

Disease

D-2-hydroxyglutaric aciduria type II^{1, 2}

Gene

IDH2

Inheritance	AD
Current Treatments	Enasidenib, riboflavin
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	urine organic acid ²
Experimental / Investigational Therapies	https://clinicaltrials.gov/search?cond=hydroxyglutaric%20acidemia&viewType=Table&checkSpell=

References

¹ <https://pubmed.ncbi.nlm.nih.gov/38736636/>

² <https://pubmed.ncbi.nlm.nih.gov/22391998/>