

Whole Exome Sequencing Re-analysis

Description:

The Genetics and Genomics Diagnostic Laboratory at Cincinnati Children's will reanalyze data generated by our ExomeSeq test at the request of the ordering provider within five years from the date of the original exome case submission. There are multiple indications for ExomeSeq re-analysis:

- New gene(s) may be reported in the literature that are associated with the patient's phenotype; or new gene functions may be reported in the literature that establish new phenotype-genotype correlations.
- Phenotypes may evolve over time and patients may present with new symptoms after their ExomeSeq results are completed. Ordering providers may request re-examination of the existing variants for genes associated with new phenotype(s).
- If inadequate coverage was attained for genes of interest during initial analysis, the laboratory will recommend customized Sanger sequencing for these genes.

Note: If inclusion of a new sibling is requested, his/her sample will be used for confirmation only.

Ordering Requirements:

- Submission of a completed ExomeSeq Re-analysis Request form to the Genetics and Genomics Diagnostic Laboratory.

Sample Requirements:

- A new sample may be required for confirmation studies if the initial ExomeSeq was performed more than 2 years ago, or if the sample was depleted during testing.

Results:

- A new report will be issued for either negative or positive results.

Turn-around time:

- 16 weeks

CPT Codes:

- Whole Exome Re-analysis (Proband only): **81417**
- Whole Exome Re-analysis (Duo): **81417**
- Whole Exome Re-analysis (Trio): **81417**
- Whole Exome Re-analysis (Extra Family Member): **81417**

For questions, please call 513-803-5390.



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