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Volume 4, Number 3

Welcome to the October 2012 PreventionGenetics Newsletter. In this issue, we introduce our new web site and also Christina Zaleski, our first genetic counselor. We also describe our Rhode Island state license and 35 new Sanger sequencing tests. In the President's corner, Dr. Jim Weber continues his discussion of the storage of patient DNA sequences in electronic medical records.

PreventionGenetics Unveils its New Website

Our business is growing, our services are evolving and our markets are expanding. Our new website will help our clients and staff keep up with all these changes.

The new website is a complete overhaul of the old. An enormous amount of work over the last several months went into transforming the site. The software and data behind the site were completely reworked. The result is not just a new look, but more importantly, greatly improved search capabilities. These changes will allow our clients to find the test information they need faster with less effort.

In particular, our test menu may now be easily searched by:
* Gene
* Disease
* Test Name
* Test Number

Previously, our clients needed to scroll through long lists of tests to find the one they wanted. Now, just a quick, simple search will bring them the information they seek. Our search engine will accept synonyms for genes and diseases. In near future, we will add search capability for categories of test, for example all tests involved in muscle or heart disease. In the more distant future, we plan to add

advanced search capabilities for clinical features.

Our website staff is dedicated to providing the best customer service on everything related to our new site. The content will continuously be updated with information on new tests, and new features will gradually be added. Check back often to see what's new.

PreventionGenetics hires Genetic Counseling Director

Christina Zaleski, MS, CGC has been hired as the Director of Genetic Counseling and Client Services.

Christina earned her Bachelor of Science degree in biology, with a psychology minor, from the University of Wisconsin-Eau Claire in 1996. She earned a Master of Science degree in human genetics/genetic counseling from Sarah Lawrence College in New York in 1999. After graduation, she continued the research work that she started as a student for Rockefeller University on mental illness in South Africa. She then returned to her home town of Marshfield, Wisconsin and has been at the Marshfield Clinic as lead genetic counselor for 13 years. Christina comes to PreventionGenetics with an extensive range of experience in genetic counseling, leadership, and a passion for service development and teaching. She has practiced in many areas including prenatal, pediatric, cancer, specialty clinics, stillbirth evaluations, child protective team, research, teaching, administration, and public health education. Highlights of her career include receiving 2007 Leadership Award for Excellence in Community Service from the March of Dimes, and being nominated by her peers as a Master Genetic Counselor in 2010.

At PreventionGenetics Christina will organize and manage all client inquiries, and supervise Client Services Representatives and Laboratory Genetic Counselor(s) as Genetic Counseling Services expand.

When asked about her new role with PreventionGenetics, Christina stated, "I look forward to a new challenge and making a difference through a clinical testing laboratory- as genetic testing can not only establish a diagnosis, but lays the ground work for the new era of gene based treatments. It's exciting how genetic testing may ultimately prevent disability, morbidity and mortality and improve our patient's and their family's quality of life."

Christina's contact information will be:
email: christina.zaleski@preventiongenetics.com
phone: 715-387-0484 ext 158

New and updated Sanger Gene Sequencing Tests at PreventionGenetics

Please follow the gene links for the corresponding test descriptions.

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Autosomal RecessiveDyskeratosis Congenita WRAPS3 (Test #1127)
Noonan Syndrome Panel *PTPN11,SOS1, RAF1, KRAS, BRAF, WYCAS, SINCZ* (Test #1115)
Amyotrophic Lateral Sclerosis, X-Linked Dominant *UBQLN2* (Test #158)
GATA2-Related Disorders and Predisposition to Myelodysplastic Syndrome (MDS) and Acute Myeloid Leukemia *GATA2* (Test #286)
Gray Platelet Syndrome *NBEAL2* (Test #448)
Ehlers-Danlos Syndrome, Kyphoscoliotic Form *PLOD1* (Test #845)
Popliteal Pterygium Syndrome and Van der Woude Syndrome *IRF6* (Test #896)
Ehlers-Danlos Syndrome *TNXB* (Test #843)
Cornelia de Large Syndrome *NIPBL, SMC1A, SMC3* (Test #890)
Meier-Gorlin Syndrome *ORC1, ORC4, ORC6, CDT1, and CDC6* (Test #950)
Autosomal Recessive Cutis Laxa Type iia (arcl2a) and Wrinkly Skin Syndrome *ATP6V0A2 (Test #1061)*
Macrocephaly, Alopecia, Cutis Laxa and Scoliosis *RIN2 (Test #1064)*
Dyskeratosis Congenita *WRAPS3* (Test #1127)
Curtis Laxa Type 1B *EFEMP2 (*Test #1062)
Autosomal Recessive Curtis Laxa Type 3A *ALDH18A1* (Test #1063)
Dyskeratosis Congenita, Autosomal Recessive *NHP2* (Test #1124)
Dyskeratosis Congenita Autosomal Recessive *NOP10* (Test #1126)
Spondyloperipheral Dysplasia (SPD) and Platyspondylic lethal skeletal dysplasia, Torrance type COL2A1 Tier789 (Test #789)
Catecholaminergic Polymorphic Ventricular Tachycardia *RYR2* and *CASQ2* (Test #940)
Sick Sinus Syndrome and Brugada Syndrome *HCN4* (Test #947)
Isolated Nonsyndromic Congenital Heart Defects *ZFPM2* (Test #946)
Tuberous Sclerosis Complex *TSC1(Test #1001)*
Tuberous Sclerosis Complex *TSC2* (Test #1002)

