Northwest Clinical Genomics Laboratory

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Name: Gender: DOB: ID(s): Date Received: Report date:

Cancer Health Assessment Reaching Many (CHARM) Study

Family Member Genetic Testing Results

This individual has enrolled in the CHARM Study which is providing testing for genetic variants identified in families. This test was for a single genetic variant; other genetic variation may be present (see limitations). **Genetic counseling is recommended and was offered through the CHARM study.**

A guide to interpreting genomic test reports can be found here: <u>http://www.ashg.org/education/csertoolkit/index.html</u>

Indication: Family member with [pathogenic variant *or* likely pathogenic variant *or* variant of uncertain significance] variant in the [gene name] gene associated with [condition].

Results: NORMAL RESULT.

THIS PARTICIPANT <u>DOES NOT HAVE</u> THE [PATHOGENIC VARIANT *or* LIKELY PATHOGENIC VARIANT *or* VARIANT OF UNCERTAIN SIGNIFICANCE] IDENTIFIED IN THEIR RELATIVE.

The specific variant tested for was c.XXX, p.XXX in the [gene name] gene which is associated with [features of condition]. ****If VUS****[This variant was tested to help interpret its clinical significance and this result does not inform cancer screening. Cancer screening for this participant should be based on their personal and family history of cancer].

OR

Results:

THIS PARTICIPANT HAS THE [PATHOGENIC VARIANT/LIKELY PATHOGENIC VARIANT/VARIANT OF UNCERTAIN SIGNIFICANCE] IDENTIFIED IN THEIR RELATIVE.

The specific variant tested for was c.XXX, p.XXX in the [gene name] gene which is associated with [features of condition] ***/*If VUS**** [This variant was tested to help interpret its clinical significance and this result does not inform cancer screening. Cancer screening for this participant should be based on their personal and family history of cancer. The vast majority of Variants of Uncertain Significance will eventually be reclassified as benign]

If positive Further Interpretation

GENE, VARIANT, rsID

Gene and Chromosome	Variant Position (hg19)	Nucleotide variant (1 = the A of the initiator methionine codon)	Protein variant (1 = the initiator methionine, the first translated amino acid of the precursor protein)

[Description of variant]

[Description of evidence supporting interpretation]

Diagnostic <u>Recommendations:</u> Genetic counseling is recommended to discuss the implications of this finding for the patient and their family. *If applicable* [NCCN guidelines for the management of patients with [condition] are available.]

Incidental finding <u>Recommendations:</u> Genetic counseling is recommended to discuss the implications of this finding for the patient and their family.

References:

Test: Sequencing of known familial variants

Method:

DNA was extracted from a submitted specimen. Primers based on the human genome reference sequence were used to amplify the genomic region harboring the known familial sequence variant. Amplified fragments were sequenced using standard dye-terminator chemistry and separated using an Applied Biosystems 3500 Genetic Analyzer. Sequence data (ABI files) was analyzed with the most recent version of Mutation Surveyor (SoftGenetics).

This laboratory test was developed and its performance characteristics determined by the Northwest Clinical Genomics Laboratory (CLIA-certified/CAP-accredited). Consistent with laboratory-developed tests, this test has not been cleared or approved by the U.S. Food and Drug Administration.

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