ORIGIN hordozóság szűrés

betegségek listája

ORIGIN

HORDOZÓSÁG SZŰRÉS

Az ORIGIN hordozóság szűrés
által vizsgált örökíthető betegségek listája

1.	ACADM	Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency
2.	AGXT	Primary hyperoxaluria type 1
3.	ARSA	Metachromatic leukodystrophy
4.	ATP7B	Wilson disease
5.	BTD	Biotinidase deficiency
6.	CBS	Homocystinuria due to cystathionine beta-synthase deficiency
7.	CFTR	Cystic fibrosis
8.	DHCR7	Smith-Lemli-Opitz syndrome
9.	EMD	Emery-Dreifuss muscular dystrophy
10.	FMR1	Fragile X syndrome
11.	GAA	Pompe disease
12.	GALC	Krabbe disease
13.	GALT	Galactosemia
14.	GBA	Gaucher disease
15.	GJB1	Charcot-Marie-Tooth disease, X-linked type 1
16.	GJB2	Nonsyndromic hearing loss, GJB2-related
17.	GJB6	Nonsyndromic hearing loss, GJB6-related
18.	GLA	Fabry disease
19.	HADHA	Long-chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency
20.	HBA1	Alpha thalassemia
21.	HBA2	Alpha thalassemia
22.	HBB	Sickle cell disease; Beta thalassemia
23.	HEXA	Tay-Sachs disease
24.	MEFV	Familial Mediterranean fever
25.	MMACHC	Methylmalonic aciduria and homocystinuria, cblC type
26.	PAH	Phenylalanine hydroxylase deficiency (Phenylketonuria)
27.	PMM2	Congenital disorder of glycosylation type 1a
28.	SERPINA	Alpha-1 antitrypsin deficiency
29.	SLC26A2	Achondrogenesis, type IB; Atelosteogenesis II; Diastrophic dysplasia
30.	SMN1	Spinal muscular atrophy