

Disease

Pancreatic agenesis 2^{1, 2}

Gene

PTF1A

Inheritance	AR
Current Treatments	Insulin, oral pancreatic enzymes
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	fecal elastase and pancreatic enzymes (pancreatic amylase and lipase) , imaging using abdominal ultrasonography, computed tomography, or magnetic resonance imaging 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=ptf1a&term=&cntry=&state=&city=&dist=

References

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

² <https://omim.org/entry/615935>

³ <https://pubmed.ncbi.nlm.nih.gov/32893856/>