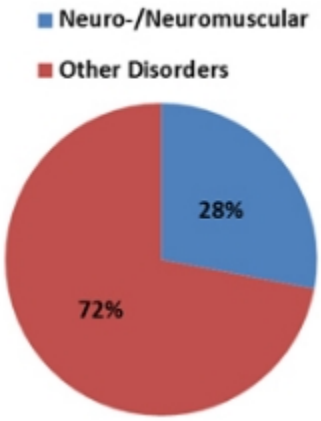
Volume 6, Number 1

Welcome to the first newsletter of 2014 from PreventionGenetics. In this issue we highlight our test portfolio for neuromuscular disorders. We present our updated test menu with 70 new single gene Sanger tests, 20 new and 6 updated NextGen Panels and 7 new Sanger panels. We also introduce Dr. Beth Buckley, a recent addition to our expanding team of geneticists. In the President's corner, Dr. Jim Weber reflects on the need for increased genetic education.

Testing for Neuromuscular/Neurological disorders at PreventionGenetics, a significant component of our comprehensive clinical DNA testing menu - 10 years and 1000 genes later

PreventionGenetics reached a new height in its 10th year with the addition of its 1000th gene to its test menu in early February. Since our inception in 2004, we have strived to constantly add to our already extensive menu of clinical DNA testing. We envision being able to offer testing for all genetic disorders. Our current portfolio of genetic testing is expansive and includes blood and lymph, dental, dermal, epilepsy, gastrointestinal, hearing loss, hereditary cancers, cardiovascular, developmental, intellectual disability, lysosomal storage, mitochondrial, neurological, neuromuscular, skeletal, metabolic and vision disorders.



Clinical testing for disorders that involve the skeletal muscle and peripheral nervous system (neuromuscular) and/or the brain (neurological) account for a significant proportion (28%) of the test menu at PreventionGenetics. We currently test for 276 genes associated with these disorders using different technologies including NextGen, Sanger sequencing and deletion/duplication analysis by gene-centric arrayCGH.

We are able to offer such an extensive menu largely due to the expertise of two of our most experienced geneticists, Dr. Tom Winder, PhD, FACMG and Dr. Khemissa Bejaoui, PhD along with our most recent addition, Dr. Beth Buckley, PhD. Dr. Winder's expertise is neuromuscular disorders (muscular dystrophies and myopathies), Dr. Bejaoui's expertise is developmental, lysosomal storage disorders, and Rasopathies and Dr. Buckley's area of focus is neurodegenerative disorders including epilepsy and intellectual disability. Together they strive to offer comprehensive testing for the diagnoses of this category of disorders.

At the end of the first quarter in 2014, we offer 14 NextGen panels covering 141 genes and 276 single gene Sanger sequencing tests for this portfolio of disorders. For phenotypes that involve multiple genes that not yet currently offered as NextGen panels, we are able to offer customized sequential testing for any genes of interest.

PreventionGenetics will continue to add more genes of clinical relevance to our test menu utilizing cutting edge technology. We offer the highest quality in testing at the lowest price possible.

NEW TESTS

NextGen Sequencing Panels

Autosomal Dominant Limb-Girdle Muscular Dystrophy (#1349)
Autosomal Recessive Limb-Girdle Muscular Dystrophy (#1347)
Cancer (#1355)
Cone-Rod Dystrophy (#1337)
Congenital Fiber Type Disproportion NextGen Sequencing Panel (#1371)
Congenital Myopathy NextGen Sequencing Panel (#1365)
Dilated Cardiomyopathy (#1339)
Distal Hereditary Motor Neuropathy (#1359)
Distal Hereditary Myopathy (#1351)
Early Epileptic Encephalopathy, Dominant and X-Linked (#1321)
Fetal Akinesia Deformation sequence/Lethal Multiple Pterygium syndrome (#1361)
Hereditary Paraganglioma-Pheochromocytoma (#1329)
Left Ventricular Noncompaction (LVNC) (#1333)
Limb-Girdle Muscular Dystrophy (#1345)
Myofibrillar Myopathy (#1357)
Pancreatic Cancer (#1343)
Steroid-Resistant Nephrotic Syndrome (SRNS)/Focal Segmental Glomerulosclerosis (FSGS) (#1335)
Renal Cancer (#1331)
Type VI-Related Collagenopathy (#1353)
Xeroderma Pigmentosum (#1341)

