

Disease

Recessive familial hypercholesterolemia-1^{1, 2}

Gene

LDLR

Inheritance	AR
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
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Familial+Hypercholesterolemia&term=&cntry=&state=&city=&d

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/>