EmblemHealth

Genetic Testing for Hereditary Pancreatitis

Last Review Date: October 12, 2018	Number: MG.MM.LA.28C3

Definitions

Acute pancreatitis	Sudden inflammation of the pancreas caused by trypsin activation within the pancreas that usually resolves after a few days with treatment.
Chronic pancreatitis	Ongoing inflammation of the pancreas caused by trypsin activation within the pancreas that progressively leads to permanent damage exocrine and endocrine pancreatic insufficiency and increased risk for pancreatic cancer.
Hereditary pancreatitis (subset of chronic pancreatitis)	An autosomal dominant disease, primarily caused by sequence variants in the protease, serine, 1 (trypsin 1) gene (PRSS1) and characterized by recurrent episodes of epigastric pain, nausea and vomiting. Symptoms of hereditary pancreatitis can start in childhood and evolve into chronic pancreatitis by 20 years of age, however disease onset is variable and some people may not exhibit symptoms until adulthood.

Related Medical Guidelines

Carrier Screening for Parents or Prospective Parents
Genetic Counseling and Testing

Guideline

Genetic testing for hereditary pancreatitis (PRSS1, SPIK1 and CFTR mutations) is considered medically necessary in children aged 18 years and under, with a confirmed diagnosis of acute or chronic pancreatitis, and for whom additional invasive diagnostic tests would be deemed unnecessary if genetic test result is positive.

Limitations/Exclusions

Genetic testing for hereditary pancreatitis is considered investigational for all for all other indications not meeting the above criteria.

Applicable Procedure Codes

81220	CFTR (cystic fibrosis transmembrane conductance regulator) (eg, cystic fibrosis) gene analysis; common variants (eg, ACMG/ACOG guidelines)
81221	CFTR (cystic fibrosis transmembrane conductance regulator) (eg, cystic fibrosis) gene analysis; known familial variants
81222	CFTR (cystic fibrosis transmembrane conductance regulator) (eg, cystic fibrosis) gene analysis; duplication/deletion variants
81223	CFTR (cystic fibrosis transmembrane conductance regulator) (eg, cystic fibrosis) gene analysis; full gene sequence
81224	CFTR (cystic fibrosis transmembrane conductance regulator) (eg, cystic fibrosis) gene analysis; intron 8 poly-T analysis (e.g., male infertility)
81401	Molecular pathology procedure, Level 2 (eg, 2-10 SNPs, 1 methylated variant, or 1 somatic variant [typically using nonsequencing target variant analysis], or detection of a dynamic mutation disorder/triplet repeat) LINC00518 (long intergenic non-protein coding RNA 518) (eg, melanoma), expression analysis PRAME (preferentially expressed antigen in melanoma) (eg, melanoma), expression analysis (Revision eff. 01/01/2018)
81404	Molecular pathology procedure, Level 5 (eg, analysis of 2-5 exons by DNA sequence analysis, mutation scanning or duplication/deletion variants of 6-10 exons, or characterization of a dynamic mutation disorder/triplet repeat by Southern blot analysis) (Revision eff. 01/01/2018)
81479	Unlisted molecular pathology procedure

Applicable ICD-10 Diagnosis Codes

K85.00	Idiopathic acute pancreatitis without necrosis or infection
K85.01	Idiopathic acute pancreatitis with uninfected necrosis
K85.02	Idiopathic acute pancreatitis with infected necrosis
K85.80	Other acute pancreatitis without necrosis or infection
K85.81	Other acute pancreatitis with uninfected necrosis
K85.82	Other acute pancreatitis with infected necrosis
K85.90	Acute pancreatitis without necrosis or infection, unspecified
K85.91	Acute pancreatitis with uninfected necrosis, unspecified
K85.92	Acute pancreatitis with infected necrosis, unspecified
K86.1	Other chronic pancreatitis

References

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