

Disease

Aminolevulinic acid dehydratase deficiency porphyria^{1, 2}

Gene

ALAD

Inheritance	AR
Current Treatments	hemin
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	urine total porphyrins and delta-aminolevulinic acid level ²
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=alad&term=&cntry=&state=&city=&dist=

References

¹ <https://www.ncbi.nlm.nih.gov/pubmed/31085196>

² <https://pubmed.ncbi.nlm.nih.gov/32809671/>