

PUT US TO THE TEST

PREVENTION GENETICS

DISEASE PREVENTION THROUGH GENETIC TESTING



IN THIS ISSUE

[New building](#)

[New and updated tests](#)

[Introducing Madhu Pantrangi, Ph.D.](#)

[President's corner](#)

QUICK LINKS

[Our website](#)

[Requisition form](#)

[Join Our Mailing List!](#)

Volume 5, Number 4

Welcome to the October 2013 PreventionGenetics newsletter. In this issue, we present 61 new and updated Next-Gen and Sanger tests. PreventionGenetics proudly introduces one of our newest geneticists, Dr. Madhu Pantrangi. We cover the dedication of our new facility, and in the President's corner, Dr. Jim Weber discusses test quality.

New building dedicated October 3

The latest step in the growth of PreventionGenetics was taken on October 3 with the dedication of our brand new 50,000 square-foot lab and office building at the Mill Creek Business Park in Marshfield, Wisconsin. Dedication of the new building was the culmination of 18 months of planning and construction.

The new building includes a 130 seat auditorium, Helix Hall, which will be used for seminars and other educational purposes. On October 4, for example, we hosted 300

eighth-grade students from local schools to learn a little about human genetics and to extract DNA from bananas. The new building was designed with many green features, especially including geothermal heating and cooling. There are no conventional furnaces or air conditioners in the building. Rather, all temperature control is achieved by extracting or contributing heat to fluid which is pumped through pipes installed beneath a field adjacent to the building. Our new building will roughly triple available operating space. This will provide ample room for future growth.



Company founder and President Dr. Jim Weber talks to the audience in Helix Hall.



DNA Banking Director Donna Dorshorst (at far left) leads one of the tours after the ribbon-cutting program.



Long-time employee and Specimen Processing Lab Manager, Mary Urban, cuts the ribbon.

New and updated tests at PreventionGenetics

Please follow the links for full descriptions of each new test.

New and updated Next-Gen sequencing tests

Marfan syndrome and related Aortopathies NGS-MSA ([Test #1212](#))

Bardet-Biedl syndrome NGS-BBS ([Test #1053](#))

Joubert and Meckel-Gruber syndromes NGS-JSMKS ([Test #1057](#))

Nephronophthisis and Senior-Loken syndrome NGS-NPH ([Test #1058](#))

New and updated sequencing tests by disease category

Cancer

Mosaic Variegated Aneuploidy syndrome

BUB1B ([Test #1290](#))

Cardiovascular

Amyloidosis *TTR* ([Test #929](#))

Hypertrophic Cardiomyopathy with Mitochondrial Myopathy and Progressive External Ophthalmoplegia *SLC25A4* ([Test #1250](#))

Marfan syndrome and related Aortopathies NGS-MSA ([Test #1212](#))

Shprintzen-Goldberg syndrome *SKI* ([Test #1158](#))

Ciliopathy

Bardet-Biedl syndrome NGS-BBS ([Test #1053](#))

Joubert syndrome *TMEM237* ([Test #1021](#))

Joubert and Meckel-Gruber syndromes NGS-JSMKS ([Test #1057](#))

Meckel-Gruber syndrome *B9D1* ([Test #577](#))

Nephronophthisis and Senior-Loken syndrome NGS-NPH ([Test #1058](#))

Cognitive

Cortical Dysplasia-Focal Epilepsy syndrome *CNTNAP2* ([Test #1522](#))

Pitt-Hopkins-Like syndrome 2 *NRXN1* ([Test #1523](#))

Rett syndrome *MECP2* ([Test #1455](#))

Smith-Magenis and Potocki-Lupski syndromes *RAI1* ([Test #1519](#))

Developmental

Alagille syndrome *JAG1* ([Test #427](#))
Bardet-Biedl syndrome NGS-*BBS* ([Test #1053](#))
Cornelia de Lange syndrome *HDAC8* ([Test #895](#))
Joubert syndrome *TMEM237* ([Test #1021](#))
Joubert and Meckel-Gruber syndromes NGS-*JSMKS* ([Test #1057](#))
Marfan syndrome and related Aortopathies NGS-*MSA* ([Test #1212](#))
Meckel-Gruber syndrome *B9D1* ([Test #577](#))

Kidney

Hyperglycemia and Hypoglycemia Disorders *CASR* ([Test #1457](#))
Hyperglycemia and Hypoglycemia Disorders *GCK* ([Test #1220](#))
Liddle Syndrome and Pseudohypoaldosteronism Type 1 *SCNN1G* ([Test #1274](#))
Nephronophthisis and Senior-Loken syndrome NGS-*NPH* ([Test #1058](#))
Nephrotic syndrome *WT1* ([Test #1230](#))
Polycystic Kidney Disease *PKD2* ([Test #187](#))

Lysosomal

Alpha-Mannosidosis *MAN2B1* ([Test #1236](#))

Metabolic/Mitochondrial

Dihydropyrimidine Dehydrogenase Deficiency *DPYD* ([Test #1238](#))
Mitochondrial Phosphate Carrier Deficiency *SLC25A3* ([Test #1252](#))
Orotic Aciduria *UMPS* ([Test #1430](#))
Progressive External Ophthalmoplegia and Hypertrophic Cardiomyopathy with Mitochondrial Myopathy *SLC25A4* ([Test #1250](#))

