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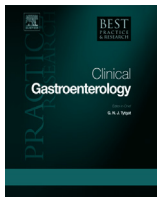

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Congenital intestinal diarrhoeal diseases: A diagnostic and therapeutic challenge

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Congenital diarrhoeal disorders
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Defects of enterocyte differentiation or polarisation
Defects of enteroendocrine cells
Defects of the intestinal immune system

A B S T R A C T

Congenital diarrhoeal disorders are a heterogeneous group of inherited malabsorptive or secretory diseases typically appearing in the first weeks of life, which may be triggered by the introduction of distinct nutrients. However, they may also be unrecognised for a while and triggered by exogenous factors later on. In principle, they can be clinically classified as osmotic, secretory or inflammatory diarrhoea. In recent years the disease-causing molecular defects of these congenital disorders have been identified. According to the underlying pathophysiology they can be classified into four main groups:
1) Defects of digestion, absorption and transport of nutrients or electrolytes
2) Defects of absorptive enterocyte differentiation or polarisation
3) Defects of the enteroendocrine cells
4) Defects of the immune system affecting the intestine
Here, we describe the clinical presentation of congenital intestinal diarrhoeal diseases, the diagnostic work-up and specific treatment aspects.

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Abbreviations

AE	acrodermatitis enteropathica
ALP	abetalipoproteinemia
APECED	autoimmune-polyendocrinopathy-candidiasis-ectodermal-dystrophy
CD	Crohn's disease
CCD	congenital chloride diarrhoea
CDD	congenital diarrhoeal disorders
CEP	congenital enteropeptidase-deficiency
CF	cystic fibrosis
CGD	chronic granulomatous disease
CGGM	congenital glucose-galactose malabsorption
CLD	congenital lactase deficiency
CLPS	cofactor protein colipase
CMD	congenital malabsorptive diarrhoea
CRD	chylomicron retention disease
CSD	congenital sodium diarrhoea
CSID	congenital sucrase-isomaltase deficiency
CVID	common variable immunodeficiency
DNA	deoxyribonucleic acid
FMF	familial Mediterranean fever
FOXP3	forkhead box P3
G6PC3	glukose-6-phosphatase catalytic subunit 3
GI	gastrointestinal
GSD1b	glycogen storage disease type Ib
HED	hypohydrotic ectodermal dysplasia
HLP	familial hypobetalipoproteinemia
HP	hereditary pancreatitis
HPS	Hermansky–Pudlak syndrome
HSCT	haematopoietic stem cell transplantation
IBD	inflammatory bowel disease
IED	intestinal epithelial dysplasia
Ig	immunoglobulin
IGAD	IgA deficiency
IL	interleukin
ILE	intestinal lymphangiectasia
IPEX	immunodysregulation, polyendocrinopathy and enteropathy X-linked
LAD1	Leucocyte adhesion deficiency 1
LPI	lysinuric protein intolerance
NLH	nodular lymphoid hyperplasia
MGAD	congenital maltase-glucoamylase deficiency
MVID	microvillus inclusion disease
MKD	mevalonate kinase deficiency
PBAM	primary bile acid malabsorption
PCD	prohormone convertase 1/3 deficiency
PNLIP	pancreas lipase
SCID	severe combined immunodeficiency
SDS	Shwachman–Diamond syndrome
SeHCAT	selenium-homocholic acid taurine
STAT	signal transducer and activator of transcription
THES	trichohepatoenteric syndrome
TNF	tumour necrosis factor
TPN	total parental nutrition