



PreventionGenetics

Disease prevention through genetic testing

Volume 1, Number 6

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Welcome to the December 2009 PreventionGenetics Newsletter. We begin the Newsletter by comparing Clinical Test prices at PreventionGenetics to those in other labs. We also present new clinical tests for Distal Arthrogyrosis and Hypertrophic Cardiomyopathy. Finally, in the President's Corner, we discuss how we are able to offer high quality clinical tests at low prices.



We would also like to take this opportunity to wish all of our clients and their patients a very Happy Holidays. We thank you for using our DNA Testing and DNA Banking services in 2009 and we look forward to growing with you in 2010.

Prices of Gene Sequencing Tests at PreventionGenetics

In the following table we compare the median per exon price for gene sequencing from PreventionGenetics and four other major U.S. clinical genetics labs. Prices were collected directly from lab websites in December 2009. Prices at PreventionGenetics are 37-75% of the prices in the other four labs. Nearly three tests could be ordered from PreventionGenetics for the price of a single test at the most costly lab surveyed.

Lab	Median Exon Price
PreventionGenetics	\$48
Lab A	\$64

Lab B	\$118
Lab C	\$123
Lab D	\$129

New Tests at PreventionGenetics

PreventionGenetics is pleased to announce the addition of new gene sequencing tests for Distal Arthrogyriposis and Hypertrophic Cardiomyopathy.

Distal Arthrogyriposis:

Distal Arthrogyriposis (DA) syndromes are a group of multiple congenital contracture disorders with distal joint involvement, variable clinical expression, and autosomal dominant inheritance (Bamshad et al. *Am J Med Genet* 65:277-281, 1996). DA syndromes include Freeman-Sheldon Syndrome (FSS; DA2A, OMIM #193700), TPM-2 related DA (DA1; OMIM#108120), and Sheldon-Hall syndrome (DA2B; OMIM#601680) which is intermediate in features between the first two.

Mutations in the *MYH3* gene appear to be a common cause of Freeman-Sheldon syndrome, with 26 of 28 cases having disease causing mutations. Among 38 Sheldon-Hall syndrome patients that tested negative for *TNNI2* and *TNNT3* mutations, 12 tested positive for *MYH3*. PreventionGenetics offers testing for all three Distal Arthrogyriposis genes: *MYH3*, *TNNI2*, and *TNNT3*.

Hypertrophic Cardiomyopathy:

Hypertrophic cardiomyopathy (HCM, OMIM#192600) is a primary disease of the cardiac muscle characterized by idiopathic hypertrophy of the left ventricle, although hypertrophy of the right ventricle may occasionally occur (Fifer and Vlahakes, *Circulation* 117:429-439, 2008). HCM is inherited in an autosomal dominant manner. It is caused by mutations in various genes encoding sarcomeric proteins. Over 500 different HCM causing mutations have been reported, mostly missense, in eight genes. These mutations account for 74%-94% of

all HCM cases with causative detectable mutations. PreventionGenetics offers tiered testing for all eight genes (see below).

Panel	Genes	HCM Detection
Panel 1	<i>MYH7, MYBPC3</i>	~80%
Panel 2	<i>TNNT2, TNNI, TPM1</i>	~12%
Panel 3	<i>MYL2, MYL3, ACTC1</i>	~3%

PreventionGenetics also offers a number of tests for genes that are very rarely mutated in HCM patients. These include sequencing tests for *TTN, MYH6, TCAP, VCL, ACTN2, PLN, CSRP3, TNNC1,* and *CAV3* genes. For more information on HCM genetic testing, contact [Dr. Khemissa Bejaoui](#).

President's Corner

Jim Weber, PhD

In our previous Newsletter, I described the quality of Clinical DNA Testing at PreventionGenetics. In this Newsletter, I focus on test prices.

I'm pleased that PreventionGenetics is able to offer high quality Clinical DNA Tests at prices that are among the lowest in the industry. We are able to offer such low prices because our relatively large size allows us to realize economies of scale that smaller labs cannot achieve, because the cost of doing business in Central Wisconsin is lower than other parts of the country, and especially because we run an extremely efficient operation. Over many years of experience, we have learned how to maximize productivity at each stage of our laboratory operation.

Our goal is certainly not to put other labs out of business. Although I support vigorous competition, we have never targeted specific labs in our development of new tests. We do not practice predatory pricing.

We do aim, however, to provide the best possible service at the lowest possible price to patients. Through offering low test prices, PreventionGenetics is helping to hold down the cost of health care. Clinical Geneticists can help more patients and order more tests by working with us.

I also call your attention to a recent major overhaul of the PreventionGenetics web site. The content of our site is mostly the same, but the look and feel has been substantially upgraded. I'm also pleased that none of the photos on our web site are stock photos or were created using professional models. We just feature a few of our employees doing their regular jobs.



Coming in the February PreventionGenetics Newsletter

Our next Newsletter will focus on new additions to the PreventionGenetics test menu as well as the inclusion of DNA test information in electronic health records.

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 Chuck Dokken by phone at 715-387-0484 x 107 or email at chuck.dokken@preventiongenetics.com.

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