

Disease

Familial hypercholesterolemia-1^{1, 2}

Gene

LDLR

Inheritance	AD
Current Treatments	diet, statin, evinacumab, ezetimibe, apheresis, evolocumab, lomitapide
Evidence for Treatment	guideline ^{3, 4}
Non-Molecular Testing Availability	yes
Non-Molecular Test	plasma low-density lipoprotein (LDL) cholesterol ¹
ClinGen	https://actionability.clinicalgenome.org/ac/Pediatric/ui/stg2SummaryRpt?doc=AC057
Disease Frequency (per 100,000)	416.5 ^{5, 6}
Genes Included in Disease Frequency	APOB, LDLR, PCSK9
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Familial+Hypercholesterolemia&term=&cntry=&state=&city=&di

References

- ¹ <https://www.ncbi.nlm.nih.gov/books/NBK174884/>
- ² <https://www.ncbi.nlm.nih.gov/pubmed/31618540>
- ³ <https://pubmed.ncbi.nlm.nih.gov/29877295/>
- ⁴ <https://pubmed.ncbi.nlm.nih.gov/31838973/>
- ⁵ <https://www.ncbi.nlm.nih.gov/pubmed/30270061>
- ⁶ <https://ghr.nlm.nih.gov/condition/hypercholesterolemia#statistics>