Monash IVF

Expanded Carrier Screen

Gene list (by gene)

Gene	Disorder	Inheritance	Population	Carrier frequency	Detection rate	Residual risk of being a carrier*	Residual risk of having an affected child^
ABCB11	Progressive familial intrahepatic cholestasis	AR	General	1 in 112	98%	1 in 5,551	1 in 2,486,848
ABCC8	Familial hyperinsulinism, ABCC8-related	AR	General	1 in 112	98%	1 in 5,551	1 in 2,486,848
ABCC8	Familial hyperinsulinism, ABCC8-related	AR	Ashkenazi Jewish	1 in 44	98%	1 in 2,151	1 in 378,576
ABCC8	Familial hyperinsulinism, ABCC8-related	AR	Finnish	1 in 25	98%	1 in 1,201	1 in 120,100
ABCC8	Familial hyperinsulinism, ABCC8-related	AR	Middle-Eastern	1 in 25	98%	1 in 1,201	1 in 120,100
ABCD1	Adrenoleucodystrophy, X-linked	XL	General	1 in 21,000	99%	1 in 2,099,901	1 in 8,399,804
ABCD4	Methylmalonic aciduria and homocystinuria, cblJ type	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
ACAD9	Acyl-CoA dehydrogenase-9 (ACAD9) deficiency	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
ACADM	Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency	AR	General	1 in 69	98%	1 in 3,401	1 in 938,676
ACADM	Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency	AR	Caucasian/European	1 in 52	99%	1 in 5,101	1 in 1,061,008
ACADM	Medium-chain acyl-CoA dehydrogenase (MCAD)deficiency	AR	East Asian	1 in 198	99%	1 in 19,701	<1 in 10 million
ACADM	Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency	AR	Native American	1 in 43	96%	1 in 1,051	1 in 180,772
ACADS	Short-chain acyl-CoA dehydrogenase (SCAD) deficiency	AR	General	1 in 85	99%	1 in 8,401	1 in 2,856,340
ACADS	Short-chain acyl-CoA dehydrogenase (SCAD) deficiency	AR	African/African American	1 in 52	99%	1 in 5,101	1 in 1,061,008
ACADS	Short-chain acyl-CoA dehydrogenase (SCAD) deficiency	AR	Caucasian/European	1 in 76	99%	1 in 7,501	1 in 2,280,304
ACADS	Short-chain acyl-CoA dehydrogenase (SCAD) deficiency	AR	Middle-Eastern	1 in 52	99%	1 in 5,101	1 in 1,061,008
ACADS	Short-chain acyl-CoA dehydrogenase (SCAD) deficiency	AR	South Asian/Indian	1 in 51	99%	1 in 5,001	1 in 1,020,204
ACADSB	Short branched-chain acyl-CoA dehydrogenase (SBCAD) deficiency	AR	General	1 in 368	99%	1 in 36,701	<1 in 10 million
ACADSB	Short branched-chain acyl-CoA dehydrogenase (SBCAD) deficiency	AR	Hmong	1 in 6	99%	1 in 501	<1 in 10 million
ACADSB	Short branched-chain acyl-CoA dehydrogenase (SBCAD) deficiency	AR	General	1 in 118	93%	1 in 1,672	1 in 789,184
ACADVL	Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency	AR	Middle-Eastern	1 in 74	93%	1 in 1,044	1 in 309,024
ACADVL	Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency	AR	Native American	1 in 61	93%	1 in 858	1 in 209,352
ACADVL	Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency	AR	South Asian/Indian	1 in 73	93%	1 in 1,030	1 in 300,760
ACAT1	3-ketothiolase deficiency	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
ACOX1	Peroxisomal acyl-CoA oxidase deficiency	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
ACSF3	Combined malonic and methylmalonic aciduria	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
ADA	Adenosine deaminase deficiency	AR	General	1 in 224	93%	1 in 3,187	1 in 2,855,552
ADAMTS2	Ehlers-Danlos syndrome, dermatosparaxis type 7C	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
ADAMTS2	Ehlers-Danlos syndrome, dermatosparaxis type 7C	AR	Ashkenazi Jewish	1 in 248	98%	1 in 12,351	<1 in 10 million
ADGRG1	Bilateral frontoparietal polymicrogyria	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
ADK	Hypermethioninaemia due to adenosine kinase deficiency	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
AGA	Aspartylglucosaminuria	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
AGA	Aspartylglucosaminuria	AR	Finnish	1 in 71	98%	1 in 3,501	1 in 994,284
AGL	Glycogen storage disease, type 3	AR	General	1 in 158	95%	1 in 3,141	1 in 1,985,112
AGL	Glycogen storage disease, type 3	AR	Faroese	1 in 28	95%	1 in 541	1 in 60,592
AGL	Glycogen storage disease, type 3	AR	Inuit	1 in 25	95%	1 in 481	1 in 48,100
AGL	Glycogen storage disease, type 3	AR	North African Jewish	1 in 37	95%	1 in 721	1 in 106,708
AGPS	Rhizomelic chondrodysplasia punctata, type 3	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
AGXT	Primary hyperoxaluria, type 1	AR	General	1 in 120	99%	1 in 11,901	1 in 5,712,480
AGXT	Primary hyperoxaluria, type 1	AR	Caucasian/European	1 in 173	99%	1 in 17,201	<1 in 10 million
AHCY	Hypermethioninaemia due to deficiency of Sadenosylhomocysteine hydrolase	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
AHI1	Joubert syndrome, AHI1-related	AR	General	1 in 448	99%	1 in 44,701	<1 in 10 million
AIPL1	Childhood-onset severe retinal dystrophy, AIPL1-related	AR	General	1 in 409	99%	1 in 40,801	<1 in 10 million
AIRE	Autoimmune polyendocrinopathy syndrome, type 1	AR	Finnish	1 in 79	98%	1 in 3,901	1 in 1,232,716
ALDH3A2	Sjögren-Larsson syndrome	AR	General	1 in 250	98%	1 in 12,451	<1 in 10 million
ALDH4A1	Hyperprolinaemia, type 2	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
ALDOB	Hereditary fructose intolerance	AR	General	1 in 122	99%	1 in 12,101	1 in 5,905,288
ALDOB	Hereditary fructose intolerance	AR	African/African American	1 in 250	99%	1 in 24,901	<1 in 10 million
ALDOB	Hereditary fructose intolerance	AR	Caucasian/European	1 in 67	99%	1 in 6,601	1 in 1,769,068
ALDOB	Hereditary fructose intolerance	AR	Middle-Eastern	1 in 97	99%	1 in 9,601	1 in 3,725,188
ALG6	Congenital disorder of glycosylation, type 1C	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
ALMS1	Alstrom syndrome	AR	General	1 in 500	98%	1 in 24,951	<1 in 10 million
ALPL	Hypophosphatasia	AR	General	1 in 158	95%	1 in 3,141	1 in 1,985,112
ALPL	Hypophosphatasia	AR	Caucasian/ European	1 in 274	95%	1 in 5,461	1 in 5,985,256
ALPL	Hypophosphatasia	AR	Mennonite	1 in 25	95%	1 in 481	1 in 48,100
AMT	Glycine encephalopathy, AMT-related	AR	General	1 in 373	98%	1 in 18,601	<1 in 10 million
AMT	Glycine encephalopathy, AMT-related	AR	Finnish	1 in 117	98%	1 in 5,801	1 in 2,714,868
AP1S2	X-linked intellectual disability, AP1S2-related	XL	General	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million

AR autosomal recessive; XL X-linked.
*If patient not shown to be a carrier. ^ If patient not identified as a carrier and partner not tested for AR conditions.

