APPENDIX. Conditions inherited in autosomal recessive or X-linked manner

Conditions	Genes
ABCC8-related hyperinsulinism	ABCC8
Achromatopsia	CNGB3
Alkaptonuria	HGD
Alpha-1 antitrypsin deficiency	SERPINA1
Alpha-mannosidosis	MAN2B1
Andermann syndrome	SLC12A6
Autosomal recessive spastic ataxia of Charlevoix-Saguenay	SACS
Aspartylglycosaminuria	AGA
Ataxia with vitamin E deficiency	TTPA
Ataxia-telangiectasia	ATM
Autosomal recessive polycystic kidney disease	PKHD1
Bardet–Biedl syndrome, BBS1-related	BBS1
Bardet–Biedl syndrome, BBS10-related	BBS10
Biotinidase deficiency	BTD
Bloom dyndrome	BLM
Canavan disease	ASPA
Carnitine palmitoyltransferase IA deficiency	CPT1A
Carnitine palmitoyltransferase II deficiency	CPT2
Cartilage-hair hypoplasia	RMRP
Citrullinemia type 1	ASS1
CLN3-related neuronal ceroid lipofuscinosis	CLN3
CLN5-related neuronal ceroid lipofuscinosis	CLN5
Cohen syndrome	VPS13B
Congenital disorder of glycosylation type Ia	PMM2
Congenital disorder of glycosylation type Ib	MPI
Congenital Finnish nephrosis	NPHS1
Costeff optic atrophy syndrome	OPA3
Cystic fibrosis	CFTR
Cystinosis	CTNS
D-bifunctional protein deficiency	HSD17B4
Factor XI deficiency	F11
Familial dysautonomia	IKBKAP
Familial Mediterranean fever	MEFV
Fanconi anaemia type C	FANCC
Fragile X syndrome	FMR1
Galactosaemia	GALT
GJB2-related DFNB1 nonsyndromic hearing loss and deafness	GJB2
Glutaric acidemia type 1	GCDH
Glycogen storage disease type Ia	G6PC
Glycogen storage disease type Ib	SLC37A4
Glycogen storage disease type III	AGL
Glycogen storage disease type V	PYGM
GRACILE syndrome	BCS1L
Hb beta chain-related haemoglobinopathy (including beta-thalassaemia and sickle cell disease)	HBB
Hereditary fructose intolerance	ALDOB
Hereditary thymine-uraciluria	DPYD
Herlitz junctional epidermolysis bullosa, LAMA3-related	LAMA3
Herlitz junctional epidermolysis bullosa, LAMB3-related	LAMB3
Herlitz junctional epidermolysis bullosa, LAMC2-related	LAMC2
Hexosaminidase A deficiency (including Tay–Sachs disease)	HEXA
Homocystinuria caused by cystathionine beta-synthase deficiency	CBS
Hypophosphatasia, autosomal recessive	ALPL
Inclusion body myopathy 2	GNE

APPENDIX. (cont'd)

Conditions	Genes
Isovaleric acidaemia	IVD
Joubert syndrome 2	TMEM216
Krabbe's disease	GALC
Limb-girdle muscular dystrophy type 2D	SGCA
Limb-girdle muscular dystrophy type 2E	SGCB
Lipoamide dehydrogenase deficiency	DLD
Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency	HADHA
Maple syrup urine disease type 1B	BCKDHB
Medium-chain acyl-CoA dehydrogenase deficiency	ACADM
Megalencephalic leukoencephalopathy with subcortical cysts	MLC1
Metachromatic leukodystrophy	ARSA
Mucolipidosis IV	MCOLN1
Muscle–eye–brain disease	POMGNT1
NEB-related nemaline myopathy	NEB
Niemann–Pick disease type C	NPC1
Niemann–Pick disease, SMPD1-associated	SMPD1
Nijmegen breakage syndrome	NBN
Northern epilepsy	
Pendred's syndrome	SLC26A4
PEX1-related Zellweger syndrome spectrum	PEX1
Phenylalanine hydroxylase deficiency	PAH
Polyglandular autoimmune syndrome type 1	AIRE
Pompe's disease	GAA
PPT1-related neuronal ceroid lipofuscinosis	PPT1
Primary carnitine deficiency	SLC22A5
Primary hyperoxaluria type 1	AGXT
Primary hyperoxaluria type 2	GRHPR
PROP1-related combined pituitary hormone deficiency	PROP1
Pseudocholinesterase deficiency	BCHE
Pycnodysostosis	CTSK
Rhizomelic chondrodysplasia punctata type 1	PEX7
Salla disease	SLC17A5
Segawa dyndrome	TH
Short-chain acyl-CoA dehydrogenase deficiency	ACADS
Sjogren–Larsson syndrome	ALDH3A2
Smith–Lemli–Opitz syndrome	DHCR7
Steroid-resistant nephrotic syndrome	NPHS2
Sulfate transporter-related osteochondrodysplasia	SLC26A2
TPP1-related neuronal ceroid lipofuscinosis	TPP1
Tyrosinemia type I	FAH
Usher's syndrome type 1F	PCDH15
Usher's syndrome type 3	CLRN1
Very long-chain acyl-CoA dehydrogenase deficiency	ACADVL
Walker–Warburg syndrome	FKTN
Wilson's disease	ATP7B
21-Hydroxylase deficient congenital adrenal hyperplasia	CYP21A2
Alpha-thalassaemia	HBA1, HBA2
Gaucher's disease	GBA
Hurler syndrome	IDUA
Spinal muscular atrophy	SMN1
Choroideremia	CHM
X-linked juvenile retinoschisis	RS1

