PREVENTION GENETICS

WHY CHOOSE US?

QUALITY

- Our genotyping panel is run on all specimens, as a quality control measure to detect specimen mislabeling.
- For genes which cannot be accurately analyzed by NextGen sequencing due to paralogy, we supplement the NextGen data with special Sanger sequencing tests. Such genes include *PKD1, GBA, PMS2* and *NEB*.
- Average coverage data for clinically relevant genes is available through the Custom Panels Tool on our website.
- All targeted prenatal testing is performed in duplicate by different lab personnel.
- If short tandem repeat regions cannot be accurately assessed using nextgen data, special sizing assays may be run.
- Brief preventive medicine summaries about specific genes and associated disorders are available on our website as resources for healthcare providers. These summaries may be particularly useful when preparing letters of medical necessity (LMN). Several LMN templates are also available on our website.

VALUE

- Free testing for variants of uncertain significance is provided for up to two family members of probands who received full gene, panel or exome sequencing at PreventionGenetics.
- All uncertain aCGH and CMA results are confirmed by PCR or another method. Breakpoints are identified in many cases through PCR. We then offer free testing for up to two family members and more cost effective PCR tests for additional family members.
- Maternal cell contamination studies are offered at no additional cost for prenatal testing.
- For chromosomal microarray (CMA) tests, we routinely report candidate genes for recessive disease which lie in regions with absence of heterozygosity.
- We analyze all exome-based NextGen sequencing data for copy number variants (CNVs). All CNV's are confirmed using a different methodology such as PCR or aCGH.
- Free reflex to the full panel when any subset of genes is ordered from one of over 200 predefined panels on our test menu.
- We provide free interpretation for any variant in any clinically relevant gene through our Variant Interpretation Service.
- One reanalysis and reinterpretation of PGxome data within two years of the original test report is available at no cost. Additional reanalysis/reinterpretation is available beyond two years for an additional fee.
- Patients that undergo testing at PreventionGenetics receive a discounted rate on DNA Banking.
- We retain excess DNA from patient testing so that it is available for future testing and for quality control. This saves considerable phlebotomy and shipping costs.

SERVICE

- All of our test results and reports are carefully reviewed by at least four highly trained and experienced individuals, including two doctorate geneticists.
- For PGxome® tests, we distribute the variants to our PhD and MD experts, each of whom focuses on a specific subset of the genome. This results in more efficient and accurate variant interpretation.
- We combine *in silico* algorithms with manual review for the best possible variant interpretations. All sequence variants that are not common are interpreted manually by our doctorate geneticists.
- Wherever practical, we utilize quantitative methods to supplement qualitative interpretation of sequence variants.
- PreventionGenetics' laboratory stewardship program saves patients over $1 million per year through review of all incoming test orders. Our team works with clinical providers to ensure the right test is ordered for every patient.
- PreventionGenetics retains complete patient sequences for reinterpretation and to benefit family members. This data is available for eventual transfer into patient electronic health records.
- We have deposited many sequence variants found in our patients to the NCBI ClinVar database and will submit more in future.
- We make all of our test data available for research purposes (while protecting patient confidentiality).

FLEXIBILITY

- Custom Panels allow you to build a panel from any of ~5,000 clinically relevant genes. Many genes can also be enhanced to increase coverage. Pricing is tiered by number genes, regardless of clinical area.
- Any PGxome-based panels, including Custom Panels, can be reflexed to whole exome sequencing for the difference in cost.
- Institutional, insurance and self-pay billing options.

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