Neurologic

Amyloidosis *TTR* ([Test #929](#))
Epilepsy *EFHC1* ([Test #1424](#))
Epilepsy *GABRA1* ([Test #1454](#))
Generalized Epilepsy with Febrile Seizures Plus and Dravet syndrome *SCN1B* ([Test #634](#))
Mosaic Variegated Aneuploidy syndrome *BUB1B* ([Test #1290](#))
Pontocerebellar Hypoplasia Type 1A *VRK1* ([Test #308](#))
Pontocerebellar Hypoplasia Type 6 *RARS2* ([Test #309](#))
Rett syndrome *MECP2* ([Test #1455](#))

Neuromuscular

Distal Arthrogryposis Sanger Panel ([Test #335](#))

Pancreas

Gitelman syndrome *SLC12A3* ([Test #1262](#))
Chronic Pancreatitis *CASR* ([Test #1444](#))
Chronic Pancreatitis *CFTR* ([Test #150](#))
Chronic Pancreatitis *CTRC* ([Test #1402](#))
Chronic Pancreatitis Sanger Panel ([Test #1445](#))
Chronic Pancreatitis *PRSS1* ([Test #1285](#))

Skeletal

Marfan syndrome and related Aortopathies NGS-*MSA* ([Test #1212](#))
Shprintzen-Goldberg syndrome *SKI* ([Test #1158](#))

Skin

Epidermolysis Bullosa Simplex *KRT14* ([Test #979](#))

Vision

Choroideremia *CHM* ([Test #1237](#))

Cone Dystrophy 3 and Cone-Rod Dystrophy 14 *GUCA1A* ([Test #678](#))

Leber Congenital Amaurosis 10 *CEP290* ([Test #823](#))

Retinitis Pigmentosa 17 *CA4* ([Test #675](#))

Retinitis Pigmentosa 26 *CERKL* ([Test #690](#))

Retinitis Pigmentosa 27 *NRL* ([Test #689](#))

Retinitis Pigmentosa 31 *TOPORS* ([Test # 674](#))

Retinitis Pigmentosa 35 and Cone-rod dystrophy 10 *SEMA4A* ([Test #670](#))

Retinitis Pigmentosa 38 *MERTK* ([Test #676](#))

RLBP1-related disorders *RLBP1* ([Test #673](#))

X-Linked Juvenile Retinoschisis *RS1* ([Test #868](#))

Human molecular geneticist focuses on vision disorders

Madhu Pantrangi, Ph.D., joined PreventionGenetics earlier this year. Her portfolio of clinical tests will focus on vision disorders.

Madhu's postdoctoral fellowship at Marshfield Clinic Research Foundation included work on the human pathogen *Staphylococcus aureus* virulence gene expression and regulation in the presence and absence of different antimicrobial agents. She was also involved in *S. aureus* new strain gene annotation, whole-genome mapping to predict *S. aureus* virulence motifs, identification of *Corynebacterium nigricans* and pregnancy outcomes. In addition, she served as co-investigator in "Metagenomics study of chronic fatigue syndrome (CFS)."



Madhu Pantrangi, Ph.D.

"There are more than 200 retinal disorders and 200 associated genes that have been identified so far (RetNet). Early diagnosis by gene tests might help slow the disease progression or sometimes prevent vision loss," she noted. Dr. Pantrangi aims to provide gene tests for all retinal disorder-associated genes and looks forward to introducing Next-Gen panels in this area.

"To better understand disease, you need to understand genetics," said Dr. Pantrangi, who comes from a long line of medical professionals. "The medical field is very exciting. Most of my family's side are physicians. I always wanted to be in the medical field."

During the spring 2013 academic term, she served on the faculty at the University of Wisconsin-Stevens Point as an assistant professor of microbiology.

Dr. Pantrangi received her Ph.D. in genetics from the University of Delhi and

her master's in biotechnology from Pondicherry University.

President's corner

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Jim Weber, Ph.D.

We recently added a new section to our website labeled, "Quality." Test quality has always been fundamental to all that we do at PreventionGenetics. Our employees sometimes get tired of hearing me say, "Get the tests right."

PreventionGenetics is CLIA accredited through CAP. Our quality assurance processes, however, far exceed regulatory requirements. For example, we have had to date zero errors out of 2,500 total Proficiency Tests, both external and internal. Our sequencing error rate is about one per million nucleotides or one per 1000 sequence variants. Sequencing error rates, although tough to measure, should I think be discussed much more in the clinical genetics community.

In addition, we offer many special quality features as a routine part of our standard clinical testing. For example, all of our test results and reports are carefully reviewed by four highly trained and experienced individuals, including two doctorate-level geneticists. All targeted mutation testing (mostly single and double exon sequencing), especially including all prenatal testing, is performed twice by different lab personnel. Our genotyping identity panel is run on all clinical and DNA bank specimens to detect specimen mislabeling. We have found that roughly one of 500 specimens received by our lab is mislabeled. A full list of these special features is available from our website. Many labs offer some of these features, but my hunch is that no other lab offers all of these features.

Competition is increasing in clinical DNA testing. This makes it tougher for us and for other labs, but competition also leads to improved quality. We challenge other labs to match or even exceed us in quality. And you have my promise that PreventionGenetics will continue to relentlessly pursue improvement in all aspects of our testing services.

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.