

2006340

Exome Sequencing, Familial Control

TRACKEXADD



Family Member Control History for Exome Sequencing



Informed Consent for Exome Sequencing

Methodology: Massively Parallel Sequencing
Performed: Varies
Reported: 4-8 weeks

Specimen Required: Collect: Lavender (K₂EDTA) or Yellow (ACD Solution A or B). Peripheral blood required. Contact ARUP's genetic counselor at (800) 242-2787 ext. 2141 if there are questions prior to test submission.

Specimen Preparation: Transport 3 mL whole blood. (Min: 1 mL)

Storage/Transport Temperature: Refrigerated.

Remarks: A completed exome consent form and patient history form is required on each parent or family member submitting a control sample.

Stability (collection to initiation of testing): Ambient: 72 hours; Refrigerated: 1 week; Frozen: Unacceptable

Reference Interval: By report

Note: Up to four family members may serve as exome sequencing controls for a proband having Exome Sequencing, Trio (2006332). Family members samples are used to aid in interpretation of the proband's exome sequencing data. Secondary findings, including pathogenic variants in genes recommended for analysis by the American College of Medical Genetics (ACMG) and other actionable secondary variants will only be reported for control individuals who complete a separate Exome Sequencing consent form. ACMG genes are only analyzed to the extent that routine exome analysis allows and single disease-causing variants in autosomal recessive ACMG genes are not reported.

CPT Code(s): N/A