

Disease

ABCC8 associated permanent neonatal diabetes mellitus^{1, 2}

Gene

ABCC8

Inheritance	AR and AD
Current Treatments	Insulin, glibenclamide, oral pancreatic enzymes
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	glucose tolerance test, hemoglobin A1C, insulin level, glucose level ²
Disease Frequency (per 100,000)	2.4 ³
Genes Included in Disease Frequency	ABCC8, GCK, GLUD1, HADH, HK1, HNF1A, HNF4A, PMM2, KCNJ11, SLC16A1, UCP2, FOXA2
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=abcc8&term=&cntry=&state=&city=&dist=

References

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

² <https://omim.org/entry/618857>

³ <https://www.ncbi.nlm.nih.gov/pubmed/?term=PMID%3A+24865345>