

Disease

Homocystinuria¹

Gene

CBS

Inheritance	AR
Current Treatments	vitamin B6 (pyridoxine), methionine-restricted diet, folate, vitamin B12, betaine
Evidence for Treatment	guideline ²
Non-Molecular Testing Availability	yes
Non-Molecular Test	plasma total homocysteine and plasma amino acids ¹
ClinGen	https://actionability.clinicalgenome.org/ac/Adult/ui/stg2SummaryRpt?doc=AC097
Disease Frequency (per 100,000)	0.82 ³
Genes Included in Disease Frequency	CBS
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Homocystinuria&term=&cntry=&state=&city=&dist=

References

- ¹ <https://www.ncbi.nlm.nih.gov/books/NBK1524/>
- ² <https://pubmed.ncbi.nlm.nih.gov/27778219/>
- ³ <https://www.ncbi.nlm.nih.gov/pubmed/32232970>