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Welcome to the November 2009 PreventionGenetics Newsletter. In this issue we present new clinical tests for congenital disorders of glycosylation. We also introduce our new Director of Research Genotyping, Dave Schlesinger, PhD, and describe our Research Genotyping services. Finally, we inaugurate a President's Corner section written by Dr. James Weber, our company's President.

New Tests at PreventionGenetics

PreventionGenetics is pleased to announce the addition of new gene sequencing tests for congenital disorders of glycosylation (CDG). These new tests are part of Dr. Tom Winder's portfolio. For more information, please contact Dr. Winder at 715-387-0484 ext. 118 or at tom.winder@preventiongenetics.com

Congenital Disorders of Glycosylation:

PreventionGenetics now has a two tiered sequential test for CDG. [Tier 1](#) includes bi-directional sequencing of *PMM2*, *MPI*, and *ALG6*. In cases with demonstrated reduced phosphoanmutase or mannosophosphate isomerase activity plus diagnostic serum transferrin glycoforms, *PMM2* and *MPI* sequencing, respectively, is nearly 100% sensitive.

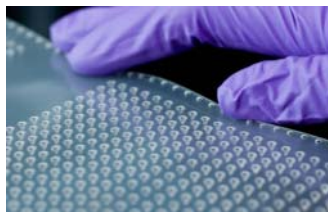
[Tier 2](#) includes genes *ALG3*, *DPM1*, *MPDU1*, *ALG12*, *ALG8*, and *ALG2*. Pathogenic mutations in the Tier 2 genes have been described in only a small number of patients, therefore clinical sensitivity cannot be estimated. Each of the genes in the sequential test are indicated for individuals with clinical and biochemical findings consistent with CDG. The genes for both tiers will be tested in the order specified by the client on the [Requisition Form](#).



Introducing Dave Schlesinger, PhD

We are pleased to announce the addition of Dave Schlesinger, PhD to our staff at PreventionGenetics. Dr. Schlesinger will lead our Research Genotyping services. Dave received his PhD from the University of Illinois at Urbana-Champaign. Prior to coming to PreventionGenetics Dr. Schlesinger was the senior investigator at Kim Laboratories where he oversaw the development of diagnostic tests for human gastrointestinal viruses. Dave brings new energy, knowledge, and experience to Research Genotyping.

Research Genotyping at PreventionGenetics focuses on Ancestry Informative Markers (AIMs), Single Nucleotide Polymorphisms (SNPs), Short Tandem Repeat Polymorphisms (STRPs), and Insertion/Deletion Polymorphisms (InDels). Our array tape technology allows us to perform high



throughput genotyping for small to medium projects (<200 polymorphisms). Array tape is essentially a series of microtiter plates on a continuous sheet of flexible plastic. This novel platform is ideally suited to genotyping many DNA samples with relatively few numbers of markers.

The staff at PreventionGenetics has 20 years of human genotyping experience and serves a global client list. Our extensive quality control and innovative genotyping system allow us to offer our services at highly competitive costs and with fast turnover without sacrificing data quality. We will continue to add new genotyping services to meet the demand from our customers. For more information on our Research Genotyping capabilities, please visit our [website](#). Dr. Schlesinger may be contacted directly at 715-387-0484 ext. 124 or by email at dave.s@preventiongenetics.com.

President's Corner

Jim Weber, PhD

In this regular feature of our Newsletter, I will cover various aspects of PreventionGenetics DNA Testing and Banking services and will also discuss general challenges in clinical genetics. My topic for this newsletter is the quality of Clinical DNA Testing at PreventionGenetics.

We are passionate about DNA test quality at our company. PreventionGenetics is fully CLIA accredited through CAP. To date, we have had zero errors out of approximately 400 external and internal Proficiency Tests.

Bidirectional Sanger sequencing of PCR products using capillary electrophoresis is the most widely used clinical DNA test methodology at PreventionGenetics. Despite well-known limitations tied mostly to PCR, this methodology will likely serve as the gold standard as much cheaper but more error-prone Next-Generation sequencing technology comes online. At PreventionGenetics our Sanger/capillary sequencing involves coverage of ~50 bp of flanking DNA beyond exon boundaries, automated calling of bases using two independent software packages, manual review of all electrophoresis traces by an experienced technician, and final review of the critical sequence data by two doctorate-level geneticists.

We also try very hard at PreventionGenetics to produce thorough and complete test reports. Although brevity is certainly a virtue in clinical test reports, we feel that completeness is even more important. For example, we report *ALL* differences between patient sequences and reference sequences in our reports. Although it may increase our liability risks somewhat, we also include our best possible full interpretation of each sequence variant using all available literature references, databases, and a variety of prediction programs.

For many months now, PreventionGenetics has had a standard turn around time for our gene sequencing tests of 40 calendar days. We are now in the process of cutting this in half. Some of our more complex tests and sequential sequencing of multiple genes will still often require more than 20 days, but the great majority of our test reports will be transmitted in three weeks.

PreventionGenetics is certainly not perfect or infallible, but we will go toe to toe on quality with any Clinical DNA Testing Lab anywhere. We also have a policy of continuous improvement at our company. Each and every working day, we become more experienced, improve our software or processes, or otherwise upgrade our services. If you're skeptical, talk with your colleagues who have already used our services, or better yet, give us a try. I am confident that you will be satisfied.

Coming in the December PreventionGenetics Newsletter

Our next Newsletter will focus on the pricing of clinical tests at PreventionGenetics and new additions to the PreventionGenetics test menu, including tests for Hypertrophic Cardiomyopathy and several Cardiac Rhythm disorders.

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.