Rx-Genes
Treatment Guidance at the Point of Care

Disease

Familial hypercholesterolemia-2[1]

Gene

APOB

Inheritance	AD
Current Treatments	diet, statin, evinacumab, ezetimibe, apheresis, evolocumab, lomitapide
Evidence for Treatment	guideline[2, 3]
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[4, 5]
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://pubmed.ncbi.nlm.nih.gov/29877295/

[3] https://pubmed.ncbi.nlm.nih.gov/31838973/

[4] https://www.ncbi.nlm.nih.gov/pubmed/30270061

[5] https://ghr.nlm.nih.gov/condition/hypercholesterolemia#statistics