Reproductive Technology Council

PGT approved conditions list V3

Condition number	RTC List of Approved Conditions	OMIM phenotype number
1	Achondroplasia (ACH)	#100800
2	Alagille Syndrome	#118450
3	Alport syndrome x-linked (ATS1)	#301050
4	Amyotrophic lateral sclerosis 1 (ALS1)	#105400
	Amyotrophic lateral sclerosis frontotemporal dementia (FTDALS1)	#105550
5	Angelman syndrome (UBE3A gene only)	#105830
6	Antithrombin III deficiency (AT3D)	#613118
7	Autosomal dominant polycystic kidney disease 1 (APKD1)	#173900
8	Beta thalassaemia	#613985
9	Breast cancer	#114480
	Breast-ovarian cancer, familial, susceptibility to,1 (BROVCA1)	#604370
	Breast-ovarian cancer, familial, susceptibility to,2 (BROVCA2)	#612555
	Breast-ovarian cancer, familial, susceptibility to,5 (BROVCA5)	#620442
10	Brugada syndrome (SUNDS)	#601144
11	Cardiomyopathy, dilated 1l	#604765
	Cardiomyopathy, familial hypertrophic 1 (CMH1)	#192600
	Cardiomyopathy, familiar hypertrophic 3 (CMH 3)	#115196
	Cardiomyopathy, familial hypertrophic 7 (CMH7)	#613690
	Cardiomyopathy, familial hypertrophic 10 (CMH10)	#608758
	Cardiomyopathy, familial hypertrophic 4 (CMH4)	#115197
	Cardiomyopathy, familial hypertrophic 26 (CMH26)	#617047
12	Carpenter's syndrome 1	#201000
13	Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL)	#125310
14	Charcot Marie Tooth disease (x-linked)	#302800
	Charcot Marie Tooth disease type 1A (CMT1A)	#118220
15	Congenital disorder of glycosylation type 1a (CDG1A)	#212065
16	Crigler Najjar syndrome type I (CN1)	#218800
17	Cystic fibrosis (CF) classical	#219700
18	Deafness, autosomal recessive type 1A (DFNB1A)	#220290
19	D-Bifunctional protein deficiency	#261515
20	Duchenne muscular dystrophy	#310200
21	Epidermolysis bullosa simplex	#619555
22	Facioscapulohumeral muscular dystrophy type 2 (FSHD2)	#158901
23	Familial adenomatous polyposis 1 (FAP1)	#175100
	Familial adenomatous polyposis 2 (FAP2) MUTYH-associated polyposis	#608456
24	Familial Creutzfeldt-Jakob disease (fCJD)	#123400
25	Fanconi anaemia type N (FANCN)	#610832

	Fanconi anameia complementation group D1 (FANCD1)	#605724
	Fanconi anameia complementation group S (FANCS)	#617883
26	Fragile X syndrome (FRAX)	#300624
27	Galactosaemia	#230400
28	Huntington's disease (HD)	#143100
29	Incontinentia pigmenti (IP) x-linked	#308300
30	Leber congenital amaurosis (LCA)	#204100
31	Lethal multiple pterygium syndrome (LMPS)	#253290
32	Long QT syndrome Type 1	#192500
	Long QT syndrome Type 3	#603830
	Long QT syndrome Type 5	#613695
	Long QT syndrome Type 6	#613693
33	Lymphoproliferative disorder x-linked (XLP1)	#308240
34	Microcephalic osteodysplastic primordial dwarfism type I (MOPD I)	#210710
35	Mucopolysaccharidosis type I (MPS I) (Hurler syndrome)	#607014
36	Multiple Lentigines syndrome (LEOPARD syndrome)	#151100
37	Myotonic dystrophy 1 (DM1)	#160900
	Myotonic dystrophy type 2 (DM2)	#602668
38	Myotubular myopathy	#300219
	Myotubular myopathy (CNMX)	#310400
39	Nemaline myopathy (NEM6)	#609273
40	Noonan syndrome 1 (NS1)	#163950
41	Ocular albinism, type 1 (OA1)	#300500
42	Optic atrophy 1 (OPA1)	#165500
43	Ornithine transcarbamylase deficiency	#311250
44	Palmoplantar keratoderma, nonepidermolytic, focal or diffuse (PPKNEFD)	#615735
45	Pancreatic cancer, susceptibility to,2	#613347
	Pancreatic cancer, susceptibility to,3	#613348
46	Pendred syndrome (PDS)	#274600
47	Pfeiffer syndrome	#101600
48	Polycystic kidney disease (PKD) 2	#613095
	Polycystic kidney disease (PKD) 3	#600666
	Polycystic kidney disease (PKD) 5	#617610
	Polycystic kidney disease (PKD) 6	#618061
49	Pontocerebellar hypoplasia Type 2A (PCH2A)	#277470
	Pontocerebellar hypoplasia Type 4 (PCH4)	#225753
	Pontocerebellar hypoplasia type 5 (PCH5)	#610204
50	Prostate cancer	#176807
51	Pseudoachondroplasia (PSACH)	#177170
52	Renpenning syndrome (RENS1)	#309500
53	Retinitis pigmentosa (RPSRDF)	#300455
54	Rhizomelic chondrodysplasia punctata type 1 (RCDP1)	#215100
55	Sickle cell disease	#603903
56	Smith Lemli Opitz syndrome (SLOS)	#270400
57	Spinal muscular atrophy, type 1 (SMA1)	#253300
	Spinal muscular atrophy, type 2 (SMA2)	#253550
	Spinal muscular atrophy, type 3 (SMA3)	#253400

	Spinal muscular atrophy and respiratory distress (SMARD1)	#604320
58	Spondylometaphyseal dysplasia, corner fracture type (SMDCF)	#184255
59	Tubulointerstitial kidney disease, autosomal dominant,1 (ADTKD1)	#162000

PGT-SR - cases that are at risk of transmitting a chromosomal abnormality that can cause repetitive pregnancy failure or developmental defects are included. * Cases that do not meet these criteria are required to submit an application to Council for approval.		
	ALL translocations	
	* Chromosomal duplication	
	* Chromosome insertions	
	* Chromosome inversions	
	* Chromosomal microdeletions	