Monash IVF

Expanded Carrier Screen

Gene list (by gene)

Gene	Disorder	Inheritance	Population	Carrier frequency	Detection rate	Residual risk of being a carrier*	Residual risk of having an affected child^
AQP2	Nephrogenic diabetes insipidus	AR	General	<1 in 500	95%	1 in 9,981	<1 in 10 million
AQP2	Nephrogenic diabetes insipidus	AR	Finnish	1 in 169	95%	1 in 3,361	1 in 2,272,036
ARG1	Arginase deficiency	AR	General	1 in 296	98%	1 in 14,751	<1 in 10 million
ARL13B	Joubert syndrome, ARL13B-related	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
ARSA	Metachromatic leucodystrophy	AR	General	1 in 100	95%	1 in 1,981	1 in 792,400
ARSA	Metachromatic leucodystrophy	AR	Caucasian/European	1 in 78	95%	1 in 1,541	1 in 480,792
ARSB	Mucopolysaccharidosis, type 6 (Maroteaux-Lamy syndrome)	AR	General	1 in 250	98%	1 in 12,451	<1 in 10 million
ARSB	Mucopolysaccharidosis, type 6 (Maroteaux-Lamy syndrome)	AR	Western Australian	1 in 283	98%	1 in 14,101	<1 in 10 million
ARX	X-linked intellectual disability, ARX-related	XL	General	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
ASL	Argininosuccinate lyase deficiency	AR	General	1 in 132	90%	1 in 1,311	1 in 692,208
ASNS	Asparagine synthetase deficiency	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
ASPA	Canavan disease	AR	General	1 in 300	97%	1 in 9,968	<1 in 10 million
ASPA	Canavan disease	AR	Ashkenazi Jewish	1 in 55	96%	1 in 1,351	1 in 297,220
ASS1	Citrullinaemia	AR	General	1 in 119	96%	1 in 2,951	1 in 1,404,676
ASS1	Citrullinaemia	AR	East Asian	1 in 132	96%	1 in 3,276	1 in 1,729,728
ATMl	Ataxia-telangiectasia	AR	General	1 in 100	92%	1 in 1,239	1 in 495,600
ATP6V1B1	Renal tubular acidosis with deafness	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
ATP7A	Menkes disease	XL	General	1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
ATP7B	Wilson disease	AR	General	1 in 87	98%	1 in 4,301	1 in 1,496,748
ATP7B	Wilson disease	AR	Caucasian/European	1 in 42	98%	1 in 2,051	1 in 344,568
ATP7B	Wilson disease	AR	Ashkenazi Jewish	1 in 70	98%	1 in 3,451	1 in 966,280
ATRX	Alpha thalassaemia X-linked intellectual disability syndrome	XL	General	<1 in 250,000	99%	1 in 24,999,901	<1 in 10 million
BBS1	Bardet-Biedl syndrome, type 1	AR	General	1 in 367	99%	1 in 36,601	<1 in 10 million
BBS10	Bardet-Biedl syndrome, type 10	AR	General	1 in 395	99%	1 in 39,401	<1 in 10 million
BBS12	Bardet-Biedl syndrome, type 12	AR	General	1 in 791	99%	1 in 79,001	<1 in 10 million
BBS2	Bardet-Biedl syndrome 2	AR	General	1 in 621	99%	1 in 62,001	<1 in 10 million
BBS2	Bardet-Biedl syndrome 2	AR	Ashkenazi Jewish	1 in 107	99%	1 in 10,601	1 in 4,537,228
BBS2	Retinitis pigmentosa 74	AR	General	1 in 621	99%	1 in 62,001	<1 in 10 million
BBS2	Retinitis pigmentosa 74	AR	Ashkenazi Jewish	1 in 107	99%	1 in 10,601	1 in 4,537,228
BCKDHA	Maple syrup urine disease, type 1A	AR	General	1 in 321	98%	1 in 16,001	<1 in 10 million
BCKDHA	Maple syrup urine disease, type 1A	AR	Mennonite	1 in 10	98%	1 in 451	1 in 18,040
BCKDHB	Maple syrup urine disease, type 1B	AR	General	1 in 364	98%	1 in 18,151	<1 in 10 million
BCKDHB	Maple syrup urine disease, type 1B	AR	Ashkenazi Jewish	1 in 97	98%	1 in 4,801	1 in 1,862,788
BCS1L	Björnstad syndrome	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
BCS1L	GRACILE syndrome	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
BCS1L	Mitochondrial complex 3 deficiency	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
BLM	Bloom syndrome	AR	General	1 in 800	87%	1 in 6,147	<1 in 10 million
BLM	Bloom syndrome	AR	Ashkenazi Jewish	1 in 134	99%	1 in 13,301	1 in 7,129,336
BRWD3	X-linked intellectual disability, BRWD3-related	XL	General	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
BSND	Bartter syndrome	AR	General	1 in 500	98%	1 in 24,951	<1 in 10 million
BTD	Biotinidase deficiency	AR	General	1 in 124	99%	1 in 12,301	1 in 6,101,296
BTD	Biotinidase deficiency	AR	Caucasian/European	1 in 71	99%	1 in 7,001	1 in 1,988,284
BTD	Biotinidase deficiency	AR	Latino	1 in 136	99%	1 in 13,501	1 in 7,344,544
BTD	Biotinidase deficiency	AR	Middle-Eastern	1 in 55	99%	1 in 5,401	1 in 1,188,220
CAPN3	Limb-girdle muscular dystrophy, type 2A	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
CAPN3	Limb-girdle muscular dystrophy, type 2A	AR	Caucasian/European	1 in 103	98%	1 in 5,101	1 in 2,101,612
CASQ2	Catecholaminergic polymorphic ventricular tachycardia, CASQ2-related	AR	General	1 in 224	99%	1 in 22,301	<1 in 10 million
CBS	Homocystinuria due to cystathionine beta-synthase deficiency	AR	General	1 in 224	99%	1 in 22,301	<1 in 10 million
CBS	Homocystinuria due to cystathionine beta-synthase deficiency	AR	Caucasian/European	1 in 86	99%	1 in 8,501	1 in 2,924,344
CBS	Homocystinuria due to cystathionine beta-synthase deficiency	AR	Middle-Eastern	1 in 21	99%	1 in 2,001	1 in 168,084
CCDC103	Primary ciliary dyskinesia, type 17	AR	General	1 in 316	98%	1 in 15,751	<1 in 10 million
CCDC151	Primary ciliary dyskinesia, type 30	AR	General	1 in 365	98%	1 in 18,201	<1 in 10 million
CCDC39	Primary ciliary dyskinesia, type 14	AR	General	1 in 211	98%	1 in 10,501	1 in 8,862,844
CD40LG	Hyper IgM syndrome, X-linked	XL	General	1 in 50,000	98%	1 in 2,499,951	1 in 9,999,904
CDH23	Usher syndrome, type 1D	AR	General	1 in 285	90%	1 in 2,841	1 in 11,364
CEP290	Bardet-Biedl syndrome 14	AR	General	1 in 190	98%	1 in 9,451	1 in 7,182,760
CEP290	Joubert syndrome 5	AR	General	1 in 190	98%	1 in 9,451	1 in 7,182,760
CEP290	Leber congenital amaurosis 10	AR	General	1 in 190	98%	1 in 9,451	1 in 7,182,760
CEP290	Meckel syndrome 4	AR	General	1 in 190	98%	1 in 9,451	1 in 7,182,760
CEP290	Senior-Løken syndrome 6	AR	General	1 in 190	98%	1 in 9,451	1 in 7,182,760
CEP290	CEP290-related disorders	AR	General	1 in 190	98%	1 in 9,451	1 in 7,182,760
CERKL	Retinitis pigmentosa 26	AR	General	1 in 148	98%	1 in 7,351	1 in 4,351,792
CFTR	Cystic fibrosis	AR	General	1 in 32	99%	1 in 3,101	1 in 396,928
CFTR	Cystic fibrosis	AR	African/African American	1 in 61	99%	1 in 6,001	1 in 1,464,244
CFTR	Cystic fibrosis	AR	Ashkenazi Jewish	1 in 24	99%	1 in 2,301	1 in 220,896
CFTR	Cystic fibrosis	AR	Caucasian/European	1 in 25	99%	1 in 2,401	1 in 240,100
CFTR	Cystic fibrosis	AR	East Asian	1 in 94	99%	1 in 9,301	1 in 3,497,176
CFTR	Cystic fibrosis	AR	Latino	1 in 58	99%	1 in 5,701	1 in 1,322,632
CHM	Choroideraemia	XL	General	1 in 25,000	95%	1 in 499,981	1 in 1,999,964
CHRNE	Congenital myasthenic syndrome, CHRNE-related	AR	General	1 in 408	99%	1 in 40,701	<1 in 10 million
CHRNG	Multiple pterygium syndrome	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
CHST6	Macular corneal dystrophy, CHST6-related	AR	General	1 in 79	99%	1 in 7,801	1 in 2,465,116
CIITA	Bare lymphocyte syndrome, type 2	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million

AR autosomal recessive; XL X-linked.
*If patient not shown to be a carrier. ^ If patient not identified as a carrier and partner not tested for AR conditions.

