

Disease

Acute intermittent porphyria^{1, 2}

Gene

HMBS

Inheritance	AD
Current Treatments	Givosiran, hemin
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	erythrocyte porphobilinogen deaminase activity ³
ClinGen	https://actionability.clinicalgenome.org/ac/Adult/ui/stg2SummaryRpt?doc=AC095
Disease Frequency (per 100,000)	1.4 ^{4, 5}
Genes Included in Disease Frequency	HMBS
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Acute+Intermittent+Porphyria&term=&cntry=&state=&city=&dis

References

- ¹ <https://www.ncbi.nlm.nih.gov/books/NBK1193/>
- ² <https://www.ncbi.nlm.nih.gov/pubmed/30726693>
- ³ <https://pubmed.ncbi.nlm.nih.gov/29317194/>
- ⁴ <https://onlinelibrary.wiley.com/doi/full/10.1111/bjh.14459>
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