

New Test

3005721

Hereditary Erythrocytosis Panel, Sequencing

ECYT NGS



Additional Technical Information



Patient History for Hereditary Erythrocytosis Testing

Methodology: Massively Parallel Sequencing
Performed: Varies
Reported: 3 weeks. If specimen is a skin punch biopsy, add 2 weeks for culturing.

Specimen Required: Patient Prep:

Collect: Cultured skin fibroblasts (preferred) or Whole blood: Lavender (EDTA) or yellow (ACD solution A or B). or Skin punch biopsy: Thaw media prior to tissue inoculation. Place skin punch biopsy in a sterile, screw-top container filled with tissue culture transport medium (ARUP Supply #32788). Available online through eSupply using ARUP Connect. If cytogenetics tissue media is not available, collect in plain RPMI, Hanks solution, sterile saline, or Ringers.

New York State Clients: Only whole blood, lavender (EDTA) tube, refrigerated

Specimen Preparation: Cultured skin fibroblasts: 2 T-25 flasks at 80 percent confluency, Fill flasks with culture media. Backup cultures must be maintained at the client's institution until testing is complete.

Skin punch biopsy **DO NOT FREEZE**. Do not place in formalin. Transport a 4 mm skin biopsy in a sterile, screw-top container filled with tissue transport medium.

Whole blood: Transport 3 mL (Min: 2 mL)

New York State Clients: Specimens must be received at performing laboratory within 48 hours of collection. For specimen requirements and direct submission instructions please contact ARUP Referral Testing at (800) 242-2787, ext. 5145.

Storage/Transport Temperature: Cultured skin fibroblasts: Critical room temperature. Must be received within 48 hours of shipment due to lability of cells

Skin punch biopsy: Room temperature

Whole Blood: Refrigerated.

Remarks: Cultured skin fibroblast backup cultures must be retained at the client's institution until testing is complete. Skin punch biopsies can be cultured at ARUP at an additional charge.

Unacceptable Conditions: Grossly hemolyzed or frozen specimens; formalin fixed tissue, FFPE

Stability (collection to initiation of testing): Cultured skin fibroblasts: Ambient: 48 hours; Refrigerated: Unacceptable; Frozen: Unacceptable,

Skin punch biopsy: Ambient: 48 hours; Refrigerated: 48 hours; Frozen: Unacceptable

Whole blood: Ambient: 72 hours; Refrigerated: 1 week; Frozen: Unacceptable

New York State Clients: Only whole blood: Ambient: 48 hours; Refrigerated: 2 weeks; Frozen: Unacceptable

Reference Interval: By report

Interpretive Data:

Refer to report.

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the U.S. Food and Drug Administration. This test was performed in a CLIA-certified laboratory and is intended for clinical purposes.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.

Note: Genes tested: *BPGM*, *EGLN1 (PHD2)*, *EPAS1 (HIF2A)*, *EPOR*, *HBB*, *HIF1A*, *JAK2*, *SH2B3*, *VHL**

*One or more exons are not covered by sequencing for the indicated gene; see Additional Technical Information.

If a skin punch biopsy is submitted, specimen will be reflexed for culturing. Additional charges apply.

CPT Code(s): 81364, 81404; 81479; for skin punch biopsy, add 88233

New York DOH approval pending. Call for status update.

HOTLINE NOTE: Refer to the Test Mix Addendum for interface build information.