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Gene	Disorder	Inheritance	Population	Carrier frequency	Detection rate	Residual risk of being a carrier*	Residual risk of having an affected child^
CLN3	Neuronal ceroid lipofuscinosis, CLN3-related	AR	General	1 in 230	98%	1 in 11,451	<1 in 10 million
CLN3	Neuronal ceroid lipofuscinosis, CLN3-related	AR	Finnish	1 in 72	98%	1 in 3,551	1 in 1,022,688
CLN5	Neuronal ceroid lipofuscinosis, CLN5-related	AR	General	<1 in 500	95%	1 in 9,981	<1 in 10 million
CLN5	Neuronal ceroid lipofuscinosis, CLN5-related	AR	Finnish	1 in 115	95%	1 in 2,281	1 in 1,049,260
CLN6	Neuronal ceroid lipofuscinosis, CLN6-related	AR	General	<1 in 500	92%	1 in 6,239	<1 in 10 million
CLN8	Neuronal ceroid lipofuscinosis, CLN8-related	AR	General	<1 in 500	95%	1 in 9,981	<1 in 10 million
CLN8	Neuronal ceroid lipofuscinosis, CLN8-related	AR	Finnish	1 in 135	95%	1 in 2,681	1 in 1,447,740
CLRN1	Usher syndrome, type 3A	AR	General	1 in 500	98%	1 in 24,951	<1 in 10 million
CLRN1	Usher syndrome, type 3A	AR	Ashkenazi Jewish	1 in 120	98%	1 in 5,951	1 in 2,856,480
CLRN1	Usher syndrome, type 3A	AR	Finnish	1 in 70	98%	1 in 3,451	1 in 966,280
CNGA1	Retinitis pigmentosa, CNGA1-related	AR	General	1 in 210	99%	1 in 20,901	<1 in 10 million
CNGB1	Retinitis pigmentosa, CNGB1-related	AR	General	1 in 296	99%	1 in 29,501	<1 in 10 million
CNGB3	Achromatopsia	AR	General	1 in 87	99%	1 in 8,601	1 in 2,993,148
CNGB3	Achromatopsia	AR	Micronesian	1 in 2	99%	1 in 101	1 in 808
COL27A1	Steel syndrome	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
COL4A3	Alport syndrome, COL4A3-related	AR	General	1 in 267	98%	1 in 13,301	<1 in 10 million
COL4A3	Alport syndrome, COL4A3-related	AR	Ashkenazi Jewish	1 in 188	98%	1 in 9,351	1 in 7,031,952
COL4A4	Alport syndrome, COL4A4-related	AR	General	1 in 267	98%	1 in 13,301	<1 in 10 million
COL4A5	Alport syndrome, COL4A5-related	XL	General	1 in 139	98%	1 in 6,901	1 in 27,604
COL7A1	Dystrophic epidermolysis bullosa	AR	General	1 in 196	97%	1 in 6,501	1 in 5,096,784
CPS1	Carbamoyl phosphate synthetase 1 deficiency	AR	General	1 in 570	98%	1 in 28,451	<1 in 10 million
CPT1A	Carnitine palmitoyltransferase 1A deficiency	AR	General	1 in 354	90%	1 in 3,531	1 in 4,999,896
CPT1A	Carnitine palmitoyltransferase 1A deficiency	AR	Hutterite	1 in 16	90%	1 in 151	1 in 9,664
CPT2	Carnitine palmitoyltransferase 2 deficiency	AR	General	<1 in 500	95%	1 in 9,981	<1 in 10 million
CPT2	Carnitine palmitoyltransferase 2 deficiency	AR	Ashkenazi Jewish	1 in 51	95%	1 in 1,001	1 in 204,204
CRB1	Leber congenital amaurosis 8	AR	General	1 in 104	98%	1 in 5,151	1 in 2,142,816
CRB1	Retinitis pigmentosa 12	AR	General	1 in 104	98%	1 in 5,151	1 in 2,142,816
CTNS	Cystinosis	AR	General	1 in 158	99%	1 in 15,701	1 in 9,923,032
CTNS	Cystinosis	AR	British	1 in 81	99%	1 in 8,001	1 in 2,592,324
CTSK	Pycnodysostosis	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
CUL4B	X-linked intellectual disability, CUL4B-related	XL	General	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
CYBA	Chronic granulomatous disease	AR	General	1 in 224	99%	1 in 22,301	<1 in 10 million
CYBB	Chronic granulomatous disease, X-linked	XL	General	1 in 149,254	99%	1 in 14925301	<1 in 10 million
CYP11B1	Congenital adrenal hyperplasia due to 11-betahydroxylase deficiency	AR	General	1 in 158	98%	1 in 7,851	1 in 4,961,832
CYP11B1	Congenital adrenal hyperplasia due to 11-betahydroxylase deficiency	AR	Moroccan Jewish	1 in 35	98%	1 in 1,701	1 in 238,140
CYP11B2	Corticosterone methyloxidase deficiency	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
CYP17A1	Congenital adrenal hyperplasia due to 17-alphahydroxylase deficiency	AR	General	1 in 500	98%	1 in 24,951	<1 in 10 million
CYP19A1	Aromatase deficiency	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
CYP1B1	Primary congenital glaucoma	AR	General	1 in 50	99%	1 in 4,901	1 in 980,200
CYP21A2	Congenital adrenal hyperplasia due to 21-hydroxylase deficiency	AR	General	1 in 61	99%	1 in 6,001	1 in 1,464,244
CYP21A2	Congenital adrenal hyperplasia due to 21-hydroxylase deficiency	AR	Inuit	1 in 9	99%	1 in 801	1 in 28,836
CYP21A2	Congenital adrenal hyperplasia due to 21-hydroxylase deficiency	AR	Middle-Eastern	1 in 35	99%	1 in 3,401	1 in 476,140
CYP27A1	Cerebrotendinous xanthomatosis	AR	General	1 in 500	98%	1 in 24,951	<1 in 10 million
CYP27A1	Cerebrotendinous xanthomatosis	AR	Moroccan Jewish	1 in 5	98%	1 in 201	1 in 4,020
DBT	Maple syrup urine disease, type 2	AR	General	1 in 481	98%	1 in 24,001	<1 in 10 million
DCLRE1C	Severe combined immunodeficiency with sensitivity to ionising radiation	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
DCX	Lissencephaly, X-linked	XL	General	1 in 42,500	98%	1 in 2,124,951	1 in 8,499,904
DHCR7	Smith-Lemli-Opitz syndrome	AR	General	1 in 30	96%	1 in 726	1 in 87,120
DHCR7	Smith-Lemli-Opitz syndrome	AR	African/African American	1 in 138	96%	1 in 3,426	1 in 1,891,152
DHCR7	Smith-Lemli-Opitz syndrome	AR	Ashkenazi Jewish	1 in 36	96%	1 in 876	1 in 126,144
DHDDS	Retinitis pigmentosa 59	AR	General	1 in 296	98%	1 in 14,751	<1 in 10 million
DHDDS	Retinitis pigmentosa 59	AR	Ashkenazi Jewish	1 in 118	98%	1 in 5,851	1 in 2,761,672
DLD	Dihydrolipoamide dehydrogenase deficiency	AR	General	1 in 500	98%	1 in 24,951	<1 in 10 million
DLD	Dihydrolipoamide dehydrogenase deficiency	AR	Ashkenazi Jewish	1 in 107	98%	1 in 5,301	1 in 2,268,828
DLG3	X-linked intellectual disability, DLG3-related	XL	General	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
DMD	Duchenne muscular dystrophy	XL	General	1 in 2,350	93%	1 in 33,558	1 in 134,260
DNAH5	Primary ciliary dyskinesia, DNAH5-related	AR	General	1 in 142	98%	1 in 7,051	1 in 4,004,968
DNAI1	Primary ciliary dyskinesia, DNAI1-related	AR	General	1 in 230	98%	1 in 11,451	<1 in 10 million
DNAI2	Primary ciliary dyskinesia, DNAI2-related	AR	General	1 in 447	98%	1 in 22,301	<1 in 10 million
DNAL1	Primary ciliary dyskinesia, DNAL1-related	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
DPYD	Dihydropyrimidine dehydrogenase deficiency	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
DUOX2	Congenital hypothyroidism, DUOX2-related	AR	General	1 in 366	91%	1 in 4,057	1 in 5,938,797
DUOXA2	Congenital hypothyroidism, DUOXA2-related	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
DYSF	Limb-girdle muscular dystrophy, type 2B	AR	General	<1 in 500	95%	1 in 9,981	<1 in 10 million
DYSF	Limb-girdle muscular dystrophy, type 2B	AR	Japanese	1 in 332	95%	1 in 6,621	1 in 8,792,688
DYSF	Limb-girdle muscular dystrophy, type 2B	AR	Libyan Jewish	1 in 18	95%	1 in 341	1 in 24,552
EDA	Hypohidrotic ectodermal dysplasia	XL	General	1 in 14,167	99%	1 in 1,416,601	1 in 5,666,472
EIF2AK3	Wolcott-Rallison syndrome	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
EIF2B5	Leucoencephalopathy with vanishing white matter	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
ELP1	Familial dysautonomia	AR	General	1 in 300	99%	1 in 29,901	<1 in 10 million
ELP1	Familial dysautonomia	AR	Ashkenazi Jewish	1 in 31	99%	1 in 3,001	1 in 372,124
EMD	Emery-Dreifuss muscular dystrophy	XL	General	1 in 81,967	99%	1 in 8,196,601	<1 in 10 million
ERCC6	Cockayne syndrome, type B	AR	General	1 in 500	99%	1 in 49,901	<1 in 10 million

