Medizinische Genetik Mainz

Genetic analyses for hereditary gastroenterological diseases

Gastrointestinal disorders occur with a high prevalence in the population and encompass a very broad spectrum of conditions. In recent years, it has become apparent that many of these disorders have underlying genetic causes. Genetic diagnosis of these diseases can enable a better progression assessment and a more targeted treatment approach. In addition, genetic testing allows to identify asymptomatic family members at risk, providing a starting point for appropriate screening and early diagnosis.

A	Alagille syndrome (ALGS)
	Alpha-1 antitrypsin deficiency *
	Arthrogryposis renal dysfunction (ARC Syndrome)
	Autosomal dominant polycystic kidney disease (ADPKD)
	Autosomal dominant polycystic liver disease (ADPLD)
	Autosomal recessive polycystic kidney disease (ARPKD)
B	Benign recurrent intrahepatic cholestasis (BRIC)
	Bile acid synthesis defects
C	Congenital hepatic fibrosis (CHF)
	Crigler-Najjar syndrome (CNS) *
	Cystic fibrosis *
F	Familial intrahepatic progressive cholestasis (PFIC)
	Fanconi-Bickel syndrome
G	Glycogen storage diseases (GSD)
	Gilbert´s syndrome
H	Hemochromatosis *
	Hereditary pancreatitis
	Heterotaxia
J	Joubert syndrome
L	Liver organ morphogenesis disorders
	Lynch syndrome
M	Meckel-Gruber syndrome
	Meulengracht disease *
	Mitochondrial Hepatopathies
	Multiple endocrine neoplasia (MEN1) *
P	Polycystic liver disease (PCLD)
	Polyposis
R	Recurrent acute liver failure

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T	Transport disorders in hepatocytes and cholangiocytes
W	Wilson disease

* Analysis provided by a group partner laboratory