Sanger Sequencing Panels

Familial Hypercholesterolemia (#875)
Hirschsprung disease (#1565)
Long QT syndrome (#1100)
Pseudohypoaldosteronism Type II (#1280)
Primary Congenital Glaucoma Sanger panel (#565)
Waardenburg syndrome (#1556)
X-Linked Retinitis Pigmentosa Sanger panel (#825)

Single Gene Sanger Testing

ADA Deficiency *ADA* (#1231)
Adrenal Hypoplasia AND 46, XY sex reversal *NR0B1* (#1462)
Alzheimer's Disease *PSEN1* (#1414)
Amelogenesis Imperfecta *AMELX* (#1596)
Amelogenesis Imperfecta *FAM20A* (#1602)

Amelogenesis Imperfecta *KLK4* (#1598)
Amelogenesis Imperfecta *MMP20* (#1597)
Amelogenesis Imperfecta *FAM83H* (#1600)
Amelogenesis Imperfecta *WDR72* (#1599)
Amyloidosis *APOA1* (#1403)
Amyloidosis *GSN* (#1409)
Argininosuccinate Lyase Deficiency *ASL* (#1181)
Aspartylglucosaminuria *AGA* (#1558)
Bartter Syndrome *BSND* (#1257)
Beta-Thalassemia and Hemoglobinopathy *HBB* (#1088)
Bjornstad syndrome/Gracile syndrome *BCS1L* (#1562)
Canavan disease *ASPA* (#1559)
Carney Complex *PRKAR1A* (#1287)
Char syndrome *TFAP2B* (#942)
Charcot-Marie-Tooth Disease 2A and Mitochondrial DNA Depletion syndrome *MFN2* (#1254)
Cherubism *SH3BP2* (#1288)
Cone-Rod Dystrophy AND Leber Congenital Amaurosis *RPGRIP1* (#1546)
Congenital Adrenal Hyperplasia *CYP21A1* (#1419)
Congenital Anomalies of Kidney and Urinary Tract *DSTYK* (#495)
Congenital Central Hypoventilation Syndrome *PHOX2B* (#1173)
Congenital Nystagmus *FMRD* (#1639)
Congenital Variant Rett syndrome/FOXG1 syndrome *FOXG1* (#1637)
Cryopyrin-Associated Periodic syndromes *NLRP3* (#1638)
Cystathioninuria *CTH* (#879)
Dubin-Johnson syndrome *ABCC2* (#1591)
Ehlers-Danlos syndrome *TNXB*-exon 35 (#877)
Familial Alzheimer's Disease *APP* (exons 16 and 17) (#604)
Familial Hypercholesterolemia *PCSK9* (#870)
Familial Mediterranean Fever *MEFV* (#1555)
FGFR1-Related disorders *FGFR1* (#498)
FGFR2-Related disorders *FGFR2* (#499)
Focal Dermal Hypoplasia *PORCN* (#958)
Fragile X syndrome *FMR1* (#558)
Geroderma Osteodysplasticum *GORAB* (#1260)
Glucose-6-Phosphate Dehydrogenase Deficiency *G6PD* (#1259)
Goldberg-Shprintzen Megacolon syndrome *KIAA1279* (#1568)
Gracile syndrome *BCS1L* (#1562)
Hemophilia B *F9* (#1577)
Hereditary Breast and Ovarian Cancer *RAD51D* (#1293)
Hereditary Paraganglioma-Pheochromocytoma Syndrome *MAX* (#1138)
Hereditary Spherocytosis *ANK1* (#1258)
Hirschsprung Disease 2 *EDNRB* (#1572)
Hirschsprung Disease 3 *GDNF* (#1566)
Hirschsprung Disease 4 *EDN3* (#1564)
Hirschsprung Disease, Cardiac Defects and Autonomic Dysfunction *ECE1* (#1569)
Kohlschutter-Tonz syndrome *ROGD1* (#1605)
Lafora disease *NHLRC1* (#635)
Long QT syndrome and Jervell and Lange-Nielsen syndrome *KCNQ1* (#1040)
Long QT syndrome *KCNH2* (#1041)
Medulloblastoma and Nevoid Basal Cell Carcinoma syndrome/Gorlin syndrome *SUFU* (#1281)
Mohr-Tranebjaerg/Jensen *TIMM8A* (#1467)
Mowat-Wilson syndrome *ZEB2* (#1567)
Muckle-Wells syndrome *NLRP3* (#1638)
Neuronal Ceroid Lipofuscinosis 3 *CLN3* (#1560)
Oculocutaneous Albinism Type 1 *TYR* (#1482)
Oculocutaneous Albinism Type II *OCA2* (#1481)