AR autosomal recessive; XL X-linked.
*If patient not shown to be a carrier. ^ If patient not identified as a carrier and partner not tested for AR conditions.

Monash IVF

Expanded Carrier Screen

Gene list (by gene)

Gene	Disorder	Inheritance	Population	Carrier frequency	Detection rate	Residual risk of being a carrier*	Residual risk of having an affected child^
ERCC6	Cockayne syndrome, type B	AR	Japanese	1 in 74	99%	1 in 7,301	1 in 2,161,096
ERCC6	De Sanctis-Cacchione syndrome	AR	General	1 in 500	99%	1 in 49,901	<1 in 10 million
ERCC6	De Sanctis-Cacchione syndrome	AR	Japanese	1 in 74	99%	1 in 7,301	1 in 2,161,096
ERCC8	Cockayne syndrome, type A	AR	General	1 in 822	98%	1 in 41,051	<1 in 10 million
ESCO2	Roberts syndrome	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
ETFA	Glutaric aciduria, type 2A	AR	General	1 in 500	98%	1 in 24,951	<1 in 10 million
ETFB	Glutaric aciduria, type 2B	AR	General	1 in 500	98%	1 in 24,951	<1 in 10 million
ETFDH	Glutaric aciduria, type 2C	AR	General	1 in 250	98%	1 in 12,451	<1 in 10 million
ETFDH	Glutaric aciduria, type 2C	AR	East Asian	1 in 74	98%	1 in 3,651	1 in 1,080,696
ETHE1	Ethylmalonic encephalopathy	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
EVC	Ellis-van Creveld syndrome, EVC-related	AR	General	1 in 142	98%	1 in 7,051	1 in 4,004,968
EVC	Ellis-van Creveld syndrome, EVC-related	AR	Amish	1 in 7	98%	1 in 301	1 in 8,428
EVC	Weyers acrofacial dysostosis, EVC-related	AR	General	1 in 142	98%	1 in 7,051	1 in 4,004,968
EVC	Weyers acrofacial dysostosis, EVC-related	AR	Amish	1 in 7	98%	1 in 301	1 in 8,428
EVC2	Ellis-van Creveld syndrome, EVC2-related	AR	General	1 in 240	98%	1 in 11,951	<1 in 10 million
EVC2	Ellis-van Creveld syndrome, EVC2-related	AR	Amish	1 in 7	98%	1 in 301	1 in 8,428
EVC2	Weyers acrodental dysostosis, EVC2-related	AR	General	1 in 240	98%	1 in 11,951	<1 in 10 million
EVC2	Weyers acrodental dysostosis, EVC2-related	AR	Amish	1 in 7	98%	1 in 301	1 in 8,428
EXOSC3	Pontocerebellar hypoplasia, type 1B	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
EYS	Retinitis pigmentosa 25	AR	General	1 in 66	98%	1 in 3,251	1 in 858,264
F11	Factor 11 deficiency	AR	General	1 in 500	98%	1 in 24,951	<1 in 10 million
F11	Factor 11 deficiency	AR	Ashkenazi Jewish	1 in 11	98%	1 in 501	1 in 22,044
F8	Haemophilia A	XL	General	1 in 3,250	48%	1 in 6,249	1 in 25,000
F9	Haemophilia B	XL	General	1 in 15,000	99%	1 in 1,499,901	1 in 5,999,804
FAH	Tyrosinaemia, type 1	AR	General	1 in 99	95%	1 in 1,961	1 in 776,556
FAH	Tyrosinaemia, type 1	AR	Ashkenazi Jewish	1 in 150	95%	1 in 2,981	1 in 1,788,600
FAH	Tyrosinaemia, type 1	AR	French Canadian	1 in 66	95%	1 in 1,301	1 in 343,464
FAH	Tyrosinaemia, type 1	AR	South Asian/Indian	1 in 172	95%	1 in 3,421	1 in 2,353,648
FAM161A	Retinitis pigmentosa 28	AR	General	1 in 296	98%	1 in 14,751	<1 in 10 million
FANCA	Fanconi anaemia group A	AR	General	1 in 239	98%	1 in 11,901	<1 in 10 million
FANCC	Fanconi anaemia group C	AR	General	1 in 535	99%	1 in 53,401	<1 in 10 million
FANCC	Fanconi anaemia group C	AR	Ashkenazi Jewish	1 in 99	99%	1 in 9,801	1 in 3,881,196
FANCG	Fanconi anaemia group G	AR	General	1 in 632	90%	1 in 6,311	<1 in 10 million
FGD1	X-linked Aarskog-Scott syndrome	XL	General	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
FH	Fumarase deficiency	AR	General	<1 in 500	90%	1 in 4,991	1 in 9,982,000
FKRP	Muscular dystrophy-dystroglycanopathy, FKRP-related	AR	General	1 in 158	98%	1 in 7,851	1 in 4,961,832
FKTN	Muscular dystrophy-dystroglycanopathy, FKTN-related	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
FKTN	Muscular dystrophy-dystroglycanopathy, FKTN-related	AR	Ashkenazi Jewish	1 in 150	99%	1 in 14,901	1 in 8,940,600
FKTN	Muscular dystrophy-dystroglycanopathy, FKTN-related	AR	Japanese	1 in 82	99%	1 in 8,101	1 in 2,657,128
FKTN	Fukuyama congenital muscular dystrophy	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
FKTN	Fukuyama congenital muscular dystrophy	AR	Ashkenazi Jewish	1 in 150	99%	1 in 14,901	1 in 8,940,600
FKTN	Fukuyama congenital muscular dystrophy	AR	Japanese	1 in 82	99%	1 in 8,101	1 in 2,657,128
FMR1	Fragile X syndrome	XL	General	1 in 151	99%	1 in 15,001	1 in 60,004
FMR1	Fragile X syndrome	XL	Ashkenazi Jewish	1 in 115	99%	1 in 11,401	1 in 45,604
FTCD	Glutamate formiminotransferase deficiency	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
FTSJ1	X-linked intellectual disability, FTSJ1-related	XL	General	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
G6PC	Glycogen storage disease, type 1A	AR	General	1 in 177	95%	1 in 3,521	1 in 2,492,868
G6PC	Glycogen storage disease, type 1A	AR	Ashkenazi Jewish	1 in 64	95%	1 in 1,261	1 in 322,816
GAA	Pompe disease	AR	General	1 in 100	98%	1 in 4,951	1 in 1,980,400
GAA	Pompe disease	AR	African/African American	1 in 60	98%	1 in 2,951	1 in 708,240
GAA	Pompe disease	AR	East Asian	1 in 112	98%	1 in 5,551	1 in 2,486,848
GALC	Krabbe disease	AR	General	1 in 158	99%	1 in 15,701	1 in 9,923,032
GALC	Krabbe disease	AR	Israeli Druze	1 in 6	99%	1 in 501	1 in 12,024
GALE	Galactose epimerase deficiency	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
GALK1	Galactokinase deficiency	AR	General	1 in 110	95%	1 in 2,181	1 in 959,640
GALK1	Galactokinase deficiency	AR	Irish	1 in 64	95%	1 in 1,261	1 in 322,816
GALNS	Mucopolysaccharidosis, type 4A (Morquio syndrome A)	AR	General	1 in 224	97%	1 in 7,434	1 in 6,660,864
GALT	Galactosaemia	AR	General	1 in 110	95%	1 in 2,181	1 in 959,640
GALT	Galactosaemia	AR	African/African American	1 in 94	95%	1 in 1,861	1 in 699,736
GAMT	Guanidinoacetate methyltransferase deficiency	AR	General	1 in 371	99%	1 in 37,001	<1 in 10 million
GBA	Gaucher disease	AR	General	1 in 77	99%	1 in 7,601	1 in 2,341,108
GBA	Gaucher disease	AR	African/African American	1 in 35	99%	1 in 3,401	1 in 476,140
GBA	Gaucher disease	AR	Ashkenazi Jewish	1 in 15	99%	1 in 1,401	1 in 84,060
GBE1	Glycogen storage disease, type 4	AR	General	1 in 387	99%	1 in 38,601	<1 in 10 million
GCDH	Glutaric aciduria, type 1	AR	General	1 in 87	98%	1 in 4,301	1 in 1,496,748
GCDH	Glutaric aciduria, type 1	AR	Amish	1 in 9	98%	1 in 401	1 in 14,436
GDAP1	Charcot-Marie-Tooth disease, GDAP1-related	AR	General	1 in 152	99%	1 in 15,101	1 in 9,181,408
GFM1	Combined oxidative phosphorylation deficiency, GFM1-related	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
GJB1	Charcot-Marie-Tooth disease, X-linked type 1	XL	General	1 in 667	90%	1 in 6,661	1 in 26,644
GJB2	Non-syndromic hearing loss, GJB2-related	AR	General	1 in 42	99%	1 in 4,101	1 in 688,968
GJB2	Non-syndromic hearing loss, GJB2-related	AR	African/African American	1 in 25	99%	1 in 2,401	1 in 240,100
GJB2	Non-syndromic hearing loss, GJB2-related	AR	Ashkenazi Jewish	1 in 21	99%	1 in 2,001	1 in 168,084

