

ORDER NAME

EPIC- Known Mutation/Familial Variant (Order Code=LAB13615)

Cerner- Known Mutation/Familial Variant

- If EPIC or Cerner is not an option, complete and submit a paper requisition to Genomicslab@upmc.edu
Paper requisition can be found on the INFONET. Search for “known familial variant form.”

BACKGROUND

Known familial variant (KFV) testing may be indicated when a pathogenic variant or likely pathogenic variant in a disease-associated gene has been identified in a blood relative. Individuals may proceed with targeted testing of the same variant.

INDICATIONS FOR TESTING

- Diagnostic
- Predictive
- Carrier status- reproductive risk

RESULT

Likely pathogenic or pathogenic variant

METHOD

Method is dependent on variant being tested: Polymerase Chain Reaction (PCR), quantitative droplet digital PCR, or next-generation sequencing.

LIMITATIONS

Limitations are dependent on the specific assay performed.

SPECIMEN REQUIREMENTS

- Whole blood – EDTA tube required, 3-5 ml
- Previously extracted DNA (concentration >25 ng/ul, volume >20 ul, minimum of 1 ug total DNA, 260/280>1.7)
- Saliva provided in Oragene (OGD-500) collection kits

TURNAROUND TIME

Up to 21 days

CPT CODE

Need to confirm if gene is assigned a specific CPT code.
Default code= 81403