

TEST CHANGE

Exome Sequencing

3016583, EXOME PRO

Specimen Requirements:

Patient Preparation:

Collect: ~~Whole blood in lavender or pink (EDTA) or yellow (ACD solution A or B). 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 should be ordered using EXOME FRPT (ARUP test code 3016589) and submitted within 7 days of the proband's sample.~~

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

Unacceptable Conditions:

Remarks: ~~Refer to Exome Sequencing, Familial Control (ARUP test code 3016589) for comparator specimen requirements. Parental comparator samples are recommended for optimal exome sequencing analysis. Comparator samples must be submitted within 7 days of the proband's sample.~~

~~Secondary findings are reported for the proband and comparators unless "opt out" is selected on the proband's Genome Sequencing Intake Form.~~

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

Methodology: Massively Parallel Sequencing

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

~~Contact ARUP's genetic counselors at 800-242-2787 ext. 2141 with questions about test submission.~~

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:

Refer to report ~~N/A~~
