

Disease

IARS1 associated growth retardation, impaired intellectual development, hypotonia, and hepatopathy^{1, 2}

Gene

IARS1

Inheritance	AR
Current Treatments	Isoleucine supplementation and protein fortification (2.5mg/kg/day, during illness 3.5 g/kg/day)
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	no
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=iars1&term=&cntry=&state=&city=&dist=

References

¹ <https://omim.org/entry/617093>

² <https://pubmed.ncbi.nlm.nih.gov/34194004/>