AR autosomal recessive; XL X-linked.
*If patient not shown to be a carrier. ^ If patient not identified as a carrier and partner not tested for AR conditions.

Monash IVF

Expanded Carrier Screen

Gene list (by gene)

Gene	Disorder	Inheritance	Population	Carrier frequency	Detection rate	Residual risk of being a carrier*	Residual risk of having an affected child^
GJB2	Non-syndromic hearing loss, GJB2-related	AR	Caucasian/European	1 in 33	99%	1 in 3,201	1 in 422,532
GJB2	Non-syndromic hearing loss, GJB2-related	AR	Latino	1 in 100	99%	1 in 9,901	1 in 3,960,400
GJB2	Non-syndromic hearing loss, GJB2-related	AR	Middle-Eastern	1 in 83	99%	1 in 8,201	1 in 2,722,732
GJB2	Non-syndromic hearing loss, GJB2-related	AR	South Asian/Indian	1 in 148	99%	1 in 14,701	1 in 8,702,992
GJB6	Non-syndromic hearing loss, GJB6-related	AR	General	1 in 423	99%	1 in 42,201	<1 in 10 million
GLA	Fabry disease	XL	General	1 in 25,000	99%	1 in 2,499,901	1 in 9,999,804
GLB1	Mucopolysaccharidosis, type 4B (Morquio syndrome B)	AR	General	1 in 134	99%	1 in 13,301	1 in 7,129,336
GLB1	Mucopolysaccharidosis, type 4B (Morquio syndrome B)	AR	Maltese	1 in 30	99%	1 in 2,901	1 in 348,120
GLB1	Mucopolysaccharidosis, type 4B (Morquio syndrome B)	AR	Roma	1 in 50	99%	1 in 4,901	1 in 980,200
GLB1	GM1-gangliosidosis	AR	General	1 in 134	99%	1 in 13,301	1 in 7,129,336
GLB1	GM1-gangliosidosis	AR	Maltese	1 in 30	99%	1 in 2,901	1 in 348,120
GLB1	GM1-gangliosidosis	AR	Roma	1 in 50	99%	1 in 4,901	1 in 980,200
GLDC	Glycine encephalopathy, GLDC-related	AR	General	1 in 193	98%	1 in 9,601	1 in 7,411,972
GLDC	Glycine encephalopathy, GLDC-related	AR	British Columbia Canadian	1 in 125	99%	1 in 12,401	1 in 6,200,500
GLDC	Glycine encephalopathy, GLDC-related	AR	Finnish	1 in 117	99%	1 in 11,601	1 in 5,429,268
GLE1	Lethal congenital contracture syndrome 1	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
GLE1	Lethal congenital contracture syndrome 1	AR	Finnish	1 in 80	98%	1 in 3,951	1 in 1,264,320
GNE	Inclusion body myopathy, type 2 (Nonaka myopathy)	AR	General	<1 in 500	80%	1 in 2,496	1 in 4,992,000
GNE	Inclusion body myopathy, type 2 (Nonaka myopathy)	AR	Iranian Jewish	1 in 11	80%	1 in 51	1 in 2,244
GNPTAB	Mucolipidosis 3 alpha/beta	AR	General	<1 in 500	95%	1 in 9,981	<1 in 10 million
GNPTAB	Mucolipidosis 2 alpha/beta	AR	General	<1 in 500	95%	1 in 9,981	<1 in 10 million
GNPTG	Mucolipidosis 3 gamma	AR	General	<1 in 500	95%	1 in 9,981	<1 in 10 million
GNRHR	Hypogonadotropic hypogonadism, GNRHR-related	AR	General	1 in 347	99%	1 in 34,601	<1 in 10 million
GNS	Mucopolysaccharidosis, type 3D (Sanfilippo syndrome D)	AR	General	1 in 500	98%	1 in 24,951	<1 in 10 million
GP1BA	Bernard-Soulier syndrome, type A1	AR	General	1 in 500	98%	1 in 24,951	<1 in 10 million
GP9	Bernard-Soulier syndrome, type C	AR	General	1 in 500	98%	1 in 24,951	<1 in 10 million
GPR143	X-linked ocular albinism, GPR143-related	XL	General	1 in 25,000	99%	1 in 2,499,901	<1 in 10 million
GRHPR	Primary hyperoxaluria, type 2	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
GUSB	Mucopolysaccharidosis, type 7	AR	General	1 in 250	98%	1 in 12,451	<1 in 10 million
HADHA	Long-chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
HADHA	Long-chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency	AR	Finnish	1 in 124	98%	1 in 6,151	1 in 3,050,896
HADHA	Trifunctional protein deficiency	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
HADHA	Trifunctional protein deficiency	AR	Finnish	1 in 124	98%	1 in 6,151	1 in 3,050,896
HAX1	Severe congenital neutropaenia, HAX1-related	AR	General	1 in 224	98%	1 in 11,151	1 in 9,991,296
HBA1	Alpha thalassaemia	AR	General	1 in 20	90%	1 in 191	1 in 15,280
HBA1	Alpha thalassaemia	AR	African/African American	1 in 3	90%	1 in 21	1 in 252
HBA1	Alpha thalassaemia	AR	Ashkenazi Jewish	1 in 13	90%	1 in 121	1 in 6,292
HBA1	Alpha thalassaemia	AR	East Asian	1 in 8	90%	1 in 71	1 in 2,272
HBA1	Alpha thalassaemia	AR	Middle-Eastern	1 in 3	90%	1 in 21	1 in 252
HBA1	Alpha thalassaemia	AR	South Asian/Indian	1 in 5	90%	1 in 41	1 in 820
HBA2	Alpha thalassaemia	AR	General	1 in 20	90%	1 in 191	1 in 15,280
HBA2	Alpha thalassaemia	AR	African/African American	1 in 3	90%	1 in 21	1 in 252
HBA2	Alpha thalassaemia	AR	Ashkenazi Jewish	1 in 13	90%	1 in 121	1 in 6,292
HBA2	Alpha thalassaemia	AR	East Asian	1 in 8	90%	1 in 71	1 in 2,272
HBA2	Alpha thalassaemia	AR	Middle-Eastern	1 in 3	90%	1 in 21	1 in 252
HBA2	Alpha thalassaemia	AR	South Asian/Indian	1 in 5	90%	1 in 41	1 in 820
HBB	Sickle cell disease	AR	General	1 in 158	95%	1 in 3,141	1 in 1,985,112
HBB	Sickle cell disease	AR	African/African American	1 in 10	95%	1 in 181	1 in 7,240
HBB	Sickle cell disease	AR	East Asian	1 in 50	95%	1 in 981	1 in 196,200
HBB	Sickle cell disease	AR	Latino	1 in 128	95%	1 in 2,541	1 in 1,300,992
HBB	Sickle cell disease	AR	Mediterranean	1 in 3	95%	1 in 41	1 in 492
HBB	Sickle cell disease	AR	South Asian/Indian	1 in 25	95%	1 in 481	1 in 48,100
HBB	Beta thalassaemia	AR	General	1 in 158	95%	1 in 3,141	1 in 1,985,112
HBB	Beta thalassaemia	AR	African/African American	1 in 10	95%	1 in 181	1 in 7,240
HBB	Beta thalassaemia	AR	East Asian	1 in 50	95%	1 in 981	1 in 196,200
HBB	Beta thalassaemia	AR	Latino	1 in 128	95%	1 in 2,541	1 in 1,300,992
HBB	Beta thalassaemia	AR	Mediterranean	1 in 3	95%	1 in 41	1 in 492
HBB	Beta thalassaemia	AR	South Asian/Indian	1 in 25	95%	1 in 481	1 in 48,100
HEXA	Tay-Sachs disease	AR	General	1 in 300	99%	1 in 29,901	<1 in 10 million
HEXA	Tay-Sachs disease	AR	Ashkenazi Jewish	1 in 27	99%	1 in 2,601	1 in 280,908
HEXB	Sandhoff disease	AR	General	1 in 600	98%	1 in 29,951	<1 in 10 million
HGD	Alkaptonuria	AR	General	1 in 250	90%	1 in 2,491	1 in 2,491,000
HGSNAT	Mucopolysaccharidosis, type 3C (Sanfilippo syndrome C)	AR	General	1 in 434	98%	1 in 21,651	<1 in 10 million
HGSNAT	Mucopolysaccharidosis, type 3C (Sanfilippo syndrome C)	AR	Caucasian/European	1 in 345	98%	1 in 17,201	<1 in 10 million
HJV	Haemochromatosis, type 2A	AR	General	1 in 500	99%	1 in 49,901	<1 in 10 million
HLCS	Holocarboxylase synthetase deficiency	AR	General	1 in 500	98%	1 in 24,951	<1 in 10 million

