Exome with CNV Evaluation

Quest Diagnostics™

Ordering guide

1 Order the Exome with CNV Evaluation in 1 of 3 ways

- Electronic ordering using Quanum™ eLabs
- An interfaced EMR
- Paper requisition

There are 3 test options:

Trio ⋮ Duo ⋮ Proband Only

Select the appropriate test for your patient based on the available and consenting family members. If you select Duo or Trio evaluation, please complete an additional requisition form for each family member with the Order Code 36939.

Notes

- If you have already obtained preauthorization, indicate on the electronic order or the paper requisition that preauthorization has been completed
- Provide the authorization number and a copy of the authorization when possible
- You can get copies of the Exome with CNV Evaluation paper requisition from your Quest Diagnostics account manager

2 Before blood draw occurs, fill out the Clinical History form in its entirety and have the patient sign the Consent form

- Both forms can be found at QuestExome.com or from your Quest Diagnostics account manager
- Send in the completed Clinical History form and Consent form with the requisition and the sample OR you can fax the forms before blood draw to the Prior Authorization Services team at 1.949.668.7818

3 Collect the patient specimen in the office and send with the requisition by a Quest Diagnostics courier, or send the patient to one of our more than 2,200 Patient Service Centers with the requisition to have the specimen collected

All orders must include proper documentation of patient and family history. Incomplete information on an order **will result** in delays and will require additional communications with your office to collect the necessary information.

Please consider doing a preauthorization before specimen collection. Fax the requisition and Clinical History form, or any supporting documentation, to the Prior Authorization Service team at 1.949.668.7818 to ensure the fastest turnaround time.

Complete information on an order includes:

- ○ Clinician name and NPI number
- ○ Patient billing and insurance info (if needed)
- ○ Patient address and phone number
- ○ Patient date of birth
- ○ ICD-10 codes
- ○ Clinical History form
- ○ Consent form
- ○ Test code selection
- ○ Patient signature
- ○ Physician signature (unless blanket form is on file)
- ○ Additional requisitions for family members (only for Duo and Trio orders)

For any questions, **please call 1.866.GENE.INFO (1.866.436.3463)** to speak to a Genomics Science Specialist or a member of the Prior Authorization Service team; or **visit QuestExome.com**

What to expect after ordering:

- The samples will be shipped to our laboratory, accessioned, and stabilized. Our Genomic Science Specialists are available to answer any questions you may have before or after ordering at 1.866.GENE.INFO (1.866.436.3463).
- If ordering with third-party insurance and you have not already received preauthorization, our Prior Authorization Services team will begin benefits investigation to discover if your patient needs a preauthorization before testing and what requirements are needed. The team may call your office if additional information is needed.

  If allowed by the health plan, we will submit all the required information for preauthorization. Please note that this process can take days to weeks to complete depending on the health plan.
- Once benefits investigation and/or preauthorization are complete, we will call the patient if the out-of-pocket amount is over $350.

  If over $350, we will discuss the patient's options from payment plans to income-based financial assistance.

  Once the patient has agreed to the out-of-pocket amount (including financial assistance and/or payment plans where applicable), or if the amount was less than $350, our laboratory will begin testing.
- The turnaround time for this test is expected to be 6 to 8 weeks once testing is initiated.
- Each case goes through a rigorous review process:
  - Genetic counselor review of clinical and family history
  - Two separate geneticist reviews
  - Thorough variant vetting and analysis by our team of in-house variant scientists, VariantIQ™
  - Case conference including our genetic and medical experts across the country
- Results can be delivered electronically through your interfaced EMR or Quanum™ eLabs. Faxing and secure emails are also available.

What types of results you can expect to receive?

Positive or likely positive

A positive or likely positive result indicates that the laboratory has found a genetic alteration that may explain the patient's disorder. It can also indicate that additional family members may be at risk for a disorder and may require follow-up testing.

Negative

A negative result indicates that the laboratory did not identify the genetic cause of the patient's disorder. This does not mean that the cause of the disease is not genetic. It also does not mean that the patient will be healthy or free of any genetic diseases or medical conditions. This could mean that the exome technology was unable to detect the alteration or that medical science has not yet associated an alteration with disease. Further testing may be necessary to diagnose the condition. Reanalysis of exome data can be ordered one year after the initial results by speaking with a Genomic Science Specialist at 1.866.GENE.INFO (1.866-436-3463).

Uncertain

An uncertain test result indicates that there was a genetic alteration detected, but it is currently unknown if this change would be causative. For example, an alteration could occur in a gene that significantly overlaps with the medical condition of the patient, but the particular difference found is not yet known to have an effect. This is not considered to be a positive result and does not clarify the patient's medical condition or risk. There may be additional information or testing necessary in order to clarify the inconclusive result.

For questions regarding variant classification updates, please call Quest Diagnostics Genomic Client Services at **1.866.GENE.INFO (1.866.436.3463)** or visit **QuestDiagnostics.com/VariantIQ**

The classification and interpretation of the variant(s) identified reflect the current state of Quest Diagnostics' understanding at the time of the report. Variant classification and interpretation are subject to professional judgment, and may change for a variety of reasons, including but not limited to updates in classification guidelines and availability of additional scientific and clinical information. This test result should be used in conjunction with the healthcare provider's clinical evaluation.

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