for bringing up new tests and services within their areas of expertise (see below). In addition, they review primary data and prepare test reports for the tests within their portfolios. PreventionGenetics will continue to expand into new areas of clinical molecular genetics through the addition of new senior staff.

Khemissa Bejaoui - Developmental and vision disorders
Margaret Chen - Metabolic disorders
Michael Chicka - Blood disorders
Bruce Krawisz - Lab Director
Keith Nykamp - Cancer and infertility
Derek Pavelec - Bioinformatics and cognitive disability
David Schlesinger - Research Genomics
Marwan Tayeh - Ciliopathies, growth disorders and aCGH
Ying Wang - Skeletal disorders
James Weber - President
Thomas Winder - Neuromuscular disorders

Last year we added aCGH to our repertoire of core technologies at PreventionGenetics. This was in addition to our well established techniques of Sanger sequencing and short tandem repeat (microsatellite) analysis. Later in 2011, we will be announcing our first tests using Next Generation sequencing technology. Through all these changes, we will continue to provide the high quality

services and low prices that you've come to expect from PreventionGenetics.

Interested in a test we don't currently offer?

PreventionGenetics continues to expand our gene sequencing test menu. If you are interested in a particular test that we don't currently offer, please [contact us](#). There is an excellent chance we can develop a test to suit your needs.

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