Blueprint Genetics

Familial Variant Testing

Test code: FVT001

- Diagnostic testing in affected family members
- Predictive testing in unaffected family members
- Carrier testing in the case of autosomal recessive and X-linked disorders
- Segregation of variants

With Familial Variant Testing, you can order for up to 10 variants per order for a fixed price.

Blueprint Genetics will only report the variant(s) of interest. If the individual being tested is suspected of being affected with an inherited disorder, then another more comprehensive test (single gene, panel, WES) may be appropriate.

All variants previously identified at Blueprint Genetics or another diagnostic laboratory are eligible for this service.

Please contact your local Genetic Services Consultant or our Customer Support team to assist with ordering. We are here to help you!

About Familial Variant Testing

Summary

- To assess for the presence or absence of known variant(s) in family members
  - Includes variants identified at Blueprint Genetics or another laboratory
  - Includes predictive testing in unaffected family members
- Carrier testing for autosomal recessive and X-linked disorders
- Allows for segregation of variants
  - For example, demonstrating that a variant is present in affected family members and absent in unaffected family members

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How to order

- Log in to our online portal Nucleus at nucleus.blueprintgenetics.com for the easiest way to order and access results.
- Fill out both the consent and requisition forms.
- Send the sample to Blueprint Genetics.
  - Sample Requirements:
    - Blood (min. 1ml) in an EDTA tube
    - Extracted DNA, min. 2 µg in TE buffer or equivalent
    - Saliva (Oragene DNA OG-500 kit/OGD-500 or OG-575 & OGD-575)
    - Read more about our sample requirements
  - Sending a positive control sample from the index patient is not required.

Methods

Familial Variant Testing is done with Next Generation Sequencing (NGS) technology and/or Sanger sequencing and

qPCR/digital PCR.

Test strengths & limitations

- CLIA-certified personnel performing clinical testing in a CLIA-certified laboratory
- Powerful sequencing technologies, advanced target enrichment methods and precision bioinformatics pipelines ensure superior analytical performance
- Comprehensive clinical statement

Test limitations

Familial Variant Testing for CNVs and testing of prenatal samples are only available for cases where the index patient was tested at Blueprint Genetics. For further information on limitations, please contact our Support team.

Familial Variant Testing may not reliably detect the following:

- Low level mosaicism

Availability

4 weeks

Test Strengths

The strengths of this test include:

- CAP accredited laboratory
- CLIA-certified personnel performing clinical testing in a CLIA-certified laboratory
- Powerful sequencing technologies, advanced target enrichment methods and precision bioinformatics pipelines ensure superior analytical performance
- Careful construction of clinically effective and scientifically justified gene panels
- Our Nucleus online portal providing transparent and easy access to quality and performance data at the patient level
- Our publicly available analytic validation demonstrating complete details of test performance
- ~2,000 non-coding disease causing variants in our clinical grade NGS assay for panels (please see 'Non-coding disease causing variants covered by this panel' in the Panel Content section)
- Our rigorous variant classification scheme
- Our systematic clinical interpretation workflow using proprietary software enabling accurate and traceable processing of NGS data
- Our comprehensive clinical statements

Test Limitations

This test does not detect the following:

- Complex inversions
- Gene conversions
- Balanced translocations
- Mitochondrial DNA variants
- Repeat expansion disorders unless specifically mentioned
- Non-coding variants deeper than ±20 base pairs from exon-intron boundary unless otherwise indicated (please see above Panel Content / non-coding variants covered by the panel).

This test may not reliably detect the following:

- Low level mosaicism (variant with a minor allele fraction of 14.6% is detected with 90% probability)
- Stretches of mononucleotide repeats
- Indels larger than 50bp

- Single exon deletions or duplications
- Variants within pseudogene regions/duplicated segments

The sensitivity of this test may be reduced if DNA is extracted by a laboratory other than Blueprint Genetics.

For additional information, please refer to the Test performance section and see our Analytic Validation.

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Eligibility for free Familial Variant Testing

Five family members are eligible for free testing for up to 10 variants. Eligibility criteria are as follows:

- The proband had a panel test performed at Blueprint Genetics.
- Institutional billing was selected for the proband's test.
- The variant(s) being tested must be classified as either pathogenic, likely pathogenic, or as a variant of uncertain significance and be reported in the primary variant table in the clinical statement. Variants reported in the additional findings table are not eligible for free family variant testing but can be tested as part of our variant specific testing service.
- The individuals are blood relatives of the proband.
- The FVT orders are placed within 90 days and samples are received within 100 days of the proband's result.
- The order is inputted via the normal FVT ordering page or paper requisition and the eligibility is checked automatically.

Available for panel orders placed after September 1st, 2019.

Accepted sample types

- EDTA blood, min. 1 ml
- Purified DNA, min. 3µg* in TE buffer or equivalent
- Saliva (Oragene DNA OG-500 kit)

Label the sample tube with your patient's name, date of birth and the date of sample collection.

Note that we do not accept DNA samples isolated from formalin-fixed paraffin-embedded (FFPE) tissue.