

TEST CHANGE

Hereditary Bone Marrow Failure Panel, Sequencing and Deletion/Duplication

3001615, BMF 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 Ringer's solution. New York State Clients: Lavender (EDTA) or yellow (ACD solution A or B) AND Prevention Genetics Healthcare Provider Statement

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 whole blood. (Min: 2 mL)
New York State Clients: Transport 5 mL whole blood (Min: 3 mL)

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: 1 week; Frozen:

Unacceptable

New York State Clients: Only whole blood: Ambient: 48 hours;
Refrigerated: 1 week; Frozen: Unacceptable

Methodology:

Massively Parallel Sequencing

Note:

Genes Tested: *ACD; ALAS2; ANKRD26; ATM; BLM; BRCA1**
(*NM_007294*); *BRCA2* (*NM_000059*); *BRIP1; CBL; CEBPA***;
CSF3R; CTC1; CXCR4; DDX41; DKC1; DNAJC21**; *ELANE; ERCC4*;
*ERCC6L2**; *ETV6; FANCA**; *FANCB; FANCC; FANCD2**; *FANCE*;
*FANCF; FANCG; FANCI; FANCL**; *G6PC3; GATA1; GATA2; GF11*;
HAX1; HOXA11; IKZF1; KRAS; MBD4; MPL; MYH9; NBN; NHP2;
*NOP10***; *NRAS; PALB2; PARN; PTPN11; RAD51C; RMRP***;
*RPL11; RPL15***; *RPL26; RPL35A; RPL5; RPS10; RPS19; RPS24*;
RPS26; RPS7; RTEL1; RUNX1; SAMD9; SAMD9L; SLX4; SRP72;
*TERC****; *TERT; TET2; TIN2; TP53; UBE2T; USB1; VPS45; WAS*;
WRAP53

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

**Deletion/duplication detection is not available for this gene.

***Duplication detection is not available for this gene.

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

CPT Codes:

81162; 81218; 81307; 81351; 81405; 81406; 81408; 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.~~ ~~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.~~

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.