Quality & Value

QUALITY MEASURES

- Only two errors over 15 years of laboratory operation out of >15,000 Proficiency Tests, both external and internal.
- Comparison of our Sanger sequencing to NextGen sequence data: Four errors out of 14 Mb sequenced (~9,000 sequence variants) (all four errors were the result of allele dropout during PCR).
- 12 specimen labeling errors out of ~70,000 opportunities, but only one of which led to an incorrect report.
- Comparison of our NextGen sequencing to Sanger sequence data and to NextGen sequence data from other labs: One error out of 7.0 Mb sequenced (~5,500 sequence variants).

DISTINGUISHING FEATURES

1. All of our test results and reports are carefully reviewed by at least four highly trained and experienced individuals, including two doctorate geneticists.
2. In our PGxome® tests, for variant interpretation, 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.
3. All sequence variants that are not common (< 10% allele frequency) are interpreted manually by our doctorate geneticists. We do not rely solely on automated approaches.
4. Wherever practical, we utilize quantitative methods for interpretation of sequence variants. We do not rely solely on qualitative approaches.
5. We apply Utilization Management to all of the specimens which come through our doors. Through this process we have saved and will continue to save patients over $1 million per year.
6. Essentially all staff who work on tests and test reports are on site in Marshfield, including our counselors and doctorate geneticists. This facilitates communication and continuing education.
7. Our genotyping panel is run on all specimens, as a quality control measure, to detect specimen mislabeling.
8. For genes which cannot be accurately analyzed by NextGen sequencing due of the presence of pseudogenes, we supplement the NextGen data with special Sanger sequencing tests. Such genes include *PKD1, GBA, PMS2* and *NEB*.
9. All targeted prenatal testing is performed in duplicate by different lab personnel.
10. We provide free interpretation for any variant in any clinically relevant gene through our Variant Interpretation Service. We make all of our test data available for research purposes (while protecting patient confidentiality). We have deposited many sequence variants found in our patients to the NCBI ClinVar database and will submit more in future.
11. We are transparent with test prices on our web site.
12. We analyze nearly all NextGen sequencing data for copy number variants (CNVs). The sensitivity of this approach for CNV detection is still lower than aCGH, but is steadily improving. All CNV calls are confirmed using a different methodology such as PCR or aCGH.
13. 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. Patients that undergo testing at PreventionGenetics receive a discounted rate on DNA Banking.
14. PreventionGenetics retains complete patient sequences for reinterpretation and to benefit family members. This data is available for eventual transfer into patient electronic health records.
15. Free testing for variants of uncertain significance is provided for up to two family members of probands who received full gene sequencing at PreventionGenetics.
16. Special sizing assays are run for regions of short tandem repeats within or near coding regions. Examples are the (T)9 sequence within exon 9 of the *ZMPSTE24* gene and the (TG)11 (T)7 sequence in intron 8 of the *CFTR* gene. Such repeat regions cannot be accurately determined by sequencing alone.
17. For chromosomal microarray tests, we routinely report candidate genes for recessive disease which lie in regions with absence of heterozygosity.
18. Maternal cell contamination studies are offered at no additional cost for fetal testing.
19. All uncertain gene-centric aCGH results are confirmed by PCR or another method. Breakpoints are identified in many cases through sequencing of the PCR products. We then offer more cost effective PCR tests for family studies.
20. Brief preventive medicine summaries about specific genes and associated disorders have been placed on our web site to act as resources for healthcare providers. These summaries may be particularly useful when preparing letters of medical necessity.

Value = Quality / Cost

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