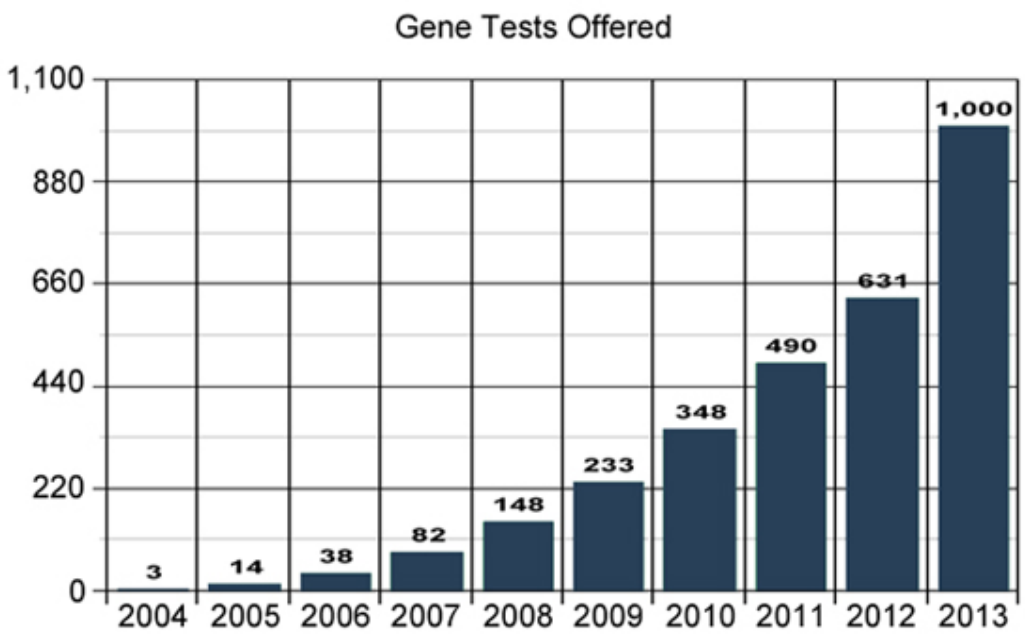
NEWSLETTER

Volume 5, Number 5

Welcome to the December 2013 PreventionGenetics newsletter. In this issue, we highlight the comprehensive nature of our testing. We also present 61 new NextGen and Sanger sequencing tests and introduce one of our newest geneticists, Dr. Honey Reddi. In the President's corner, Dr. Jim Weber reflects on the year gone by and the new year to come.

PreventionGenetics marches on toward comprehensive coverage of clinically relevant genes.

PreventionGenetics has been aggressively adding new tests to our test menu since our company started 10 years ago. As we have grown, the rate of addition of new tests has increased. PreventionGenetics currently offers about 900 germline clinical DNA tests covering 1,000 genes. This is the largest test menu of any lab in America. We also have many new tests covering hundreds of additional genes in various stages of development.
Our goal is not a secret. PreventionGenetics intends to offer comprehensive coverage of the clinically relevant portion of the genome over the next few years. This coverage includes NextGen and Sanger sequencing, along with aCGH testing for larger deletions and duplications. We offer single gene (and single exon) tests along with many gene sequencing panels.



Why is comprehensive coverage important? Because it saves time and money. Instead of spending hours hunting for labs that offer particular tests and dealing with a myriad of different requisition forms and test report formats, providers now have the option to use one primary lab with a consistent, logical set of order forms, report formats and billing processes. No longer is it necessary to send the proband's specimen to one lab and specimens from family members to a second. No longer is it necessary to navigate complex and out of date lab databases. One stop at the PreventionGenetics web site provides all the information that you need.

As emphasized in previous Newsletters, as well as in the quality section of our website, we have not sacrificed quality as we have grown. If anything, our increased size has provided resources to improve our already outstanding quality. The same basic lab processes that we have used in the past are still in place and are used for all genes. You can rely on PreventionGenetics for continued highest quality, low prices, excellent service and now comprehensive coverage of clinically-relevant genes.

We encourage testing contracts with client institutions. We are happy to tailor our services to your particular needs. Quantity price discounts are also available. Please contact us to discuss contracts.

NEW TESTS

Autoimmune
Autoimmune Polyendocrinopathy syndrome Type 1 AIRE (#1224)

Blood and Lymph
Hemophilia C *F11* (#239)

Cancer
Autoimmune Polyendocrinopathy syndrome Type 1 *AIRE* (#1224)
Hereditary Breast and Ovarian Cancer *RAD50* (#1294)
Lung Cancer *EGFR* (#1207)
Lynch syndrome *EPCAM* (#1282)

Cardiovascular
Arrhythmogenic Right Ventricular Cardiomyopathy ARVC NGS Panel (#1315)

Hypertrophic Cardiomyopathy HCM NGS Panel (#1313)

Developmental
Familial Dysautonomia *IKBKAP*-targeted (#847)
Familial Dysautonomia *IKBKAP*-full (#846)
Multiple Epiphyseal Dysplasia NGS-MED (#1317)
Noonan syndrome 8 RIT1 (#1114)
Pontocerellar Hypoplasia Type 1A *EXOSC3* (#1208)
Stickler syndrome NGS-SS (#1319)

Endocrine
Autoimmune Polyendocrinopathy syndrome Type 1 AIRE (#1224)

