Put us to the test.

PreventionGenetics
Disease prevention through genetic testing

Volume 2, Number 5

IN THIS ISSUE

QUICK LINKS

Our Website

Requisition Form

Join Our Mailing List!

Happy Holidays and welcome to the December 2010 PreventionGenetics Newsletter. In this issue, we present new DNA sequencing tests for several Short Stature disorders, Fanconi Anemia, Thrombotic Thrombocytopenic Purpura (TTP), Juvenile Polyposis Syndrome, Peutz-Jeghers Syndrome, Li-Fraumeni Syndrome, Male Infertility and Usher Syndrome. In addition, we outline the services offered by our newly renamed Research Genomics division. In the President's Corner, Dr. Jim Weber discusses FDA regulation of genetic testing.

We also take this opportunity to thank our many customers and colleagues for their patronage in 2010. We enjoyed working with you this past year, and we very much look forward to helping you serve your patients in 2011.

New Tests at PreventionGenetics

Please follow the gene links for the corresponding test description.

Idiopathic short stature genes (SHOX, GHSR, GHR, & LHX4)
Spondylo-Meta-Epiphyseal Dysplasia, Short Limb-Hand Type (SMED-SL) (DDR2)
For more information on these disorders, please contact Marwan Tayeh, PhD, by email or at 715-387-0484 ext. 108.

Thrombotic Thrombocytopenic Purpura (TTP) (ADAMTS13)
Fanconi Anemia (FANCA, FANCC,FANCE, FANCF, FANCG, FANCJ/BRIP1, FANCM, & FANCN/PALB2)
For more information on these disorders, please contact Michael Chicka, PhD, by email or at 715-387-0484 ext. 141.

Juvenile Polyposis Syndrome (SMAD4 & BMPR1A)
Peutz-Jeghers Syndrome (STK11)
Li-Fraumeni Syndrome (TP53)
Male Infertility (AURKC)
For more information on these disorders, please contact Keith Nykamp, PhD, by email or at 715-387-0484 ext. 140.

Usher Syndrome Type 1 (MYO7A, CDH23, PCDH15, USH1C, & panel test)
For more information on these disorders, please contact Khemissa Bejaoui, PhD, by email or at 715-387-0484 ext. 119.

New Research Genomics Services

PreventionGenetics is pleased to announce the expansion of our Research Genomics (formerly Research Genotyping) services. The reason for the name change is to more accurately reflect the services we offer.

We specialize in custom genotyping projects for up to 250 SNPs with virtually unlimited sample capacity. We have developed Ancestry Informative Marker (AIM) panels for determining geoancestry. In addition to SNP genotyping, we are one of the few laboratories that offer custom STRP (microsatellite) genotyping. This includes genome-wide polymorphism scans.

In addition to our genotyping services PreventionGenetics offers array comparative genomic hybridization (aCGH) services based on the NimbleGen platform. We offer processing and analysis of any of the off-the-shelf arrays from NimbleGen, including Whole-Genome Exon-Focused, Chromosome-Specific, CNV, and Cytogenetics arrays. We can also design and utilize custom arrays tailored for more specific research needs.

And for 2011 we will be introducing next generation sequencing services based on Illumina's platform (GAIIx). We will have the capacity to produce up to 100Gb (gigabases) of sequence data per run (single read, paired-end reads, or mate-pair reads). We will also provide services for developing custom capture arrays for targeted resequencing experiments.

For more information about these Research Genomics services, please contact Dave Schlesinger, PhD, by email or phone: 715-387-0484 ext. 124.

PRESIDENT'S CORNER

Jim Weber, PhD

My topic for this Newsletter is FDA regulation of clinical DNA testing. In general, I'm a big fan of the FDA. There is no doubt that since passage of the Food and Drug Act in 1906, the FDA (and earlier versions of this federal government agency) has done a marvelous job of improving the safety and efficacy of our foods and medicines. However, I am quite concerned about proposals to expand

FDA regulation of clinical DNA testing.

On the surface, increased regulation of clinical DNA tests sounds great. We all want more powerful, accurate and useful testing. However, when one looks carefully at the ramifications of increased regulation, very quickly all kinds of problems arise. I am convinced that patients would be greatly harmed by increased regulation.

Clinical DNA testing is still in its infancy and is evolving rapidly. Hundreds, if not thousands, of new tests are added each year. Testing is moving quickly from assays for individual mutations like Factor V Leiden, to full gene tests, to tests for gene clusters, to genome-wide tests like for example aCGH, whole exome and even whole genome sequencing. The quality of this testing is improving by leaps and bounds. Nearly all current tests are "laboratory-developed". There are very few commercial test kits, and only a fraction of these have FDA approval.

In its most Draconian form, FDA regulation could bring this amazing progress almost to a standstill. A lengthy and expensive (up to $100,000 per application) FDA pre-approval process would mean that minor changes to tests would be impractical. Revenue for many DNA tests for ultra-rare disorders is very low (only a few thousand dollars annually). FDA regulation could make many of these tests economically unfeasible. Even for tests that are still available, the regulatory costs would have to be passed on to the patients. Test prices would likely increase several fold. It's also unclear if the FDA has the resources to rapidly process applications for thousands of new genetic tests.

It is important to keep in mind that clinical labs are already tightly regulated through the Clinical Lab Improvement Amendments (CLIA), passed by Congress in 1988. There are literally hundreds of rules and regulations with which PreventionGenetics and other CLIA-accredited labs must comply. CLIA labs are also subject to regular external, unannounced inspections and proficiency testing.

In the real world, we also need to realize that many government employees have an intrinsic self interest in expanding regulation. Expanded regulation means more job security, better pay, and additional support staff. I am leery of all proposals for increased regulation that come solely from Washington. The original Food and Drug Act of 1906 was spurred by broad public outcry over the dangers of unsafe foods and medicines. I hear no such outcry today about clinical DNA testing.

With the rapid advances in clinical DNA testing, the dream of the Human Genome Project to incorporate genetics

routinely into health care and to substantially improve the welfare of all patients is in sight. The FDA should not tie our hands and prevent us from reaching this goal as rapidly as possible.

Interested in a test we don't currently offer?

PreventionGenetics already offers one of the largest gene sequencing test menus, and we continue to add new tests. If you are interested in a test we don't currently offer, please contact us. There is a good chance we will develop the test.

Volume Discounts

In addition to our industry leading low pricing for clinical DNA testing, we offer volume based discounts. For more information, contact David Schlesinger by phone: 715-387-0484 ext. 124 or email.

PreventionGenetics | 3700 Downwind Drive | Marshfield | WI | 54449