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Volume 5, Number 3

Welcome to the September 2013 PreventionGenetics newsletter. In this issue, we introduce Disease Prevention articles. Dr. Khemissa Bejoui discusses new tests for Amyotrophic Lateral Sclerosis (ALS). We welcome Dr. Wuyan Chen, human molecular geneticist.
We present 73 new and updated Next-Gen and Sanger tests. In the President's corner, Dr. Jim Weber discusses disease prevention through genetic testing.

Prevention articles to help with insurance preauthorizations

Katie Willfahrt
PreventionGenetics Intern

PreventionGenetics has launched Disease Prevention articles to help clients in writing letters of medical necessity.

Our prevention pages show how genetic testing can decrease morbidity and mortality of genetic disease and improve the quality of life for individuals and families.

In some cases such as Malignant Hyperthermia and Phenylketonuria, disease and disability may be prevented entirely. In other cases, it may only be currently possible to delay the onset or reduce the severity of disease.

Through the efforts of our college intern, Katie Willfahrt, about 15 articles are now available on our website.

Willfahrt completed the first 12-week genetic counseling internship at PreventionGenetics. She is a junior at the University of Minnesota and plans to pursue a career in genetic counseling.

"This project has been a great opportunity to gain experience and knowledge in all aspects of genetic counseling," she said. In addition to the Prevention

articles, she also completed a number of other projects to help prepare her for graduate school.

Go to this link for a full listing of the Disease Prevention articles currently available on our website.

New test offered for C9orf72 ALS gene

Khemissa Bejoui, Ph.D.

Amyotrophic Lateral Sclerosis (ALS) is a debilitating disease that affects nerve cells in the brain and the spinal cord. PreventionGenetics is pleased to offer a new test for the hexanucleotide repeat within the C9orf72 ALS gene.

Testing strategy

The C9orf72 Gene Hexanucleotide Repeat Expansion test consists of a combination of two complementary analyses. The repeat-primed PCR assay is used as a screening method for the presence or absence of the GGGGCC hexanucleotide repeat expansion in the C9orf72 gene. The fluorescent fragment-length assay confirms the results obtained from the repeat-primed PCR assay.

PreventionGenetics also offers an ALS Gene Sequencing Panel.

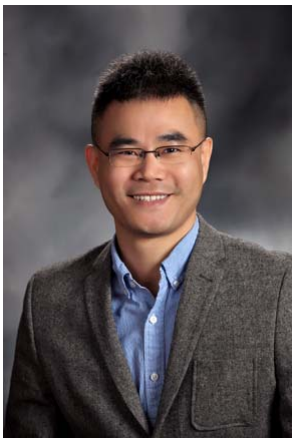
Indications for test

All patients with symptoms suggesting ALS, including autosomal dominant ALS and sporadic ALS with or without frontotemporal dementia (FTD), are candidates for this test.

"Because a pathogenic expansion of the GGGGCC hexanucleotide repeat in a non-coding region of C9orf72 has been reported as the most common cause of ALS with or without FTD, we will first screen the patients' DNA for the presence or absence of this expansion," noted Khemissa Bejaoui, Ph.D., PreventionGenetics clinical molecular geneticist. "When the pathogenic expansion is excluded, we will perform bidirectional DNA sequencing of all coding exons of five ALS genes (SOD1, FUS, TARDBP, ANG and OPTN)."

Human molecular geneticist joins PreventionGenetics

Wuyan Chen, Ph.D., joined PreventionGenetics earlier this year as a human molecular geneticist.



Wuyan Chen, Ph.D.

His postdoctoral fellowship at the National Institute on Aging provided well-rounded training in both clinical and laboratory genetics. His postdoctoral research focused on medical genetics of congenital adrenal hyperplasia due to 21-hydroxylase deficiency.

"Most of my postdoctoral training is similar to what we do here at PreventionGenetics, so my background matches very well. It was a very natural change from academia to a clinical diagnostics lab."

Dr. Chen's portfolio of specialties includes mitochondrial, kidney, endocrine and reproductive disorders.

What excites him about coming to work each day is knowing he can make a difference in patients' lives. "What we are doing is helping, in an effective way, to provide genetic information to patients. That's what's important for me."

