

Disease

Friedreich ataxia^{1, 2}

Gene

FXN

Inheritance	AR
Current Treatments	Omaveloxolone
Evidence for Treatment	guideline ³
Non-Molecular Testing Availability	yes
Non-Molecular Test	mass spectrometry analysis of whole blood frataxin proteoforms ⁴
Disease Frequency (per 100,000)	3 ¹
Genes Included in Disease Frequency	FXN
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Friedreich+Ataxia+1&term=&cntry=&state=&city=&dist=

References

¹ <https://www.ncbi.nlm.nih.gov/books/NBK1281/>

² <https://pubmed.ncbi.nlm.nih.gov/36444905/>

³ <https://pubmed.ncbi.nlm.nih.gov/36371255/>

⁴ <https://pubmed.ncbi.nlm.nih.gov/35573317/>