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## Volume 9, Number 1

Welcome to the March 2017 PreventionGenetics newsletter. In this issue, we highlight our PGxome™ Custom Panels as well as our expanded suite of PGxome tests. We also introduce one of our new geneticists, Greg Fischer, PhD. In the President's Corner, Jim Weber, PhD discusses paralogous genes.

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# PreventionGenetics Leaps Ahead with PGxome™ Custom Panels

You already know PreventionGenetics for expert sequencing of individual genes and defined panels. You may also know that we provide one of the most powerful and cost effective whole exome sequencing tests, PGxome. PreventionGenetics is now pleased to announce that we are offering custom panels based on whole exome sequencing. Any subset of genes can be ordered from the list of ~4,000 clinically relevant genes. In addition, because of our unsurpassed expertise with Sanger sequencing, PGxome Custom Panels can be enhanced to achieve complete coverage when necessary.


From pricing to CPT codes to gene coverage, we pride ourselves on transparency. Our new PGxome Custom Panel Tool displays this information as you are building your custom panel. Choose exactly the genes you want sequenced, and choose the level of coverage you need. Get started with PGxome Custom Panels.

Read more.

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## Upcoming Webinar:

### Genetic Testing for Kidney Disorders

Speakers: Wuyan Chen, PhD


Register Here

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## One size does not fit all - flexible PGxome™ options to suit your patient's needs

We are excited to announce the expansion of PGxome, PreventionGenetics' whole exome sequencing test! We now offer PGxome diagnostic for individuals, duos, and trios. We also offer PGxome health screen, an exome-based test for individuals or couples seeking carrier status and/or susceptibility information for adult onset conditions.

What sets us apart? Using NextGen sequencing technology, PGxome assesses almost all genes from the human genome including coding regions and adjacent introns. While this is common for most exome platforms, as an added quality measure, variants identified in the ~4,000 genes known to be involved in Mendelian disorders are first manually reviewed by MD and PhD geneticists who have specialization in specific disease areas to ensure the best variant interpretation. This specialty-focused approach harnesses the collective knowledge and experience of many Geneticists, and through the power of many, we are able to help determine your patient's diagnosis. For more details regarding our PGxome testing options, click here.

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## DR. FISCHER SPECALIZES IN AUTISM DISORDERS AND HLA-TYPING

Greg Fischer, PhD, joined PreventionGenetics in October 2016 as a Human Molecular Geneticist. His portfolio includes Human Leukocyte Antigen (HLA) typing and Autism Spectrum Disorders.

Dr. Fischer earned his BSc in Biochemistry and Molecular Biology from the University of Wisconsin-Eau Claire in 2011 and his PhD in Genetics from the University of Wisconsin-Madison in 2016. Prior to joining PreventionGenetics, Dr. Fischer's doctoral work focused on immune recognition of the opportunistic fungal pathogen, *Aspergillus fumigatus,* and the genetic players of both humans and fungi that influence recognition and clearance of the organism.

Dr. Fischer was previously an undergraduate intern at PreventionGenetics in the summer of 2008, which was the leading factor behind his decision to pursue a career in Genetics. Originally from Central Wisconsin, Greg is proud to be back at PreventionGenetics where he has the opportunity to expand his clinical genetics knowledge beyond immunity. "PreventionGenetics has changed a lot since 2008, but the company's commitment to provide high-quality and accurate results to clients has not. The knowledge and dedication of all staff is impressive, and I am honored to be part of that team."

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## PRESIDENT'S CORNER

James L. Weber, PhD

### Paralogous Genes

One of the greatest technical challenges facing clinical DNA testing labs these days is sequencing of genes that are present in more than one copy in the haploid genome. Such genes are often called paralogs. Perhaps the toughest situation is a functional gene adjacent to a pseudogene with nearly

identical sequence and with sequence similarity extending throughout both exons and introns. Such genes are susceptible to gene conversion events, production of hybrids between the functional gene and pseudogene and copy number variations in the functional gene. Testing such genes is very complicated. My strong hunch is that no clinical lab today does a *perfect* job of assaying such genes.

The problem of paralogs is highly relevant to exome and genome sequencing. Labs need to know which genes can be sequenced accurately and which not. At PreventionGenetics we have tackled this problem head on.

We have carefully constructed a list of paralogous genes using multiple informatics approaches. These approaches involve both BLAT and Nextgen short read mapping algorithms. Our geneticists know when a called variant is in a paralogous region. In cases where we have validated an accurate Sanger sequencing test for one of these paralogous regions, we of course confirm the variant using Sanger. We have already validated tests for some of the paralogous genes involved most commonly in clinical genetics such as *PKD1*, *CYP21A2* and *NEB*. In cases where we have not yet validated a test, we do not report the variants. We are also gradually whittling down the list of paralogous genes that we cannot yet accurately sequence by tweaking our analysis pipelines and validating new tests.

In the more distant future, we hope that long read sequencing technology and *de novo* sequence assembly will come to the rescue. Until then, both clinicians and testing labs will need to use extreme caution.

## Visit us at these Upcoming Conferences:

**American College of Medical Genetics, March 22-24** - Phoenix Arizona, Booth #709
**Clinical Laboratory Management Association KnowledgeLab, March 26-29** - Nashville, TN
**American Academy of Neurology, April 22-29** - Boston, MA, Booth #961
**Canadian College of Medical Gentics, April 30-May 3** - Montreal, QC

## More Information:

- New Tests
-Tests by Category
- Requisition Forms
- Billing Policy
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- Order Testing Kits

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