

Disease

Long QT syndrome 1^{1, 2, 3}

Gene

KCNQ1

Inheritance	AD
Current Treatments	nadolol, propranolol, mexiletine, ranolazine, flecainide, left cardiac sympathetic denervation, implantable cardiac defibrillator (ICD)
Evidence for Treatment	guideline ^{4, 5}
Non-Molecular Testing Availability	yes
Non-Molecular Test	EKG ⁶
ClinGen	https://actionability.clinicalgenome.org/ac/Pediatric/ui/stg2SummaryRpt?doc=AC131
Disease Frequency (per 100,000)	40 ⁶
Genes Included in Disease Frequency	KCNQ1, KCNH2, SCN5A
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Long+QT+Syndrome&term=&cntry=&state=&city=&dist=

References

- ¹ <https://omim.org/entry/192500>
- ² <https://pubmed.ncbi.nlm.nih.gov/34646680/>
- ³ <https://pubmed.ncbi.nlm.nih.gov/34039680/>
- ⁴ <https://pubmed.ncbi.nlm.nih.gov/24011539/>
- ⁵ <https://pubmed.ncbi.nlm.nih.gov/35936045/>
- ⁶ <https://www.ncbi.nlm.nih.gov/books/NBK1129/>