

Disease

Vissers-Bodmer syndrome Arg535Cys variant¹

Gene

CNOT1

Inheritance	AD
Current Treatments	Insulin, pancreatic enzyme
Evidence for Treatment	Expert opinion
Non-Molecular Testing Availability	Yes
Non-Molecular Test	Glucose tolerance test, hemoglobin A1C, insulin level, glucose level, fecal chymotrypsin, elastase levels ¹
Experimental / Investigational Therapies	https://clinicaltrials.gov/search?cond=Neonatal%20Diabetes&viewType=Table

References

¹ <https://pubmed.ncbi.nlm.nih.gov/41911374/>