PreventionGenetics

Disease prevention through genetic testing

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Welcome to the April 2010 PreventionGenetics Newsletter. In this issue, we are pleased to introduce Marwan Tayeh, PhD, and a new test panel for Bardet-Biedl syndrome.

The topic for the President's Corner is human gene patenting.

Introducing Marwan Tayeh, PhD

PreventionGenetics is pleased to introduce our newest Clinical Molecular Geneticist, Marwan Tayeh, Ph.D. Dr. Tayeh recently completed his Clinical Molecular Genetics fellowship at Emory University after receiving his PhD in Genetics at the University of Iowa. Dr. Tayeh will be responsible for developing a portfolio of tests that will include among others; ciliopathies, Marfan syndrome, and microarray analysis. Dr. Tayeh recently won the Richard King Award from the American College of Medical Genetics (ACMG) for best publication of the year for a manuscript entitled "Targeted comparative genomic hybridization (CGH) array for the detection of single- and multi- exon gene deletions and duplications", published in the April 2009 issue of Genetics in Medicine.

New Tests at PreventionGenetics

PreventionGenetics is pleased to announce a new test panel for Bardet-Biedl syndrome (BBS). BBS is a pleiotrophic disorder characterized by retinal degeneration, obesity, post-axial polydactyly, cognitive impairment, hypogenitalism, and renal and cardiovascular anomalies. BBS clinical features overlap with a group of diseases known as ciliopathies, which include Meckel-Gruber syndrome, Joubert syndrome, Bardet-Biedl syndrome, nephronophthisis, Senior-Loken syndrome, and Leber congenital amaurosis. These disorders may represent a phenotypic continuum of a single clinical entity.

Similar to other ciliopathies, BBS exhibits locus heterogeneity: 12 BBS genes have been identified to date. PreventionGenetics has recently developed bi-directional gene sequencing tests for all 12 BBS genes. The BBS genes can be ordered as a panel or individually.

The BBS Panel at PreventionGenetics:

Gene	Disease	Percentage of reported BBS mutations
BBS1	BBS	~23%
BBS10	BBS	~20%
BBS2	BBS	~8%
MKKS/BBS6	BBS	~6%
BBS12	BBS	~5%
BBS4	BBS	~3%
BBS7	BBS	~2%
BBS8	BBS	~2%
BBS5	BBS	~2%
ARL6/BBS3	BBS	~1%
BBS9	BBS	~1%
TRIM32/BBS11	BBS	<1%

Overall sensitivity for the BBS panel is at least 70%. Turn around time for the entire panel is a maximum of 100 days.

For more information, please contact Marwan Tayeh, PhD, at 715-387-0484 ext. 108 or email Dr. Tayeh at marwan.tayeh@preventiongenetics.com.

President's Corner

Jim Weber, PhD

Notwithstanding the recent New York Court decision, banning the patenting of human DNA sequences must remain a high priority for the human genetics community. An excellent, thorough discussion of this matter appeared in the February 2010 issue of CAP Today. PreventionGenetics is absolutely opposed to the patenting of human genes or other human DNA sequences for purposes of clinical testing.

Gene patents benefit a miniscule fraction of the population and harm everyone else. Exclusive patent licenses create testing monopolies with accompanying high prices and low quality. Even non-exclusive licenses raise test prices unnecessarily and sometimes prevent labs form offering tests at all. As PreventionGenetics and other labs have amply shown, patent licenses are not required to develop new DNA tests.

Gene patents will also substantially weaken clinical whole exome and whole genome sequencing. Patients will not be pleased to receive "Swiss cheese" genomes with many critical gene sequences removed. Why pay, for example, $2500 for a whole exome sequence and then have to pay many thousands more for the missing genes.

Gene patents are not required to motivate scientists to identify new disease genes. Many new disease genes are identified each year by European scientists, and gene patents are almost entirely ignored by European Clinical Labs. Nearly all disease genes are identified at non-profit institutions. Scientists at non-profit institutions are already strongly motivated by the need to win high profile publications, grants and tenure. They do not require additional motivation from patents.

To my knowledge no legislation was passed initially to allow human gene patents. This was merely a (very unwise) decision on the part of the Patent Office. Although we hope that the ACLU *BRCA* gene lawsuit will ultimately prevail, neither lawsuits nor an act of Congress should be required to reverse the original decision. The Patent Office can simply acknowledge their mistake and stop issuing new patents.

Coming in the June PreventionGenetics Newsletter

Our June newsletter will focus on our new exon deletion/duplication microarray, as well as recent additions to our test menu.

Interested in a test we don't currently offer?

PreventionGenetics already offers one of the largest gene sequencing test menus, and we continue to add new tests. If you are interested in a test we don't currently offer, please contact us. There is a good chance we will develop the test.

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 us at clinicaltesting@preventiongenetics.com.