AR autosomal recessive; XL X-linked.
*If patient not shown to be a carrier. ^ If patient not identified as a carrier and partner not tested for AR conditions.

Monash IVF

Expanded Carrier Screen

Gene list (by gene)

Gene	Disorder	Inheritance	Population	Carrier frequency	Detection rate	Residual risk of being a carrier*	Residual risk of having an affected child^
HMGCL	3-hydroxy-3-methylglutaryl-CoA lyase deficiency	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
HOGA1	Primary hyperoxaluria, type 3	AR	General	1 in 184	99%	1 in 18,301	<1 in 10 million
HPS1	Heřmanský-Pudlák syndrome 1	AR	General	1 in 354	98%	1 in 17,651	<1 in 10 million
HPS1	Heřmanský–Pudlák syndrome 1	AR	Puerto Rican	1 in 21	98%	1 in 1,001	1 in 84,084
HPS3	Heřmanský–Pudlák syndrome 3	AR	General	1 in 354	98%	1 in 17,651	<1 in 10 million
HSD17B4	D-bifunctional protein deficiency	AR	General	1 in 158	98%	1 in 7,851	1 in 4,961,832
HSD3B2	Congenital adrenal hyperplasia due to 3-betahydroxysteroid dehydrogenase 2 deficiency	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
HYAL1	Mucopolysaccharidosis, type 9	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
HYLS1	Hydrolethalus syndrome	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
HYLS1	Hydrolethalus syndrome	AR	Finnish	1 in 50	98%	1 in 2,451	1 in 490,200
IDH3B	Retinitis pigmentosa, IDH3B-related	AR	General	1 in 296	99%	1 in 29,501	<1 in 10 million
IDS	Mucopolysaccharidosis, type 2 (Hunter syndrome)	XL	General	1 in 50,000	91%	1 in 555,545	1 in 2,222,204
IDUA	Mucopolysaccharidosis, type 1 (Hurler syndrome)	AR	General	<1 in 500	95%	1 in 9,981	<1 in 10 million
IDUA	Mucopolysaccharidosis, type 1 (Hurler syndrome)	AR	Caucasian/European	1 in 153	95%	1 in 3,041	1 in 1,861,092
IL1RAPL1	X-linked intellectual disability, IL1RAPL1-related	XL	General	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
IL2RG	Severe combined immunodeficiency, X-linked	XL	General	1 in 25,000	99%	1 in 2,499,901	1 in 9,999,804
IVD	Isovaleric acidaemia	AR	General	1 in 167	90%	1 in 1,661	1 in 1,109,548
IVD	Isovaleric acidaemia	AR	African/African American	1 in 100	90%	1 in 991	1 in 396,400
IVD	Isovaleric acidaemia	AR	Caucasian/European	1 in 115	90%	1 in 1,141	1 in 524,860
IVD	Isovaleric acidaemia	AR	East Asian	1 in 407	90%	1 in 4,061	1 in 6,611,308
IYD	Thyroid dyshormonogenesis, IYD-related	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
JAK3	Severe combined immunodeficiency, JAK3-related	AR	General	1 in 299	99%	1 in 29,801	<1 in 10 million
KCNJ11	Congenital hyperinsulinism	AR	General	1 in 423	99%	1 in 42,201	<1 in 10 million
KCNJ11	Congenital hyperinsulinism	AR	Caucasian/European	1 in 232	99%	1 in 23,101	<1 in 10 million
KCNJ11	Permanent neonatal diabetes mellitus	AR	General	1 in 423	99%	1 in 42,201	<1 in 10 million
KCNJ11	Permanent neonatal diabetes mellitus	AR	Caucasian/European	1 in 232	99%	1 in 23,101	<1 in 10 million
KDM5C	X-linked intellectual disability, KDM5C-related	XL	General	<1 in 50,000	98%	1 in 2,499,951	<1 in 10 million
L1CAM	L1 syndrome	XL	General	1 in 15,000	99%	1 in 1,499,901	1 in 5,999,804
LAMA2	Muscular dystrophy, LAMA2-related	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
LAMA2	Muscular dystrophy, LAMA2-related	AR	Caucasian/European	1 in 125	99%	1 in 12,401	1 in 6,200,500
LAMA3	Junctional epidermolysis bullosa, LAMA3-related	AR	General	1 in 781	98%	1 in 39,001	<1 in 10 million
LAMA3	Laryngo-onycho-cutaneous syndrome	AR	General	1 in 781	98%	1 in 39,001	<1 in 10 million
LAMB3	Junctional epidermolysis bullosa, LAMB3-related	AR	General	1 in 781	98%	1 in 39,001	<1 in 10 million
LAMC2	Junctional epidermolysis bullosa, LAMC2-related	AR	General	1 in 781	98%	1 in 39,001	<1 in 10 million
LCA5	Leber congenital amaurosis 5	AR	General	1 in 500	98%	1 in 24,951	<1 in 10 million
LHX3	Combined pituitary hormone deficiency 3	AR	General	1 in 45	98%	1 in 2,201	1 in 396,180
LIFR	Stüve-Wiedemann syndrome	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
LIPA	Lysosomal acid lipase deficiency	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
LIPA	Lysosomal acid lipase deficiency	AR	Caucasian/European	1 in 112	99%	1 in 11,101	1 in 4,973,248
LMBRD1	Methylmalonic aciduria and homocystinuria, cblF type	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
LOXHD1	Non-syndromic hearing loss, LOXHD1-related	AR	General	1 in 500	98%	1 in 24,951	<1 in 10 million
LOXHD1	Non-syndromic hearing loss, LOXHD1-related	AR	Ashkenazi Jewish	1 in 180	98%	1 in 8,951	1 in 6,444,720
LPL	Familial lipoprotein lipase deficiency	AR	General	1 in 500	99%	1 in 49,901	<1 in 10 million
LPL	Familial lipoprotein lipase deficiency	AR	French Canadian	1 in 46	99%	1 in 4,501	1 in 828,184
LRPPRC	Leigh syndrome with complex 4 deficiency	AR	General	1 in 447	98%	1 in 22,301	<1 in 10 million
LRPPRC	Leigh syndrome with complex 4 deficiency	AR	Faroese	1 in 21	98%	1 in 1,001	1 in 84,084
LRPPRC	Leigh syndrome with complex 4 deficiency	AR	French Canadian	1 in 22	98%	1 in 1,051	1 in 92,488
LYST	Chediak-Higashi syndrome	AR	General	<1 in 500	90%	1 in 4,991	1 in 9,982,000
MAN2B1	Alpha-mannosidosis	AR	General	1 in 354	99%	1 in 35,301	<1 in 10 million
MAN2B1	Alpha-mannosidosis	AR	Caucasian/European	1 in 274	99%	1 in 27,301	<1 in 10 million
MCCC1	3-methylcrotonyl-CoA carboxylase 1 deficiency (3-MCC deficiency)	AR	General	1 in 95	98%	1 in 4,701	1 in 1,786,380
MCCC2	3-methylcrotonyl-CoA carboxylase 2 deficiency (3-MCC deficiency)	AR	General	1 in 95	98%	1 in 4,701	1 in 1,786,380
MCEE	Methylmalonyl-CoA epimerase deficiency	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
MCOLN1	Mucolipidosis 4	AR	General	1 in 300	99%	1 in 29,901	<1 in 10 million
MCOLN1	Mucolipidosis 4	AR	Ashkenazi Jewish	1 in 100	99%	1 in 9,901	1 in 3,960,400
MED17	Postnatal progressive microcephaly with seizures and brain atrophy	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
MESP2	Spondylocostal dysostosis	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
MFSD8	Neuronal ceroid lipofuscinosis, MFSD8-related	AR	General	<1 in 500	95%	1 in 9,981	<1 in 10 million
MKS1	Joubert syndrome 28	AR	General	1 in 260	98%	1 in 12,951	<1 in 10 million
MKS1	Joubert syndrome 28	AR	Finnish	1 in 47	98%	1 in 2,301	1 in 432,588
MKS1	Meckel syndrome 1	AR	General	1 in 260	98%	1 in 12,951	<1 in 10 million
MKS1	Meckel syndrome 1	AR	Finnish	1 in 47	98%	1 in 2,301	1 in 432,588
MKS1	Bardet-Biedl syndrome 13	AR	General	1 in 260	98%	1 in 12,951	<1 in 10 million
MKS1	Bardet-Biedl syndrome 13	AR	Finnish	1 in 47	98%	1 in 2,301	1 in 432,588

