

Disease

Thiamine-responsive megaloblastic anemia syndrome with diabetes mellitus and sensorineural deafness^{1, 2}

Gene

SLC19A2

Inheritance	AR
Current Treatments	B1 (thiamine), insulin
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	glucose tolerance test, hemoglobin A1C, insulin level, glucose levels, complete blood count with MCV, B12 and folate levels, hearing test ³
Disease Frequency (per 100,000)	0.87 ¹
Genes Included in Disease Frequency	ABCC8, GCK, INS, KCNJ11, SLC19A2, PDX1, GATA4, GATA6, PTF1A, EIF2AK3, GLIS3, RFX6, NEUROD1, NEUROG3, PAX4, WFS1, MNX1, NKX2-2, IER3IP1
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=SLC19A2&term=&cntry=&state=&city=&dist=

References

¹ <https://www.ncbi.nlm.nih.gov/pubmed/29406006>

² <https://www.ncbi.nlm.nih.gov/pubmed/20301459>

³ <https://www.ncbi.nlm.nih.gov/books/NBK1282/>