Popliteal Pterygium syndrome 2 *RIPK4* (#897)
Primary Hyperoxaluria Type 1 *AGXT* (#857)
Primary Hyperoxaluria Type 2 *GRHPR* (#858)
Primary Open Angle Glaucoma *OPTN* (#568)
Protein C Deficiency *PROC* (#1575)
Pseudohypoaldosteronism Type II *CUL3* (#1279)
Pseudohypoaldosteronism Type II *KLHL3* (#1278)
Pseudohypoaldosteronism Type II *WNK1* (#1276)
Pseudohypoaldosteronism Type II *WNK4* (#1277)
Renal Coloboma syndrome and Isolated Renal Hypoplasia *PAX2* (#493)
Renal Cysts and Diabetes syndrome *HNF1B* (#494)
Renal Hypomagnesemia *CLDN16* (#1464)
Renal Hypomagnesemia *CLDN19* (#1465)
Spondylocostal Dysostosis *HES7* (#1422)
Tubular Aggregate Myopathy *STIM1* (#328)

Dr. Buckley Specializes in Neurological and Neurocognitive Disorders

Her doctoral thesis at the University of Wisconsin-Madison focused on epigenetic regulation of gene expression. Using the roundworm as a model system, Dr. Buckley studied how a process called RNA interference (RNAi) could silence gene expression in the nucleus. Specifically, she studied RNA-based gene silencing that could be inherited for multiple generations, without altering the sequence of the DNA. Dr. Buckley helped identify an Argonaute protein that directed repressive histone modifications which were heritable across generations.

Dr. Buckley, who is also the director of PreventionGenetics's specimen processing lab, is excited to work in a clinical setting and to "be able to take all the research about the genetics of disease and use it to help patients decode their DNA. The hope is that clinical genetics will not only help identify the cause of disease, but that this knowledge will also inform future health care decisions and empower patients."

President's Corner

Along with many other human geneticists, I eagerly look forward to the day when essentially all patients have their genomes sequenced as a routine part of health care. However, I don't think that we are ready yet for universal genomic sequencing. Some of the major obstacles are listed below.

- Sequencing technology is insufficient.
- Patient sequences are not being stored in electronic health records.
- Patient sequences are not being shared across providers and family members.
- Sequence interpretation is in its infancy.

- Health care providers and patients are not ready.

In the next few President Corners I'll discuss these obstacles, starting in this issue with the last one on the list.

Recent polls suggest that while many people would accept full genomic sequencing on newborns, significant numbers would not (Goldenberg et al. Genetics Med 16:78, 2014; Bombard et al. Eur J Hum Genet 2014.22). Why are so many resistant to genomic sequencing? I don't pretend to fully understand, but maybe we have at least identified some of the factors. Despite GINA, people still fear genetic discrimination. People worry that they won't be able to sleep if they know their genomes. Health care providers foresee being buried under an avalanche of complicated data. There is concern about stigmatization -- about being labeled "genetically flawed". But perhaps the greatest reason is that most people know little about genetics, and are naturally leery about a potent technology that they don't understand.

I think therefore that the key to gaining people's acceptance is genetics education. Providers and patients need to learn more about genetics at all levels. I'm not personally an educator, and PreventionGenetics' primary task is not education. Nevertheless, we try in our own small way to help. Here are a few examples. At least four technicians who have worked in my labs over the years are now certified genetic counselors. Three more of our techs are currently applying to counseling school. Clinical molecular genetics fellows from the new University of Wisconsin – Madison program have started three month rotations at PreventionGenetics. Last fall we hosted over 300 local middle school students to learn a little about human genetics and to extract DNA from bananas. This spring, several of our staff, including Christina Zaleski our lead genetic counselor, will teach a four week mini-course on genetics open to the public and sponsored by the local branch of the University of Wisconsin.

No group alone is going to fill the education gap. However, if we all work together and if we are persistent, then I think the general public will gradually come to recognize and accept the enormous benefits of clinical genomic sequencing.

Quick Links

Our website
Req forms
Kit requests
Join our mailing list

Visit us at:

ARVO	Booth	#446
DDW	Booth	#2949
ASCO	Booth	#2042
NSGC	Booth	#208
ASHG	Booth #1034	

PREVENTION GENETICS

DISEASE PREVENTION THROUGH GENETIC TESTING

3800 South Business Park Ave. Marshfield, WI 54449 Ph: 715-387-0484 Fax: 715-207-6602