

**TEST CHANGE**

**Hereditary Erythrocytosis Panel, Sequencing**

3005721, ECYT NGS

**Specimen Requirements:**

**Patient Preparation:**

**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(TM).- If cytogenetics tissue media is not available, collect in plain RPMI, Hanks solution, sterile saline, or Ringers.  
New York State Clients: Whole blood: Lavender (EDTA)

**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.  
If ARUP receives a sample below the minimum confluence, Cell Culture for Genetic Testing (3020627) will be added on by ARUP, and additional charges will apply. If clients are unable to culture specimens, Cell Culture for Genetic Testing should be added to initial order.  
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. 5161.

**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.

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

**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.

**Stability:** 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: 2 weeks; Frozen: Unacceptable

New York State Clients: Whole blood: Ambient: 48 hours

Methodology: Massively Parallel Sequencing

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 Codes: 81364, 81404; 81479; for skin punch biopsy, add 88233

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](#) [By](#) report

**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.**