PreventionGenetics

Disease prevention through genetic testing

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In This Issue

New Tests at PreventionGenetics
President's Corner
Coming in April

Quick Links...

Our Website
Requisition Form
Price List

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Welcome to the February 2010 PreventionGenetics Newsletter. In this issue, we are pleased to introduce new tests for Arrhythmogenic Right Ventricular Cardiomyopathy, Nephronophthisis, Congenital Myasthenic Syndrome, Multiple Pterygium Syndromes, and Pena-Shokeir Syndrome.

The topic for the President's Corner is DNA test results and electronic health records.

New Tests at PreventionGenetics

Arrhythmogenic Right Ventricular Cardiomyopathy

PreventionGenetics is pleased to continue developing genetic tests for heart disease. Four new gene tests for Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC) are now ready. ARVC primarily affects the right ventricle. It is characterized by myocardial atrophy and fibrofatty ventricle involvement. Heart failure may result. ARVC is a heterogeneous disease that is inherited in about half the cases. The mode of inheritance is most often

autosomal dominant. Mutations in four genes encoding desmosomal proteins account (*PKP2, DSP, DSG2* and *DSC2)* for the great majority of cases with known cause (McNally et al. GeneReviews, www.genetests.org, 2009; Bhuiyan et al. Circ Cardiovasc Genet 2:418-427, 2009). PreventionGenetics offers sequencing of these genes individually or sequentially as a panel.

For more information on cardiomyopathy genetic testing, please contact Khemissa Bejaoui PhD, by email or at 715-387-0484 ext 119.

Nephronophthisis

Nephronophthisis (NPH) is the most common genetic cause of progressive renal failure in children and young adults. NPH is characterized by polyuria, growth retardation and progressive deterioration of renal function with normal or slightly reduced kidney size (Hildebrandt et al. Nat Genet 17:149-153, 1997; Hildebrandt et al. J Am Soc Nephrol 20:23-35, 2009). NPH exhibits autosomal recessive inheritance. PreventionGenetics offers sequencing of all nine NPH genes that have been identified to date (*NPHP1, INVS/NPHP2, NPHP3, NPHP4, IQCB1/NPHP5, CEP260/NPHP6, GLIS2/NPHP7, RPGRIP1L/NPHP8* and *NEK8/NPHP9*).

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

Congenital Myasthenic Syndromes

PreventionGenetics now has testing for the following disorders:

- Congenital Myasthenic Syndromes
- Fetal Akinesia Deformation Sequence (Pena-Shokeir Phenotype)
- Lethal Multiple Pterygium Syndrome
- Escobar Variant of Multiple Pterygium Syndrome
- Familial Limb Girdle Myasthenia Syndrome

The genes involved are *CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, MUSK, CHAT, AGRN, COLQ, DOK7* and *RAPSN*. Individual gene tests and panels are available.

For more information, please contact Tom Winder, PhD, FACMG by email or at 715-387-0484 ext. 118.

Come see us in Albuquerque!

We are happy to be exhibiting at the American College of Medical Genetics meeting in Albuquerque, NM. Please stop by booth #514 and say hello!

President's Corner

Jim Weber, PhD

We all know that there is a big push underway in America to expand the use of electronic health records (EHRs). Many providers already use quite sophisticated EHRs, while others are just getting started. Unfortunately, I don't know of a single EHR that is yet ready to store and utilize DNA test results.

We need to add DNA results to EHRs for at least three really important reasons. First, it will not be possible to make full use of the sequence information in health care without electronic storage. Using the information in drug prescription and in reproductive planning, for example, will only be efficient and practical when the information is readily available in the EHR. Second, our ability to interpret DNA sequencing results is improving steadily. A variant of unknown significance today may become fully interpretable in the future. If we save the sequences in the EHRs, then the sequences can be reinterpreted periodically using continuously upgraded software. Third, patients pay a lot of money for clinical DNA tests. This information should be used throughout the patient's lifetime and by their descendants even after the patient's death. If this information is not saved in the EHR, then patients or family members may need to unnecessarily pay for the same test again and again.

PreventionGenetics will be happy today to provide files of DNA test results to clients either on CD or via secure electronic transfer. We will also store this information until clients are ready to receive the data. But PreventionGenetics cannot change health care alone. It will take a unified effort by clinical geneticists

everywhere to make this happen.

With affordable whole exome and whole genome sequencing just around the corner, the dream of human geneticists to dramatically improve health care is at hand. This revolution will not happen, however, until the sequences are added to the EHRs. I invite everyone in the human genetics community to work with PreventionGenetics to make this brighter future a reality.

Coming in the April PreventionGenetics Newsletter

Our next Newsletter will focus on new additions to the PreventionGenetics test menu as well as introduction Marwan Tayeh PhD, the newest member of our senior staff. The topic for the President's Corner will be human gene patenting.

Provide the very best care for your patients. Quality, low prices, and excellent service. You get all three with PreventionGenetics.

In addition to our industry leading low pricing for clinical DNA testing, we offer volume based discounts. For more information, contact Chuck Dokken by phone at 715-387-0484 x 107 or email at chuck.dokken@preventiongenetics.com.

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