Wilms Tumor *WT1* (Test #1003)
Hereditary Breast And Ovarian Cancer *BARD1*(Test #1004)
Leber Congenital Amaurosis And Retinitis Pigmentosa *RB1*(Test#795)
Ataxia-telangiectasia-like disorder *MRE11A* (Test #796)
Nijmegen Breakage Syndrome *NBN* (Test #797)
Hereditary Diffuse Gastric Cancer CDH1(Test #798)
Hereditary Paragangliama-Pheochromocytoma Syndrome *TMEM127* (Test #1136)
Hereditary Paragangliama-Pheochromocytoma Syndrome *SDHAF2* (Test #1134)
Von Hippel-Lindau Disease *VHL* (Test #1118)
Von Hippel-Lindau Disease *RSPH4A* (Test #747)
Early Onset Mopathy, Areflexia, Respiratory Distress and Dysphagia *MEGF10* (Test #922)
Gle1-Related Disorders *GLE1*(Test #590)
Myofibrillar Myopathy *CRYAB* (Test #362)
Lethal Multiple Pterygium Syndrome / Fetal Akinesia Deformation Sequence Panel Test *CHRNA1, CHRND, CHRNG, DOK7, GLE1, RAPSN* (Test #413)
Dyskeratosis Congenita Autosomal Recessive *NOP10* (Test #1126)

Rhode Island License

PreventionGenetics is now licensed as a Clinical Laboratory by the State of Rhode Island. This should make it easier for our Rhode Island colleagues to make use of our services.

Jim Weber, PhD

President's Corner

Patient Sequences in Electronic Medical Records

President's Corner
October 2012

This is the second in a series of three articles on the storage of patient DNA sequences in Electronic Medical Records (EMRs). In the first article, I emphasized the importance of placing patient sequences in EMRs for reinterpretation of the clinical significance of the sequences and to provide health care providers with crucial medical alerts. In this second article, I address sharing of sequence information electronically among family members.

If a person breaks their leg in an automobile accident and is rushed to the emergency room, that person is the "unit of treatment". The emergency room personnel (as they should) care little if at all about family history or genomic sequence of the patient. However, in clinical genetics, it is

not the individual who is the "unit of treatment", but rather the *family*. Because, of course, all individuals inherit their DNA sequences with almost no change from their parents, family members are inextricably linked genetically.

Very often, we cannot properly interpret a sequence variant in an affected patient without combining sequence and phenotypic information from family members. Here are three examples. When an affected child is compound heterozygous for a known causative sequence variant and a variant of unknown significance (VUS), we need to determine if the two variants are in cis or trans phase. This is usually accomplished by sequencing the parents. When we find a VUS in a patient affected with a dominant disorder, we need to determine if that VUS segregates with the disease in the family. If through testing of family members, we find that a VUS is the result of a *de novo* mutation, then this is usually very strong evidence in favor of pathogenicity.

In addition, there are other important applications of sharing of information among family members. Examples include identifying sequence errors, filling gaps in sequence coverage, determining haplotypes, identifying deletions, and confirming family trees.

I think that sharing of sequences and phenotypes among family members should become routine in clinical genetics. This will require, however, vastly improved interchange of EMRs among different health care providers, perhaps through regional EMR repositories. It may also require blanket consent of individuals to share their sequences and phenotypes. Here is a suggestion for such a consent statement:

"I consent through accredited health care providers to the anonymous sharing of my DNA sequences and relevant clinical features with all of my biological relatives, both close and distant."

Note the important term anonymous in the statement. There should be no need to reveal medical information of family members to patients. Similarly, there should be no need to reveal the identities of family members to health care providers.

In our next newsletter, I will present the third article in this series on the topic of communication of sequence information among family members.

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.