Reproductive Technology Council

PGT Approved Conditions List - January 2024

Condition number	Approved conditions	OMIM number
1	Achondroplasia (ACH)	100800
2	Alagille syndrome	118450
3	Amyotrophic lateral sclerosis 1 (ALS1)	105400
4	Angelman syndrome (UBE3A gene only)	105830
5	Antithrombin III deficiency (AT3D)	613118
6	Autosomal dominant polycystic kidney disease 1 (APKD1)	173900
7	Beta thalassaemia	141900
8	BRCA 1 gene mutation	113705
	BRCA 2 gene mutation	600185
9	Brugada syndrome (SUNDS)	601144
10	Carpenter's syndrome 1	201000
11	Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL)	125310
12	Charcot Marie tooth disease type 1A (CMT1A)	302800
	Charcot Marie tooth disease type 1A (CMT1A)	118220
13	Congenital disorder of glycosylation type 1a (CDG1A)	212065
14	Crigler Najjar syndrome type I (CN1)	218800
15	Cystic fibrosis (CF) classical	219700
16	D-bifunctional protein deficiency	261515
17	Duchenne muscular dystrophy	310200
18	Epidermolysis bullosa simplex	619555
19	Facioscapulohumeral muscular dystrophy type 2 (FSHD2)	158901
20	Familial adenomatous polyposis 1 (FAP1)	175100
	Familial adenomatous polyposis 2 (FAP2) MUTYH-associated polyposis	608456
21	Familial Creutzfeldt-Jakob disease (fCJD)	123400
22	Familial hypertrophic cardiomyopathy (CMH)	607450
23	Fragile X syndrome (FRAX)	300624
24	Galactosaemia	230400
25	Huntington's disease (HD)	143100
26	Leber congenital amaurosis (LCA)	204100
27	Lethal multiple pterygium syndrome (LMPS)	253290
28	Long QT syndrome type 1	192500
	Long QT syndrome type 2	613600
	Long QT syndrome type 3	603830
	Long QT syndrome type 5	613695
	Long QT syndrome type 6	613693
29	Microcephalic osteodysplastic primordial dwarfism type I (MOPD I)	210710
30	Mucopolysaccharidosis type I (MPS I)	607014
31	Multiple Lentigines syndrome (LEOPARD syndrome)	151100
32	Myofibrillar myopathy	617047

33	Myotonic dystrophy	160900
	Myotonic dystrpohy type 2	602668
34	Myotubular myopathy	310400
	Myotubular myopathy	300219
35	Nemaline myopathy	609273
36	Noonan syndrome type 1	163950
37	Optic atrophy 1 (OPA1)	118450
38	Palmoplantar keratoderma, nonepidermolytic, focal or diffuse	615735
39	Pendred syndrome (PDS)	274600
40	Pfeiffer syndrome	101600
41	Polycystic kidney disease (PKD) 2	613095
	Polycystic kidney disease (PKD) 3	600666
	Polycystic kidney disease (PKD) 5	617610
	Polycystic kidney disease (PKD) 6	618061
42	Pontocerebellar hypoplasia type 2A	277470
	Pontocerebellar hypoplasia type 4	225753
	Pontocerebellar hypoplasia type 5	610204
43	Pseudoachondroplasia	177170
44	Renpenning syndrome	309500
45	Retinitis pigmentosa	300455
46	Rhizomelic chondrodysplasia punctata (RCDP1) type 1	215100
47	Smith Lemli Opitz syndrome (SLOS)	270400
48	Spinal muscular atrophy and respiratory distress (SMARD1)	604320
49	Spondylometaphyseal dysplasia, corner fracture type (SMDCF)	184255
50	Tubulointerstitial kidney disease (ADTKD-UMOD) also known as MCKD2	603860
51	X-linked Alport syndrome	301050
52	X-linked lymphoproliferative disorder	308240
53	X-linked ocular albinism	300500
54	ALL translocations - PGT-SR	
	Chromosomal duplication	
	Chromosomal microdeletions	
	Chromosome insertions	
	Chromosome inversions	