

Disease

Isovaleric acidemia¹

Gene

IVD

Inheritance	AR
Current Treatments	low protein diet, l-carnitine, glycine
Evidence for Treatment	guideline ^{2, 3}
Non-Molecular Testing Availability	yes
Non-Molecular Test	Urine organic acids ¹
Disease Frequency (per 100,000)	0.4 ⁴
Genes Included in Disease Frequency	IVD
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=ISOVALERIC&term=&cntry=&state=&city=&dist=

References

- ¹ <https://omim.org/entry/243500>
- ² https://www.e-imd.org/files/medias/files/news/meeting2018/11-%20Prasentation_brussel_Et_RE.pdf
- ³ http://www.bimdg.org.uk/store/guidelines/ER-IVA-v4_101727_09092016.pdf
- ⁴ <https://www.ncbi.nlm.nih.gov/pubmed/25022222>