

Disease

KCNJ11 associated permanent neonatal diabetes mellitus

1, 2, 3

Gene

KCNJ11

Inheritance	AD
Current Treatments	Insulin, oral pancreatic enzymes; early sulphonylurea treatment (glibenclamide) in patients with specific KCNJ11 p.(V59M) variant may improve cognitive outcome
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 ⁴
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=kcnj11&term=&cntry=&state=&city=&dist=

References

- ¹ <https://www.ncbi.nlm.nih.gov/pubmed/29406006>
- ² <https://omim.org/entry/618856>
- ³ <https://pubmed.ncbi.nlm.nih.gov/29880308/>
- ⁴ <https://www.ncbi.nlm.nih.gov/pubmed/?term=PMID%3A+24865345>