Rx-Genes
Treatment Guidance at the Point of Care

Disease

Hyperinsulinism-hyperammonemia syndrome[1, 2, 3]

Gene

GLUD1

Inheritance	AD
Current Treatments	Diazoxide, somatostatin analogs, nifedipine, glucagon, IGF-1, glucocorticoids, growth hormone, pancreatic resection, mTOR inhibitors, GLP-1 receptor antagonists, sirolimus
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	ammonia, glucose, insulin, free fatty acid levels[1]
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=Hyperinsulinism&term=&cntry=&state=&city=&dist=

References

[1] https://www.ncbi.nlm.nih.gov/books/NBK1375/#hi.Summary

[2] https://pubmed.ncbi.nlm.nih.gov/27383869/

[3] https://pubmed.ncbi.nlm.nih.gov/35752848/

[4] https://www.ncbi.nlm.nih.gov/pubmed/?term=PMID%3A+24865345