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DISEASE PREVENTION THROUGH GENETIC TESTING

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Volume 5, Number 1

Welcome to the March 2013 PreventionGenetics newsletter. In this issue, we introduce Dr. Juan Dong. We also present 70 new and updated Sanger sequencing, Sanger panel tests and Next-Gen Panel tests. In the President's Corner, Dr. Jim Weber concludes his three-part series on storage of patient DNA sequences in electronic medical records.

Come see us at AAN, ACMG conferences

If you are planning to attend these two upcoming conferences, be sure to visit the PreventionGenetics booth.
At the ACMG Annual Clinical Genetics Meeting in Phoenix March 19-23, we will feature a brand new, 10 X 20 booth that highlights the many features of our genetic testing services. Come chat with us at booth #616. Members of our Ph.D. team and sales representatives will be available to answer your questions and provide more information on PreventionGenetics.
At the AAN American Academy of Neurology Conference in San Diego March 16-23, we will be at booth #1306. We look forward to speaking with you at the world's largest gathering of neurologists and neuroscience professionals.

New and updated Sanger Sequencing and Panel Tests at PreventionGenetics

We are featuring 70 new and updated genetic tests. Please click on this link to see the full range of tests.

Clinical molecular geneticist joins PreventionGenetics

Juan Dong, M.D., Ph.D., FACMG

Juan Dong, M.D., Ph.D., FACMG, joined PreventionGenetics as a clinical molecular geneticist.

Dr. Dong brings with her more than 11 years of research expertise related to craniofacial genetic disorders at the University Health Science Center at San Antonio and the University of Alabama in Birmingham.

Her portfolio at PreventionGenetics focuses on skeletal, skin and dental genetic disorders such as Ectodermal dysplasia, Epidermolysis bullosa, Cutis Laxa and Dyskeratosis Congenita.
"My passion has always been in medical service," said Dr. Dong. "I chose to work at PreventionGenetics because it offers the best quality of genetic testing to physicians and their patients."
Dr. Dong is board certified in clinical molecular genetics by the American Board of Medical Genetics. She is a Fellow of the American College of Medical Genetics.
She received her medical and master's degrees from Harbin Medical University in China and her Ph.D. degree from the Heidelberg University in Germany. She completed the American Board of Medical Genetics training in clinical molecular genetics at the University of Alabama in Birmingham.

Jim Weber, Ph.D.

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President's Corner

Jim Weber, Ph.D., PreventionGenetics founder and president

DNA sequence storage: Part III

DNA sequences should be kept confidential

Note: This is the final in a series of three articles on the storage of patient DNA sequences in electronic medical records (EMRs). In the first article, I addressed the use of patient sequences in EMRs to automatically reinterpret the clinical significance of the sequences and to provide medical alerts. In the second, I emphasized the importance of sharing sequence information electronically among family members. This article outlines a superior approach for sharing sequence information electronically among family members, and includes my support of the widely shared notion that patient DNA sequences should be kept strictly private.

It's often important for individuals to learn if they carry a pathogenic sequence variant that was originally detected in another member of the family. The pathogenic variant could, for example, predispose a person to cancer, to a severe drug reaction or have reproductive implications.

Today, we rely primarily on family members to transmit such genetic information among themselves. However, the current approach has some serious weaknesses:
* First, many family members simply won't communicate the relevant information. They may be too busy, think it's not important or don't care to make the effort.
* Second, even when family members communicate the information, they will routinely make fundamental mistakes in genetics. For example, they may not understand the difference between "dominant" and "recessive" or grasp concepts of genetic risk. The technical aspects of genetics go well beyond the basic understanding of most people.
* Third, genetic privacy is compromised when family members attempt to communicate this information. The more people who know a person's genotype, the greater the chance that this

information will get back to an employer, banker or college admissions officer. This can result in devastating consequences for the individual through genetic discrimination, whether legal or not.

A much better approach was described in the second article in the series. Sharing of electronic genetic information among interconnected health care providers is clearly superior to the attempts by lay people to communicate this information. Trained health care providers are best suited to transmit genetic information among family members. In addition, through this approach, the identities of family members who carry the causative mutations can be kept private.

People need to have access to their own genomes, but except in a few special cases (particularly in the case of parents who need to know critical sequence variants carried by their children) people need not and should not know the genomes of others. Rather than encouraging patients to reveal their genotypes, we should encourage them to keep this information strictly private. We need to develop a genetic etiquette that emphasizes confidentiality rather than disclosure.

Despite the great potential of patient sequences in EMRs, the current reality is that progress in this area has been slow. For the time being, we must rely on patient-to-patient transmission of genetic information. However, by working together and by being persistent, we can eventually achieve the long-term goal of storage and sharing of patient sequences in EMRs.

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.