

TEST CHANGE

Exome Sequencing

3016583, EXOME PRO

Specimen Requirements:

Patient Preparation:

Collect: Lavender or pink (EDTA) or yellow (ACD solution A or B). Peripheral blood required. Contact ARUP's genetic counselor at 800-242-2787 ext. 2141 prior to test submission. Refer to EXOME FRPT (ARUP test code 3016589) for parental specimen requirements. Two parental controls are recommended for EXOME PRO. Controls ~~These~~ should be ordered using EXOME FRPT (ARUP test code 3016589) and submitted within 7 days of the probands sample. ~~above.~~ New York State Clients: ARUP cannot facilitate testing for New York patients. Please work directly with a New York-approved laboratory.

Specimen Preparation: Transport 2mL whole blood (Min 1.0mL) Refer to EXOME FRPT (ARUP test code 3016589) for parental specimen requirements.

Transport Temperature: Refrigerated. Refer to EXOME FRPT (ARUP test code 3016589) for parental specimen requirements.

Unacceptable Conditions:

Remarks: DNA extraction will be performed on the proband and comparator samples upon receipt to ensure stability. DNA Extract and Hold (ARUP test 3005714) will be added to each sample by ARUP. A DNA extraction fee will only be charged once per sample.

Stability: Ambient: 72 hours; Refrigerated: 1 week; Frozen: Unacceptable

Methodology: Massively Parallel Sequencing

Performed: Varies

Reported: 21-28 days

Note: The ability to identify causative variant(s) for the patient's presentation is strongly influenced by the quality of the clinical information provided.

CPT Codes: 81415: per familial comparator, 81416 is added

New York DOH Approval Status: Specimens from New York clients will be sent out to a New York DOH approved laboratory, if possible.

Interpretive Data:

Refer to report.

Reference Interval:

N/A

HOTLINE NOTE: There is a reflexive pattern change associated with this test. One or more orderable or component has been added or removed to the reflexive pattern. Refer to the Hotline Test Mix for interface build information.