

Test Definition: FFRBS

Friedreich Ataxia, Frataxin, Quantitative, Blood Spot

Reporting Title: Frataxin, Quant, BS **Performing Location:** Rochester

Necessary Information:

Provide a reason for testing with each specimen.

Specimen Requirements:

Supplies: Card-Blood Spot Collection (Filter Paper) (T493)

Container/Tube:

Preferred: Blood spot collection card

Acceptable: PerkinElmer 226 (formerly Ahlstrom 226) Filter Paper and Whatman Protein Saver 903 Paper

Specimen Volume: 2 blood spots

Collection Instructions:

1. An alternative blood collection option for a patient older than 1 year is a fingerstick. For detailed instructions, see How

- to Collect Dried Blood Spot Samples.
- 2. Let blood dry on the filter paper at ambient temperature in a horizontal position for a minimum of 3 hours.
- 3. Do not expose specimen to heat or direct sunlight.
- 4. Do not stack wet specimens.
- 5. Keep specimen dry.

Additional Information:

- 1. For collection instructions, see Blood Spot Collection Instructions
- 2. For collection instructions in Spanish, see Blood Spot Collection Card-Spanish Instructions (T777)
- 3. For collection instructions in Chinese, see Blood Spot Collection Card-Chinese Instructions (T800)

Specimen Minimum Volume:

1 Blood spot

Forms:

1. New York Clients-Informed consent is required. Document on the request form or electronic order that a copy is on file. The following documents are available:

-Informed Consent for Genetic Testing (T576)

-Informed Consent for Genetic Testing-Spanish (T826)

2. Biochemical Genetics Patient Information (T602)

3. If not ordering electronically, complete, print, and send a Biochemical Genetics Test Request (T798) with the specimen.

Specimen Type	Temperature	Time	Special Container	
Whole blood	Ambient (preferred)	30 days	FILTER PAPER	
	Frozen	30 days	FILTER PAPER	



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Refrigerated	30 days	FILTER PAPER
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Result Codes:

Result ID	Reporting Name	Туре	Unit	LOINC®
32249	Reason for Referral	Alphanumeric		42349-1
32250	Method	Alphanumeric		85069-3
32251	Frataxin	Numeric	ng/mL	80980-6
32252	Interpretation	Alphanumeric		59462-2

LOINC and CPT codes are provided by the performing laboratory.

Supplemental Report:

No

CPT Code Information:

83520

Reference Values:

Pediatric (<18 years) normal frataxin: > or =15 ng/mL Adults (> or =18 years) normal frataxin: > or =21 ng/mL