Dr. Chen enjoys working at PreventionGenetics and living in Marshfield. "We have a pleasant working environment here," he said. "We have a strong team of technicians in our R&D and clinical lab, along with talented and knowledgeable Ph.D. geneticists. The company's rapid growth provides me with wonderful opportunities to learn new things and develop my career."

A native of Guangdong Province in southern China, Dr. Chen received his Ph.D. in biochemistry and molecular biology at Shanghai Jiao Tong University, Shanghai, China.

New and updated Next-Gen, Sanger and Panel Tests at PreventionGenetics

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

Next-Gen
Noonan spectrum disorders/Rasopathies NGS-RAS (Test #1309)
Blood
Acute Myeloid Leukemia CEBPA (Test #996)
Cancer
Hereditary Leiomyomatosis and Renal Cell Cancer or

Fumurase Deficiency FH (Test #1286)
CDC73-Related disorders *CDC73* (Test #1206)

Cardiovascular

Cathecholaminergic Polymorphic Ventricular Tachycardia *CPVT (Test #1311)*
Supravalvular Aortic Stenosis *ELN* (Test #966)
Loey-Dietz syndrome TGFB2 (Test #1159)

Developmental

Weaver syndrome *EZH2 (Test #133)*
Noonan spectrum disorders/Rasopathies NGS-RAS (Test #1309)
Joubert syndrome *TMEM138* (Test #1023)
Cornelia De Lange syndrome
RAD21 (Test #894)
46,XX Disorder of Sex Development WNT4 (Test #904)

Lysosomal

Mucolipidosis Type IV *MCOLN1* (Test #519)

Metabolic / Mitochondrial

Mitochondrial Neurogastrointestinal Encephalopathy *TYMP* (Test #1242)
Ethymalonic Encephalopathy *ETHE1* (Test #1232)
Mitochondrial Disorders *RRM2B* (Test #1246)
Glycine Encephalopathy *AMT* (Test #1209)
Encephalomyopathic form of Mitochondiral DNA Depletion syndrome *SUCGL1* (Test #1249)
Mitochondrial disorders *POLG* (Test #1241)
Fumurase Deficiency FH (Test #1286)

Neurologic

Non-syndromic autosomal recessive mental retardation *TRAPPC9*
(Test #1517)
Mental Retardation *TUSC3 (Test #1516*)
French Canadian type of Leigh syndrome *LRPPRC* (Test #1234)
Autosomal dominant Nocturnal Frontal Lobe Epilepsy *CHRNB2*
(Test #1180)
Neuronal Ceroid-Lipofuscinosis *PPT1* (Test #1427)
Progressive Myoclonic Epilepsy with or without Renal Failure *SCARB2* (Test #1428)
Early Infantile Epileptic Encephalopathy-4/Ohtahara syndrome *STXBP1* (Test #1429)
Amyotrophic Lateral Sclerosis *C9orf72* (Test #151)
Kleefstra syndrome *EHMT1* (Test #1518)
Fumurase Deficiency FH (Test #1286)

Neuromuscular

Distal Arthrogryposis
ECEL1 (Test #1199)
Gnathodiaphyseal Dysplasia *ANO5* (Test #856)
Dystroglycanopathy *B3GALNT2* (Test #1299)
Dystroglycanopathy *B3GNT1* (Test #1198)

Distal Hereditary Motor Neuropathy, Type 7A *SLC5A7* (Test #1197)

Other

Pseudohypoaldosteronism Type 1 NR3C2 (Test #1271)
Pseudohypoaldosteronism Type 1 *SCNN1A* (Test #1272)
Liddle Syndrome and Pseudohypoaldosteronism Type 1 *SCNN1B* (Test #1273)
Congenital Hyperinsulinism *ABCC8* (Test #1221)
Congenital Hyperinsulinism *KCNJ11*(Test #1218)
Congenital Nephrotic syndrome *NPHS1*(Test #1223)
Steroid-Resistant Nephrotic syndrome *NPHS2*(Test #1229)
Hereditary Chronic Pancreatitis *SPINK1* (Test #1401)

