

Test Definition: HUNTS

Hunter Syndrome, Full Gene Analysis

Reporting Title: Hunter Syndrome, Full Gene Analysis

Performing Location: Rochester

Specimen Requirements:

Forms:

1. Molecular Genetics-Biochemical Disorders Patient Information Sheet (Supply T527) in Special Instructions

2. New York Clients-Informed consent is required. Please document on the request form or electronic order that a copy is on file. An Informed Consent for Genetic Testing (Supply T576) is available in Special Instructions.

Specimen must arrive within 96 hours of collection.

Submit only 1 of the following specimens:

Preferred:

Specimen Type: Whole blood

Container/Tube:

Preferred: Lavender top (EDTA) or yellow top (ACD)

Acceptable: Any anticoagulant Specimen Volume: 3 mL Collection Instructions:

Invert several times to mix blood.
 Send specimen in original tube.

Specimen Stability Information: Ambient (preferred)/Refrigerated

Specimen Type: Cultured fibroblasts Container/Tube: T-75 or T-25 flask

Specimen Volume: 1 full T-75 flask or 2 full T-25 flasks

Specimen Stability Information: Ambient (preferred)/Refrigerated <24 hours

Specimen Type: Skin biopsy

Container/Tube: Sterile container with any standard cell culture media (eg, minimal essential media, RPMI 1640). The solution should be supplemented with 1% penicillin and streptomycin. Tubes can be supplied upon request (Eagle's

minimum essential medium with 1% penicillin and streptomycin [Supply T115]).

Specimen Volume: 4-mm punch

Specimen Stability Information: Refrigerated (preferred)/Ambient

Acceptable:

Specimen Type: Blood spot

Container/Tube:

Preferred: Collection card (Whatman Protein Saver 903 Paper)

Acceptable: Ahlstrom 226 filter paper, or Supplemental Newborn Screening Card (Supply T493)

Specimen Volume: 2 to 5 Blood Spots on collection card (Whatman Protein Saver 903 Paper; Ahlstrom 226 filter paper;

or Supplemental Newborn Screening Card, Supply T493)

Collection Instructions:

- 1. An alternative blood collection option for a patient >1 year of age is finger stick.
- 2. Let blood dry on the filter paper at ambient temperature in a horizontal position for 3 hours.
- 3. Do not expose specimen to heat or direct sunlight.
- 4. Do not stack wet specimens.
- 5. Keep specimen dry



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Specimen Stability Information: Ambient (preferred)/Refrigerated

Specimen Type	Temperature	Time
Varies	Varies	

Ask at Order Entry (AOE) Questions:

Test ID	Question ID	Description	Туре	Reportable
FBC	CG030	Source: • Autopsy • Chorionic Villi • Products of Conception or Stillbirth	Answer List	No
FBC	CG033	Reason for Referral	Plain Text	No

Result Codes:

Result ID	Reporting Name	Туре	Unit	LOINC®
33910	Reason for Referral	Alphanumeric		In Process
33911	Result	Alphanumeric		In Process
33912	Interpretation	Alphanumeric		In Process
33913	Method	Alphanumeric		In Process
33914	Amendment	Alphanumeric		In Process
33915	Specimen	Alphanumeric		In Process
33916	Source	Alphanumeric		In Process
33917	Reviewed By	Alphanumeric		In Process
33918	Release Date	Alphanumeric		N/A

CPT Code: 1 × 81405

Reflex Tests:

Te	est ID	Reporting Name	CPT Units	CPT Code	Always Performed	Orderable Separately
FE	3C	Fibroblast Culture for Genetic Test		Profile	No	Yes



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Result Codes for Reflex Tests:

Test ID	Result ID	Reporting Name	Туре	Unit	LOINC®
FBC	16167	Specimen	Alphanumeric		31208-2
FBC	16337	Specimen ID	Alphanumeric		N/A
FBC	CG030	Source	Alphanumeric		N/A
FBC	16168	Order Date	Alphanumeric		N/A
FBC	CG033	Reason For Referral	Alphanumeric		42349-1
FBC	16172	Method	Alphanumeric		In Process
FBC	16175	Interpretation	Alphanumeric		69965-2
FBC	16176	Amendment	Alphanumeric		In Process
FBC	16177	Consultant	Alphanumeric		N/A
FBC	16178	Report Date	Alphanumeric		N/A

Reference Values:

An interpretive report will be provided.