

Disease

Barth Syndrome^{1, 2}

Gene

TFAZZIN

Inheritance	XL
Current Treatments	Cardiac transplantation, granulocyte colony-stimulating factor (G-CSF), uncooked cornstarch given prior to bedtime, elamipretide
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	monolysocardiolipin/cardiolipin (MLCL:L4-CL) ratio testing, urine organic acids ¹
Disease Frequency (per 100,000)	0.43 ¹
Genes Included in Disease Frequency	TFAZZIN
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Barth+Syndrome&term=&cntry=&state=&city=&dist=

References

¹ <https://www.ncbi.nlm.nih.gov/books/NBK247162/>

² <https://pubmed.ncbi.nlm.nih.gov/33077895/>