

EXOMESEQ

Whole Exome Sequencing Re-analysis of Data:

The Molecular Genetics 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:

A. 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.

1. The referring provider may request gene coverage information from the patient's existing data for up to five gene(s) of interest. The laboratory will examine original data free of charge to assess coverage for these genes.
2. The referring provider may request analysis of up to five genes of interest at coverage obtained during original order.
3. If inadequate coverage was attained during initial analysis, the laboratory will recommend customized Sanger sequencing for these genes.

B. Phenotypes may evolve over time and patients may present with new symptoms after their ExomeSeq results are completed.

- For these situations, ordering providers may request re-examination of the existing variants for genes associated with new phenotype(s).

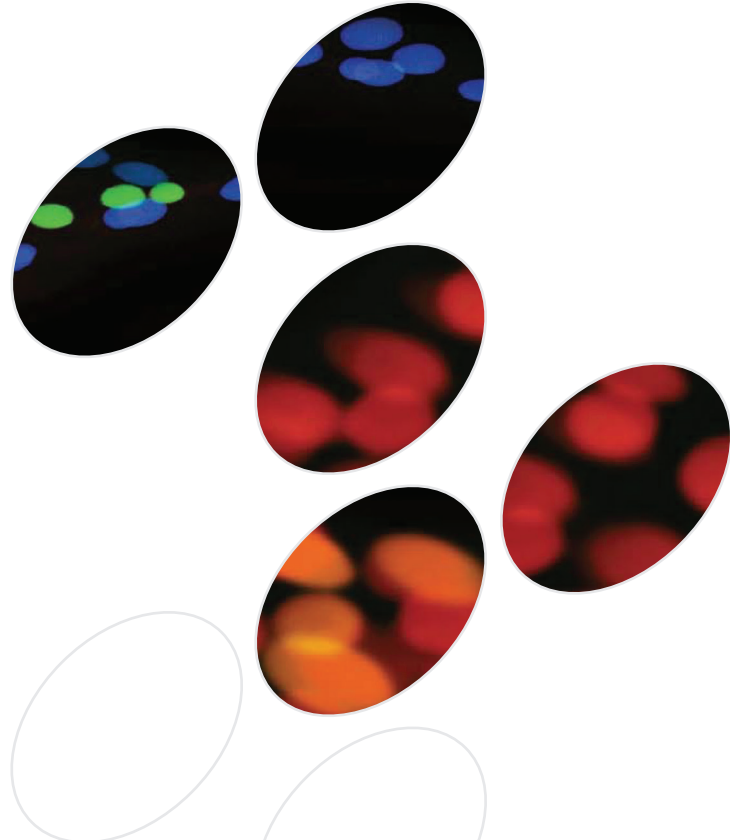
Note: If inclusion of a new sibling is requested, his/her sample will be used for confirmation only.

Requirements for Requesting ExomeSeq Re-analysis:

- Submit completed ExomeSeq Re-analysis Request form to Molecular Genetics Laboratory.
- 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.

CPT Codes:

- Coverage inquiry: N/A
- Sequencing for low-coverage areas of new gene(s) of interest: gene-specific, please call the lab to confirm CPT code(s)
- Re-analysis: 81417 (x1) for proband only, 81417 (x3) for trio



What you will receive:

Types of Exome Re-analysis	Results Issued	Turn-around Time
A1. Coverage inquiry	A brief report will be issued with coverage information for up to five genes of interest.	4 weeks
A2. Analysis of up to five genes of interest at coverage obtained during original order.	A new report will be issued for either negative or positive results.	16 weeks
A3. Custom gene sequencing for genes with low coverage during initial test.	A new report will be issued for either negative or positive results.	16 weeks
B. Complete re-analysis if new symptoms have developed	A new report will be issued for either negative or positive results.	16 weeks

For questions, please call 513-803-5390.