Hearing
Branchiootorenal syndrome SIX1 (#1474)
Branchiootorenal syndrome 1 *EYA1* (#1473)
Branchiootorenal syndrome 2 *SIX5* (#1476)
Nonsyndromic hearing loss and deafness GJB2 (#986)
Waardenburg syndrome type II MITF (#1178)
Waardenburg syndrome types I and III PAX3 (#1468)
WFS1-related disorders *WFS1* (#1477)
Wolfram syndrome Type 2 *CISD2* (#1478)
Wolfram syndrome WFS Sanger Panel (#1480)

Kidney
Apparent mineralocorticoid Excess HSD11B2 (#1269)
Branchiootorenal syndrome SIX1 (#1474)
Branchiootorenal syndrome 1 EYA1 (#1473)
Branchiootorenal syndrome 2 SIX5 (#1476)
Nail-Patella syndrome LMX1B (#1458)

Lysosomal
Neuronal Ceroid Lipofuscinosis 2 TPP1 (#867)
Neuronal Ceroid Lipofuscinosis 5 CLN5 (#926)
Neuronal Ceroid Lipofuscinosis 8 and CLN8 Northern Epilepsy Variant CLN8 (#848)

Mitochondrial
Autosomal Dominant Progressive External Ophthalmoplegia DNA2 (#1253)
Encephalomyopathic form of Mitochondrial DNA Depletion syndrome SUCLA2 (#1251)

Neurologic
Amyotrophic Lateral Sclerosis *18PFN1* (#1037)
Andermann syndrome *SLC12A6* (#1557)
Epilepsy: Dravet syndrome and Generalized Epilepsy with Febrile Seizures Plus *SCN1A* (#1456)
Familial Dysautonomia *IKBKAP*-targeted (#847)
Familial Dysautonomia *IKBKAP*-full (#846)
Neuronal Ceroid Lipofuscinosis 2 *TPP1* (#867)
Neuronal Ceroid Lipofuscinosis 5 *CLN5* (#926)
Neuronal Ceroid Lipofuscinosis 8 and CLN8 Northern Epilepsy Variant *CLN8* (#848)

Neuromuscular
Andermann syndrome *SLC12A6* (#1557)
Congenital Myasthenic syndrome NGS-CMS Panel (#1323)
Limb Girdle Muscular Dystrophy Type 2S *TRAPPC11* (#1515)
Muscular dystrophy- dystroglycanopathy *GMPPB* (#1492)
Nemaline Myopathy *KLHL40* (#1491)
Welander Distal Myopathy *TIA1* (#898)

Sex development
Disorders of sex development DHH (#905)
Disorders of sex development SRY (#901)

Skeletal
Multiple Epiphyseal Dysplasia NGS-MED (#1317)
Stickler syndrome NGS-SS (#1319)

Vision
Aniridia PAX6 (#1483)
Bietti Crystalline Corneoretinal Dystrophy CYP4V2 (#1541)
Ectopia Lentis 2 ADAMTSL4 (#1542)
Primary Congenital Glaucoma CYP1B1 (#1544)
Primary Congenital Glaucoma LTBP2 (#1545)
Retinitis Pigmentosa 48 GUCA1B (#1571)
WFS1-related disorders WFS1 (#1477)
Wolfram syndrome Type 2 CISD2 (#1478)
Wolfram syndrome WFS Sanger Panel (#1480)

Clinical molecular geneticist specializes in hearing loss, pancreas disorders

Honey Reddi, Ph.D., DABMG, joined PreventionGenetics in July 2013 as a clinical molecular geneticist. Her portfolio of specialties includes hearing loss, pancreatitis and amyloidosis.
Dr. Reddi was most recently with Mayo Clinic in Rochester, Minnesota, where she conducted research on the clinical and translational aspects of thyroid cancer, including certain biomarkers for early detection of thyroid cancer. She served as assistant professor for translational research at Mayo Clinic for the past seven years.
Much of her postdoctoral work at Evanston Northwestern Healthcare, Northwestern University and Mayo Clinic concentrated on mouse models of human disease, with a focus on cancer research.

“My interest is in being able to dissect out disease mechanisms,” she said. A desire to pair her research background with the clinical side of DNA testing led her to a clinical fellowship in molecular genetics at Mayo. In fall 2013, Dr. Reddi received certification as a Diplomate of the American Board of Medical Genetics.
“I’ve always focused on trying to do the best for patients, in terms of translating research to the clinic and beyond, leading up to my current position at PreventionGenetics. This ties in to our mission at P.G. - preventing human disease and disability through genetic testing.” At PreventionGenetics, Dr. Reddi is excited to be working on hearing loss tests, “because it is much more common than we think. One in 500 newborns has bilateral sensorineural hearing loss, and just a couple of companies offer molecular testing. Our aim is to be able to soon offer the most comprehensive hearing loss panel testing.”
In addition to her work with genetic testing, Dr. Reddi is the director of Sales and Marketing.
A native of Hyderabad, India, Dr. Reddi received her Ph.D. in biotechnology from the International Center for Genetic Engineering & Biotechnology (ICGEB) and Hamdard University in India.

President's Corner

It has been an eventful year at PreventionGenetics. We added about 400 new gene sequencing tests

(both Sanger and NextGen) to our menu. Seven new doctorate geneticists were recruited to our staff along with our second genetic counselor. And we moved into our brand new 50,000 sq. ft. lab and office building in the Marshfield Mill Creek Business Park.
The New Year will bring additional progress. Many new tests will continue to be added, including our first Chromosomal Microarray (CMA) test and our first genome-wide sequencing test. A number of new client support features will be introduced making it easier for customers to order tests and to receive test reports. Our insurance preauthorization services will also be expanded.
We thank all of our clients for their generous support in 2013 and look forward to serving you in 2014.

Happy holidays from PreventionGenetics

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