Volume 8, Number 4

Welcome to the December 2016 PreventionGenetics newsletter. In this issue, we highlight our Comprehensive Miscarriage, Stillbirth, and Neonatal Death Panel as well as our expanded kidney test menu with a recent case example. We also introduce one of our new geneticists, Dr. Diane Allingham-Hawkins. In the President’s Corner, Dr. Jim Weber discusses DNA Banking.

Finding Answers: Comprehensive Miscarriage, Stillbirth & Neonatal Death Panel

When families experience miscarriage, stillbirth, and neonatal deaths, there are often many difficult questions. Why did this happen? Will it happen again? Is there anything that could have prevented the outcome? At PreventionGenetics, we are dedicated to helping answer these questions.

Our Comprehensive Miscarriage, Stillbirth & Neonatal Death Panel was developed under collaboration between dual-certified molecular geneticist and cytogeneticist, Dr. Diane Allingham-Hawkins, and Dr. Elizabeth McPherson, experienced Medical Geneticist and Director of the Wisconsin Stillbirth Service Program, who has more than 30 years of experience evaluating stillbirths and neonatal deaths. It includes Chromosomal Microarray (CMA-ISCA) followed by the 40 gene NextGen sequencing panel in cases with a normal microarray result.

To read about genetic causes of stillbirth, our testing strategy, and more click here.

Test Details:

Comprehensive Miscarriage, Stillbirth, and Neonatal Death Panel (includes CMA) $2580

Miscarriage, Stillbirth, and Neonatal Death Sequencing Panel $1590
Recent Webinar:
Finding Answers Comprehensive Miscarriage, Stillbirth & Neonatal Death Panel 20161109

Speakers: Diane Allingham-Hawkins Ph.D., FCCMG, FACMG Elizabeth McPherson M.D.
Click here to watch recorded webinar

Expanded Kidney Test Menu - Teamwork Increases Value of Genetic Testing

We are excited to announce additions to our menu of genetic tests for kidney disorders. PreventionGenetics provides a comprehensive menu of kidney and endocrine system tests, with the same unsurpassed quality as all our tests. For a full list of our kidney tests, click here.

Performing the correct test for each patient is important to us. Below is a recent case for Alport syndrome that demonstrates teamwork between our lab, the clinician and the patient.

A patient with Alport Syndrome was in need of a kidney transplant. The clinician sent his patient's specimen to PreventionGenetics for testing and ordered the full Alport Syndrome panel along with del/dup testing. During the test preview process, Dr. Wuyan Chen and our Genetic Counseling team discovered that a pathogenic variant in the X-linked gene COL4A5 had already been identified through a research study of the patient's family.

With permission from the ordering clinician, we were able to convert the panel test to targeted testing resulting in a cost-savings of over $2000. After confirming the pathogenic variant in the affected patient, we were able to provide assurance to the family and clinician that the patient's son (potential organ donor) was not at risk for Alport's syndrome. An invasive kidney biopsy was avoided for the son, adding even more value.
By working together, we were able to provide the best outcome for the patient and his family. The patient, his family and his clinicians offered gratitude for PreventionGenetics’ outstanding customer service and utilization management. As a gold sponsor of Seattle Children’s Pediatric Laboratory Utilization Guidance Services (PLUGS®), which shares our values, we are proud to offer the same unsurpassed level of quality and service to all our patients.

Dr. Allingham-Hawkins specializes in cyto genetics, hereditary cancer syndromes and prenatal loss

Dr. Diane Allingham-Hawkins is a Laboratory Director at PreventionGenetics, LLC, in Marshfield, WI. She is dual-certified in molecular genetics and cytogenetics by the Canadian College of Medical Geneticists (CCMG). Dr. Allingham-Hawkins earned her BSc in Honors Genetics from the University of Western Ontario in 1987 and her PhD from McMaster University in 1993. She completed her post-doctoral fellowship in Molecular Genetics at the Hospital for Sick Children in Toronto, Canada in 1995. She is also a past president of the Canadian College of Medical Genetics (CCMG). Prior to joining PreventionGenetics, Dr. Allingham-Hawkins was a private genetics laboratory consultant from 2015 to 2016 and Senior Director of Genetic Test Evaluation and Technical Editing at Hayes, Inc. from 2008 to 2015.
PRESIDENT'S CORNER

James L. Weber, PhD

DNA Banking

Since the founding of our Company about thirteen years ago, PreventionGenetics has operated a clinical DNA Bank. We define DNA Banking as the long term secure storage of DNA from an individual for the purpose of future clinical DNA testing. Our Bank is still relatively small, but is steadily growing.

The rationale for DNA Banking is difficult to convey even to experienced geneticists. For the audience of this Newsletter, I’ll try my three fact approach.

Fact #1. Interpretation of sequence variants is greatly improved with clinical and sequence information from family members.

This one is really tough for lay people to understand, but human geneticists should not have a big problem. To give just one example, we prefer to sequence trios (both parents and an affected child) in exome tests because then we can detect de novo variants and compound heterozygous variants in trans and can depreciate many dominant uncertain variants (because they are found in one of the unaffected parents). With an exome sequence of just the child, there is a much lower chance of reaching a definitive diagnosis.

Fact #2. When a person is buried or cremated, it is difficult or impossible to obtain a suitable DNA specimen.

Fact #3. When properly extracted and stored, DNA is stable for at least centuries.

These three facts together make a truly compelling case for DNA Banking. Banking is not something you do for yourself, but rather for your family. DNA Banking has similarities to a will, but is far simpler and much less expensive.

As our Bank deposits have grown, the number of withdrawals, although lagging deposits, has also grown. Essentially all of our withdrawals are for testing of deceased individuals. We recently asked some of the close family members of these deceased individuals to provide testimonials about the importance of DNA Banking. We were struck by the responses. These may be found here on our DNA Banking web site in the individual’s own words.

Although we hope to someday, PreventionGenetics has not yet made a profit on DNA Banking. We provide Banking because it’s an important service for the families we serve.

I hope all of you will carefully consider DNA Banking for yourselves and family members. DNA Banking makes a wonderful gift. Happy Holidays from PreventionGenetics.
Recent Webinars:

Finding Answers Comprehensive Miscarriage, Stillbirth & Neonatal Death Panel
Speakers: Diane Allingham-Hawkins Ph.D., FCCMG, FACMG Elizabeth McPherson M.D.

Genetic Testing in Ophthalmology - Focus on Eye Panels at PreventionGenetics
Speaker: Madhulatha Pantrangi, PhD

Click here to watch recorded webinars