Reproductive Technology Council

Preimplantation Genetic Testing Approved Conditions List

The Reproductive Technology Council (Council) has pre-approved the following list of genetic conditions (including structural rearrangements) for Preimplantation Genetic Testing (PGT) on embryos.

Assisted Reproductive Technology clinics no longer need to apply to Council for PGT approval if the condition (and the online mendelian inheritance in man (OMIM®) number) is on the approved conditions list.

The conditions on the list are identified by their phenotype number, which has a hash (#) before the number. (Genotypes are identified by an asterisk (*) before the number.)

Clinics are required to keep records of all diagnostic testing, including the OMIM phenotype number for annual reporting to the Department of Health's Reproductive Technology Unit.

Clinics should continue to provide assessment of suitability for PGT testing by a clinical geneticist and provide genetic counselling to patients regarding risks and relevant issues.

Condition number	RTC List of Approved Conditions	OMIM phenotype number
1	Achondroplasia (ACH)	#100800
2	Adrenoleukodystrophy (Adrenomyeloneuropathy) (ALD)	#300100
3	Alagille syndrome	#118450
4	Alport syndrome x-linked (ATS1)	#301050
5	Agammaglobulinaemia, x-linked (XLA)	#300755
6	Amyotrophic lateral sclerosis 1 (ALS1)	#105400
	Amyotrophic lateral sclerosis frontotemporal dementia (FTDALS1)	#105550
7	Angelman syndrome (UBE3A gene only)	#105830
8	Antithrombin III deficiency (AT3D)	#613118
9	Autosomal dominant polycystic kidney disease 1 (APKD1)	#173900
10	Bardet-Biedel Syndrome	#209900
11	Beta thalassaemia	#613985
12	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
13	Brugada syndrome (SUNDS)	#601144
14	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
15	Carpenter syndrome 1	#201000
16	Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL)	#125310
17	Cerebral cavernous malformation (CCM)	#116860
18	Charcot Marie Tooth disease (x-linked)	#302800

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	Charcot Marie Tooth disease demyelinating type 1A (CMT1A)	#118220
19	Congenital adrenal hyperplasia (21 hydroxylase deficiency)	#201910
20	Congenital disorder of glycosylation type 1a (CDG1A)	#212065
21	Crigler Najjar syndrome type I (CN1)	#218800
22	Cystic fibrosis (CF) classical	#219700
23	D-Bifunctional protein deficiency	#261515
24	Deafness, autosomal recessive type 1A (DFNB1A)	#220290
25	Developmental and epileptic encephalopathy 28 (DEE28)	#616211
26	Duchenne muscular dystrophy	#310200
27	Epidermolysis bullosa simplex	#619555
28	Fabry disease	#301500
29	Facioscapulohumeral muscular dystrophy type 2 (FSHD2)	#158901
30	Familial adenomatous polyposis 1 (FAP1)	#175100
31	Familial adenomatous polyposis 2 (FAP2) MUTYH-associated polyposis	#608456
32	Familial Creutzfeldt-Jakob disease (fCJD)	#123400
33	Fanconi anaemia type N (FANCN)	#610832
	Fanconi anaemia complementation group D1 (FANCD1)	#605724
	Fanconi anaemia complementation group S (FANCS)	#617883
34	Fragile X syndrome (FRAX)	#300624
35	Frontotemporal dementia and/or amyotrophic lateral sclerosis 1 (FTDALS1)	#105550
36	Galactosaemia	#230400
37	Haemophilia A	#306700
38	Holt-Oram syndrome	#142900
39	Hunter syndrome mucopolysaccharidosis type II	#309900
40	Huntington's disease (HD)	#143100
41	Hyperinsulinemic hypoglycenia, familial 1 (HHF1)	#256450
	Hyperinsulinemic hypoglycenia, familial 2 (HHF2)	#601820
42	Hyper IgE syndrome (HIES1)	#147060
43	Hypohidrotic ectodermal dysplasia, x-linked (XHED)	#305100
44	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3 (IHPRF3)	#616900
45	Ichthyosis (x-linked) (XLI)	#308100
46	Incontinentia pigmenti (IP) x-linked	#308300
47	Intellectual developmental disorder, x-linked, syndromic 34	#300967
48	Leber congenital amaurosis (LCA)	#204100
49	Lethal multiple pterygium syndrome (LMPS)	#253290
50	Li-Fraumeni syndrome	#151623
51	Long QT syndrome Type 1	#192500
	Long QT syndrome Type 3	#603830
	Long QT syndrome Type 5	#613695
	Long QT syndrome Type 6	#613693
52	Lynch Syndromes - MLH1	#276300
	Lynch Syndromes - MSH2	#619096
	Lynch Syndromes -MSH6	#619097
	Lynch Syndrome -PMS2	# 619101