AR autosomal recessive; XL X-linked.
*If patient not shown to be a carrier. ^ If patient not identified as a carrier and partner not tested for AR conditions.

Monash IVF

Expanded Carrier Screen

Gene list (by gene)

Gene	Disorder	Inheritance	Population	Carrier frequency	Detection rate	Residual risk of being a carrier*	Residual risk of having an affected child^
MLC1	Megalencephalic leucoencephalopathy with subcortical cysts	AR	General	<1 in 500	97%	1 in 16,634	<1 in 10 million
MMAA	Methylmalonic aciduria, cblA type	AR	General	1 in 301	97%	1 in 10,001	<1 in 10 million
MMAB	Methylmalonic aciduria, cblB type	AR	General	1 in 435	98%	1 in 21,701	<1 in 10 million
MMACHC	Methylmalonic aciduria and homocystinuria, cblC type	AR	General	1 in 134	90%	1 in 1,331	1 in 713,416
MMADHC	Methylmalonic aciduria and homocystinuria, cblD type	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
MPI	Congenital disorder of glycosylation, type 1B	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
MPL	Congenital amegakaryocytic thrombocytopaenia	AR	General	1 in 102	98%	1 in 5,051	1 in 2,060,808
MPL	Congenital amegakaryocytic thrombocytopaenia	AR	Ashkenazi Jewish	1 in 55	98%	1 in 2,701	1 in 594,220
MPV17	Hepatocerebral mitochondrial DNA depletion syndrome, MPV17-related	AR	General	<1 in 500	96%	1 in 12,476	<1 in 10 million
MPV17	Hepatocerebral mitochondrial DNA depletion syndrome,MPV17-related	AR	Native American	1 in 20	96%	1 in 476	1 in 38,080
MTM1	Myotubular myopathy, X-linked	XL	General	1 in 25,000	98%	1 in 1,249,951	1 in 4,999,904
MTMR2	Charcot-Marie-Tooth disease, type 4B1	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
MTRR	Homocystinuria-megaloblastic anaemia, cobalamin E type	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
MTTP	Abetalipoproteinaemia	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
MTTP	Abetalipoproteinaemia	AR	Ashkenazi Jewish	1 in 180	98%	1 in 8,951	1 in 6,444,720
MUT	Methylmalonic acidaemia, MUT-related	AR	General	1 in 195	96%	1 in 4,851	1 in 3,783,780
MUT	Methylmalonic acidaemia, MUT-related	AR	East Asian	1 in 53	96%	1 in 1,301	1 in 275,812
MUT	Methylmalonic acidaemia, MUT-related	AR	Middle-Eastern	1 in 52	96%	1 in 1,276	1 in 265,408
MVK	Hyperimmunoglobulinaemia D syndrome	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
MVK	Mevalonate kinase deficiency	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
MYO7A	Non-syndromic hearing loss, MYO7A-related	AR	General	1 in 206	98%	1 in 10,251	1 in 8,446,824
MYO7A	Non-syndromic hearing loss, MYO7A-related	AR	East Asian	1 in 62	98%	1 in 3,051	1 in 756,648
MYO7A	Usher syndrome, type 1B	AR	General	1 in 206	98%	1 in 10,251	1 in 8,446,824
MYO7A	Usher syndrome, type 1B	AR	East Asian	1 in 62	98%	1 in 3,051	1 in 756,648
NAGLU	Mucopolysaccharidosis, type 3B (Sanfilippo syndrome B)	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
NAGLU	Mucopolysaccharidosis, type 3B (Sanfilippo syndrome B)	AR	Caucasian/European	1 in 346	99%	1 in 34,501	<1 in 10 million
NAGLU	Mucopolysaccharidosis, type 3B (Sanfilippo syndrome B)	AR	East Asian	1 in 298	99%	1 in 29,701	<1 in 10 million
NAGS	N-acetylglutamate synthase deficiency	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
NBN	Nijmegen breakage syndrome	AR	General	1 in 158	99%	1 in 15,701	1 in 9,923,032
NDP	Norrie disease	XL	General	<1 in 50,000	98%	1 in 2,499,951	<1 in 10 million
NDRG1	Charcot-Marie-Tooth disease, type 4D	AR	General	1 in 22	98%	1 in 1,051	1 in 92,488
NDUFAF5	Mitochondrial complex 1 deficiency (Leigh syndrome), NDUFAF5-related	AR	General	1 in 447	98%	1 in 22,301	<1 in 10 million
NDUFAF5	Mitochondrial complex 1 deficiency (Leigh syndrome), NDUFAF5-related	AR	Ashkenazi Jewish	1 in 290	98%	1 in 14,451	<1 in 10 million
NDUFS6	Mitochondrial complex 1 deficiency (Leigh syndrome), NDUFS6-related	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
NEB	Nemaline myopathy	AR	General	1 in 112	98%	1 in 5,551	1 in 2,486,848
NEB	Nemaline myopathy	AR	Amish	1 in 11	98%	1 in 501	1 in 22,044
NEB	Nemaline myopathy	AR	Ashkenazi Jewish	1 in 108	98%	1 in 5,351	1 in 2,311,632
NEB	Nemaline myopathy	AR	Finnish	1 in 112	98%	1 in 5,551	1 in 2,486,848
NPC1	Niemann-Pick disease, type C1	AR	General	1 in 194	90%	1 in 1,931	1 in 1,498,456
NPC2	Niemann-Pick disease, type C2	AR	General	1 in 194	99%	1 in 19,301	<1 in 10 million
NPHP1	Joubert syndrome 4	AR	General	1 in 480	98%	1 in 23,951	<1 in 10 million
NPHP1	Joubert syndrome 4	AR	Finnish	1 in 124	98%	1 in 6,151	1 in 3,050,896
NPHP1	Senior-Løken syndrome 1	AR	General	1 in 480	98%	1 in 23,951	<1 in 10 million
NPHP1	Senior-Løken syndrome 1	AR	Finnish	1 in 124	98%	1 in 6,151	1 in 3,050,896
NPHP1	Nephronophthisis	AR	General	1 in 480	98%	1 in 23,951	<1 in 10 million
NPHP1	Nephronophthisis	AR	Finnish	1 in 124	98%	1 in 6,151	1 in 3,050,896
NPHS1	Congenital nephrotic syndrome, type 1	AR	General	1 in 289	98%	1 in 14,401	<1 in 10 million
NPHS1	Congenital nephrotic syndrome, type 1	AR	Finnish	1 in 50	98%	1 in 2,451	1 in 490,200
NPHS2	Congenital nephrotic syndrome, type 2	AR	General	1 in 289	98%	1 in 14,401	<1 in 10 million
NPHS2	Congenital nephrotic syndrome, type 2	AR	Finnish	1 in 50	98%	1 in 2,451	1 in 490,200
NR0B1	Congenital adrenal hypoplasia, X-linked	XL	General	1 in 6,250	99%	1 in 624,901	1 in 2,499,804
NR2E3	Enhanced S-cone syndrome	AR	General	1 in 209	98%	1 in 10,401	1 in 8,695,236
NR2E3	Retinitis pigmentosa 37	AR	General	1 in 209	98%	1 in 10,401	1 in 8,695,236
NTRK1	Congenital insensitivity to pain with anhidrosis	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
OAT	Gyrate atrophy of choroid and retina	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
OCRL	Dent disease 2	XL	General	1 in 250,000	95%	1 in 4,999,981	<1 in 10 million
OCRL	Lowe syndrome	XL	General	1 in 250,000	95%	1 in 4,999,981	<1 in 10 million
OPA3	Costeff syndrome	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
OPA3	Costeff syndrome	AR	Iraqi Jewish	1 in 50	98%	1 in 2,451	1 in 490,200
OPHN1	X-linked intellectual disability with cerebellar hypoplasia and distinctive facial appearance	XL	General	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
OTC	Ornithine transcarbamylase deficiency	XL	General	1 in 7,000	90%	1 in 69,991	1 in 279,984
OTOF	Non-syndromic hearing loss, OTOF-related	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
OTOF	Non-syndromic hearing loss, OTOF-related	AR	Spanish	1 in 106	99%	1 in 10,501	1 in 4,452,424
P3H1	Osteogenesis imperfecta, type 8	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
P3H1	Osteogenesis imperfecta, type 8	AR	West African	1 in 67	99%	1 in 6,601	1 in 1,769,068
P3H1	Osteogenesis imperfecta, type 8	AR	African American	1 in 250	99%	1 in 24,901	<1 in 10,000,000

