

Negative exome or genome sequencing results

Information for individuals with a negative result

Testing performed

Exome and genome sequencing look at many components of a person's DNA, including approximately 20,000 genes, to assess for changes (also called variants) that may contribute to, or explain, an individual's symptoms or medical history.

Test result

- **This test did not identify any genetic changes that explain the symptoms or medical concerns reported in the individual. Therefore, a genetic cause for the individual's condition was not identified. This is considered a non-diagnostic result. Possible reasons why a change was not identified include, but are not limited to:**
- The individual has a genetic cause for their symptoms that was not identified by this test.
 - ◊ There may be genetic changes that are outside of the regions analyzed.
 - ◊ There may also be genetic changes that are not recognized as causative for the individual's medical history. This may be due to insufficient knowledge about the variant or the gene and its function.
- Further testing or evaluation of the individual, additional time for the individual to grow and develop, and/or additional evidence in the scientific literature may be needed to identify a genetic cause for the individual's symptoms. Future reanalysis of the individual's genetic data, combined with updated clinical and family histories, may enable a genetic diagnosis in the future. Talk to your healthcare provider about requesting a reanalysis in the future.
- A copy of the results has been sent to the healthcare provider who ordered this test. Please contact them to obtain a copy of the results.

Management

- Follow up with a healthcare provider regarding symptoms and any additional questions. Please speak with the individual's healthcare provider about requesting a reanalysis in the future. Even in the setting of negative results, changes in care may be indicated based on symptoms and/or family history.
- In some cases, an evaluation by a medical genetics provider, to include a physical examination and review of the medical and family histories, should be considered for the diagnosis and management of a possible genetic condition, and to guide further genetic investigation. A copy of your results has been sent to your provider.