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53	Lymphoproliferative disorder x-linked (XLP1)	#308240
54	Machado-Joseph disease	#109150
55	Marfan syndrome (MFS)	#154700
56	Microcephalic osteodysplastic primordial dwarfism type I (MOPD I)	#210710
57	Mucopolysaccharidosis type I (MPS I) (Hurler syndrome)	#607014
58	Muscular dystrophy (Becker) BMD	#300376
59	Multiple endocrine neoplasia type 1	#131100
60	Multiple lentigines syndrome (LEOPARD syndrome)	#151100
61	Myopathy, myofibrillar 5 (MFM5)	#609524
62	Myotonic dystrophy 1 (DM1)	#160900
	Myotonic dystrophy type 2 (DM2)	#602668
63	Myotubular myopathy	#300219
	Myotubular myopathy (CNMX)	#310400
64	Nemaline myopathy (NEM6)	#609273
65	Neurofibromatosis Type 1	#162200
	Neurofibromatosis Type 2	#101000
66	Noonan syndrome 1 (NS1)	#163950
67	Ocular albinism, type 1 (OA1)	#300500
68	Oculocutaneous albinism type 1A (OCA1A)	#203100
	Oculocutaneous albinism type 1B (OCA1B)	#606952
69	Optic atrophy 1 (OPA1)	#165500
70	Ornithine transcarbamylase deficiency	#311250
71	Osteogenesis imperfecta type 1	#166200
72	Palmoplantar keratoderma, epidermolytic 1 (EPPK1)	#144200
	Palmoplantar keratoderma, nonepidermolytic, focal or diffuse (PPKNEFD)	#615735
73	Pancreatic cancer, susceptibility to,2	#613347
	Pancreatic cancer, susceptibility to,3	#613348
74	Paragangliomas 4 (PGL4) SDHB gene defect	#115310
75	Pendred syndrome (PDS)	#274600
76	Periventricular nodular heterotopia 1 (PVNH1)	#300049
77	Pfeiffer syndrome	#101600
78	Phenylketonuria	#261600
79	Polycystic kidney disease (PKD) 2	#613095
	Polycystic kidney disease (PKD) 3	#600666
	Polycystic kidney disease (PKD) 5	#617610
	Polycystic kidney disease (PKD) 6	#618061
80	Pontocerebellar hypoplasia Type 2A (PCH2A)	#277470
	Pontocerebellar hypoplasia Type 4 (PCH4)	#225753
	Pontocerebellar hypoplasia type 5 (PCH5)	#610204
81	Prostate cancer	#176807
82	Pseudoachondroplasia (PSACH)	#177170
83	Pulmonary arterial hypertension	#178600
84	Renpenning syndrome (RENS1)	#309500
85	Retinitis pigmentosa (RPSRDF)	#300455
86	Rhizomelic chondrodysplasia punctata type 1 (RCDP1)	#215100

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87	Sickle cell disease	#603903
88	Smith Lemli Opitz syndrome (SLOS)	#270400
89	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
90	Spondylometaphyseal dysplasia, corner fracture type (SMDCF)	#184255
91	Tuberous sclerosis 2 (TSC2)	#613254
92	Tubulointerstitial kidney disease, autosomal dominant,1 (ADTKD1)	#162000
93	Von Hippel Lindau syndrome	#193300
94	Waardenburg syndrome type 2A (WS2A)	#193510
95	Wiskott-Aldrich syndrome (WAS)	#301000
96	X-linked Choroideremia	#303100
97	X-linked retinoschisis (RS1)	#312700

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 apply to Council for approval.

- ALL translocations
- Chromosomal duplication
- Chromosome insertions
- Chromosome inversions
- Chromosomal microdeletions