Skeletal

Spondyoenchondodysplasia with immune dysregulation *ACP5 (Test #1151*)
Ulnar-Mammary syndrome *TBX3 (Test #1154*)
Spondylocostal Dysostosis *TBX6 (Test #1421)*
Mandibulofacial Dysostosis *EFTUD2 (Test #873)*
Klippel-Feil syndrome *MEOX1* (Test #838)
Achondrogenesis *TRIP11* (Test #1152)
Treacher Collins syndrome *POLR1C* (Test #1110)
Treacher Collins syndrome *TCOF1* (Test #1108)
Treacher Collins syndrome *POLR1D* (Test #1109)
Agnathia-Otocephaly Complex *PRRX1* (Test #1298)
Miller syndrome *DHODH* (Test #1019)
FLNB-related disorders. *FLNB* (Test #1153)
Otopalatodigital Spectrum Disorders, Periventricular Nodular Heterotopia and Cardiac Valvular Dystrophy FLNA (Test #948)

Skin

Autosomal Recessive Congenital Ichthyosis Sanger panel *ARCI* (Test #1160)
Autosomal Recessive Congenital Ichthyosis *ALOXE3* (Test #1164)
Autosomal Recessive Congenital Ichthyosis *CYP4F22* (Test #1163)
Autosomal Recessive Congenital Ichthyosis *NIPAL4* (Test #1166)
Epidermolysis Bullosa with Pyloric Atresia *PLEC* (Test #970)
Autosomal Recessive Congenital Ichthyosis *PNPLA1* (Test #1167)
Autosomal Recessive Congenital Ichthyosis AB*CA12* (Test #1165)
Autosomal Recessive Congenital Ichthyosis *ALOX12B* (Test #1162)
Epidermolysis Bullosa with Pyloric Atresia *ITGA6* (Test #977)
Epidermolysis Bullosa Simplex *KRT5* (Test #978)
Autosomal Recessive Congenital Ichthyosis *TGM1* (Test #1161)
Kindler syndrome *FERMT1* (Test #969)

Epidermolysis Bullosa with Pyloric Atresia *ITGB4* (Test #976)
Ectodermal Dysplasia/Skin Fragility syndrome *PKP1* (Test #968)

Vision

"Bull's Eye" Macular Dystrophy, Cone-Rod Dystrophy 12, Retinitis Pigmentosa 41 and Stargardt Disease 4 *PROM1* (Test #672)
Leber Congenital Amaurosis 4 *AIPL1* (Test #820)
Leber Congenital Amaurosis 1 and Cone-Rod Dystrophy 6 *GUCY2D* (Test #822)
Vitelliform Macular Dystrophy/ Bestrphinopathies *BEST1* (Test #671)
Autosomal Dominant Optic Atrophy, *OPA1* (Test #566)
Autosomal Dominant Optic atrophy with cataaract and Costeff Syndrome or 3-methylglutaconic aciduria, type III OPA3 (Test #567)

Jim Weber, Ph.D.

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President's corner

Disease prevention through genetic testing

Jim Weber, Ph.D.
PreventionGenetics founder and president

I've known of the tight connection between disease prevention and DNA testing for many years (see for example Nature Genetics 7:343-344, 1994). That's why when we started our Company about 10 years ago, I chose the name PreventionGenetics. But after reading the new prevention articles on our website, even I was surprised to learn just how many disorders can be better managed with early and accurate diagnosis.

While complete "cures" are usually not possible, amelioration of Mendelian disease through a whole host of medical interventions is the rule today rather than the exception. As just a few examples, pacemakers can prevent sudden cardiac arrest. Surgery can prevent cancer. Avoiding triggering agents can prevent severe adverse drug reactions. Special diets will often help those with metabolic disorders. I (and many others) think that the ideal approach to health care is one in which health is established at as high a level and as early in life as possible, and then is maintained throughout life with preventive measures.

I invite you to check out our prevention articles. After reading just a few, I think that you will agree that early and accurate disease diagnosis is enormously valuable, and that DNA testing plays a major role in reaching those diagnoses. We really can Prevent Disease through Genetic Testing.

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.