Table 26-1

Causes of Pancreatic Insufficiency in Children

Disease	*OMIM*	*Gene/Locus*	*Comments*
CF	219700	CFTR	Most common
SDS	260400	SBDS	Hematologic abnormalities and malignancies
Johanson-Blizzard syndrome (JBS)	243800	UBR1	Nasal alar hypoplasia, congenital deafness
Pearson marrow-pancreas syndrome (PMPS)	557000	mtDNA	Refractory anemia in infancy
Pancreatic agenesis	260370	IPF1	Both endocrine and exocrine PI
Congenital lipase deficiency	614338	PNLIP	Steatorrhea, but usually without FTT
Congenital enterokinase deficiency	226200	PRSS7	Protein malabsorption; no steatorrhea
Syndrome of PI, dyserythropoietic anemia, calvarial hyperostosis	612714	COX4I2	Steatorrhea, FTT, anemia
Pancreatic and cerbellar agenesis	609069	PTF1A	Diabetes mellitus, cerebellar agenesis
Chronic pancreatitis (CP)			PI seen mainly in adults
After extensive pancreatectomy, or rarely, pancreatic obliteration after a severe, necrotic bout of acute pancreatitis			

OMIM: Online Mendelian Inheritance in Man. CF: cystic fibrosis; SDS: Shwachman-Diamond syndrome; JBS: Johanson-Blizzard syndrome; PMPS: Pearson marrow-pancreas syndrome; PI: pancreatic insufficiency; CP: chronic pancreatitis; FTT: failure to thrive.

load, their reduction leads to the symptoms of steatorrhea during PI. The lipase output, however, needs to drop to less than 10% below normal levels in order to develop fat malabsorption. These observations not only demonstrate the large reserve capacity of lipase secretion by the pancreas, but they also suggest that nonpancreatic sources of lipase, such as gastric lipase or in breastfed infants, carboxyester lipase from breast milk can contribute to lipolysis.

Cystic Fibrosis

In children, CF is the most common cause of PI and, for this reason, always needs to be considered in the differential diagnosis of PI. The autosomal recessive defect in the *cystic fibrosis*