

TEST CHANGE

Genome Sequencing

3019943, WGS PRO

Specimen Requirements:

Patient Preparation:

Collect: Preferred: Whole blood in lavender (EDTA) or pink (EDTA)
Acceptable: Oragene(TM) saliva collection kit ([ARUP Supply #66513, available online through eSupply using ARUP Connect\(TM\) or contact ARUP Client Services at \(800\)522-2787](#)); or equivalent saliva collection device [suitable for human DNA extraction](#), collected in accordance with manufacturer instructions. ~~Saliva in collection device suitable for human DNA extraction.~~
New York State Clients: ARUP cannot facilitate testing for New York patients. Please work directly with a New York-approved laboratory.

Specimen Preparation: Transport 2 mL whole blood (Min: 0.5 mL) or 2 mL saliva.

Transport Temperature: Refrigerated.

Unacceptable Conditions:

Remarks: Refer to Genome Sequencing, Familial Comparator (ARUP test code 3019951) for comparator specimen requirements.

Parental comparator samples are recommended for optimal whole genome 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: Qualitative 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.

[When testing cord blood specimens, the presence of maternal cell contamination \(MCC\) is possible, which may impact result interpretation. If clinically warranted, testing for MCC is available, at a charge, through ARUP Laboratories.](#)

CPT Codes: 81425; 81460; add 81426; 81460 per familial comparator

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:

HOTLINE NOTE: There is a price change associated with this test. Please contact ARUP Client Services at (800) 522-2787 for additional information.