



Volume 2, Number 3

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Welcome to the August, 2010 PreventionGenetics Newsletter.

In this issue, we introduce our new aCGH test for clinical specimens and for research. We also present new DNA sequencing tests for Mucopolysacchardiosis, Marfan and Marfan-like syndromes, Lissencephaly, Lynch syndrome, ARC syndrome, 3-methylglutaconic aciduria, and Congenital Generalized Lipodystrophy Types 1 & 2. In the President's Corner, Dr. Jim Weber discusses confidentiality of patient information at PreventionGenetics.

Introducing our new aCGH clinical test

PreventionGenetics is pleased to offer Array Comparative Genomic Hybridization (aCGH) for the clinical detection of deletion and amplification mutations within a targeted collection of genes.

PreventionGenetics' high density gene-centric array is a custom designed oligonucleotide array with 720K probes. This array targets a collection of over 300 genes for which we offer sequencing tests. The array will be periodically expanded to cover new genes that are added to our sequencing test menu.

This test is not designed for whole-genome scanning for allele copy number variations. Instead, the test detects copy number changes within a selected set of genes. Applications include testing of genes for which deletion mutations are particularly common and testing of genes in patients affected with recessive disorders for which only one likely causative mutation is identified by sequencing.

Our aCGH test utilizes Roche NimbleGen technology. As a research service, we are pleased to generate data using customer-supplied samples for any standard array product available from NimbleGen, including whole genome arrays. We will also work with customers to produce custom-designed arrays.

New Tests at PreventionGenetics

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Mucopolysacchardiosis

MPS I (IDUA)

MPS II (<u>IDS</u>)

MPS IIIA (SGSH)

MPS IIIB (NAGLU)

MPS IIIC (HGSNAT)

MPS IIID (GNS)

For more information on these disorders, please contact Khemissa Bejaoui, PhD, by <u>email</u> or at 715-387-0484 ext. 119.

Congenital Generalized Lipodystrophy

Type 1 (AGPAT2)

Type 2 (BSCL2)

3-methylglutaconic aciduria (AUH)

ARC syndrome (VIPAR & VPS33B)

For more information on these disorders, please contact Tom Winder, PhD, FACMG by <u>email</u> or at 715-387-0484 ext. 118.

Marfan and Marfan-like syndromes (FBN1, FBN2,

TBFBR1, TBFBR2, & MED12)

Lissencephaly (DCX, PAFAH1B1, & TUBA1A)

For more information on these disorders, please contact Marwan Tayeh, PhD, by email or at 715-387-0484 ext. 108.

Lynch syndrome (MLH1 & MSH2)

For more information on these disorders, please contact Keith Nykamp, PhD, by email or at 715-387-0484 ext. 140.

Come see us in Washington DC, Nova Scotia, and Dallas!

We are happy to be exhibiting this fall at the American Society for Human Genetics (ASHG) 60th annual meeting in Washington DC (Booth #1207), at the Annual Scientific Meeting of the Canadian College of Medical Geneticists (CCMG) in Halifax, Nova Scotia, and at the Annual Meeting of the National Society of Genetic Counselors (NSGC) in Dallas, TX (Booth #404). Please stop by our booths and say hello!

Volume Discounts

In addition to our industry-leading low pricing for clinical DNA testing, we also offer volume-based discounts. For additional information, please contact Dave Schlesinger by phone: 715-387-0484, extension 124, or via email: dave.s@preventiongenetics.com.

Interested in a test we don't currently offer?

PreventionGenetics offers one of the largest gene

sequencing test menus in the industry; however if you are interested in a test we don't currently offer, please contact us. With our experienced R&D staff, it is likely we would be able to quickly accommodate your request.

PRESIDENT'S CORNER

Jim Weber, PhD

Privacy of patient information has always been a top priority at PreventionGenetics (see <u>Genetic Ethical Principles</u>). Each employee is required to sign a Confidentiality Agreement annually. This agreement includes provisions which limit access to patient identifiers. Specimens and data are never labeled during the testing process other than with our internal test codes. The Agreement also includes a pledge to never discuss patient results with anyone who doesn't have a right to know.

Another way to ensure the privacy of patient information is through our internal computer networks. When PreventionGenetics was first started, we established two separate computer networks, with separate wiring, servers, workstations and printers. Our Office (public) network is connected to the internet and does not contain any identifiable patient information (except transiently in a very limited number of cases). Our Laboratory (private) network is not connected to the internet and is used for all laboratory processes and to store all test data and reports. It has cost us considerable trouble and expense to establish and maintain two separate computer networks; however we feel this approach provides an additional level of protection for patients.

Even with such precautions, PreventionGenetics is continually working to improve the security of patient specimens and information. And while no system is perfect, we believe the measures we currently have in place provide excellent peace of mind for patients and providers.

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