AR autosomal recessive; XL X-linked.
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Monash IVF

Expanded Carrier Screen

Gene list (by gene)

Gene	Disorder	Inheritance	Population	Carrier frequency	Detection rate	Residual risk of being a carrier*	Residual risk of having an affected child^
PAH	Phenylalanine hydroxylase deficiency (Phenylketonuria)	AR	General	1 in 93	99%	1 in 9,201	1 in 3,422,772
PAH	Phenylalanine hydroxylase deficiency (Phenylketonuria)	AR	Caucasian/European	1 in 63	99%	1 in 6,201	1 in 1,562,652
PAH	Phenylalanine hydroxylase deficiency (Phenylketonuria)	AR	Middle-Eastern	1 in 74	99%	1 in 7,301	1 in 2,161,096
PAH	Phenylalanine hydroxylase deficiency (Phenylketonuria)	AR	South East Asian	1 in 59	99%	1 in 5,801	1 in 1,369,036
PAK3	X-linked intellectual disability, PAK3-related	XL	General	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
PANK2	Pantothenate kinase-associated neurodegeneration	AR	General	1 in 289	99%	1 in 28,801	<1 in 10 million
PC	Pyruvate carboxylase deficiency	AR	General	1 in 250	95%	1 in 4,981	1 in 4,981,000
PCBD1	Tetrahydrobiopterin deficiency, PCBD1-related	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
PCCA	Propionic acidaemia, PCCA-related	AR	General	1 in 224	96%	1 in 5,576	1 in 4,996,096
PCCA	Propionic acidaemia, PCCA-related	AR	Native American	1 in 85	96%	1 in 2,101	1 in 714,340
PCCB	Propionic acidaemia, PCCB-related	AR	General	1 in 224	99%	1 in 22,301	<1 in 10 million
PCCB	Propionic acidaemia, PCCB-related	AR	Native American	1 in 85	99%	1 in 8,401	1 in 2,856,340
PCDH15	Non-syndromic hearing loss, PCDH15-related	AR	General	1 in 395	98%	1 in 19,701	1 in 78,804
PCDH15	Non-syndromic hearing loss, PCDH15-related	AR	Ashkenazi Jewish	1 in 72	98%	1 in 3,551	1 in 14,204
PCDH15	Usher syndrome, type 1F	AR	General	1 in 395	98%	1 in 19,701	1 in 78,804
PCDH15	Usher syndrome, type 1F	AR	Ashkenazi Jewish	1 in 72	98%	1 in 3,551	1 in 14,204
PDE6A	Retinitis pigmentosa, PDE6A-related	AR	General	1 in 133	99%	1 in 13,201	1 in 7,022,932
PDHA1	Pyruvate dehydrogenase E1-alpha deficiency	XL	General	<1 in 250,000	98%	1 in 12499951	<1 in 10 million
PDHB	Pyruvate dehydrogenase E1-beta deficiency	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
PEX1	Zellweger syndrome, PEX1-related	AR	General	1 in 147	95%	1 in 2,921	1 in 1,717,548
PEX10	Zellweger syndrome, PEX10-related	AR	General	1 in 500	95%	1 in 9,981	<1 in 10 million
PEX10	Zellweger syndrome, PEX10-related	AR	Japanese	1 in 354	95%	1 in 7,061	1 in 9,998,376
PEX12	Zellweger syndrome, PEX12-related	AR	General	1 in 373	95%	1 in 7,441	<1 in 10 million
PEX2	Zellweger syndrome, PEX2-related	AR	General	1 in 500	95%	1 in 9,981	<1 in 10 million
PEX2	Zellweger syndrome, PEX2-related	AR	Ashkenazi Jewish	1 in 123	95%	1 in 2,441	1 in 1,200,972
PEX6	Zellweger syndrome, PEX6-related	AR	General	1 in 280	95%	1 in 5,581	1 in 6,250,720
PEX7	Rhizomelic chondrodysplasia punctata, type 1	AR	General	1 in 158	99%	1 in 15,701	1 in 9,923,032
PFKM	Glycogen storage disease, type 7	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
PGK1	Phosphoglycerate kinase 1 deficiency	AR	General	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
PHF8	X-linked intellectual disability, Siderius type	XL	General	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
PHGDH	Phosphoglycerate dehydrogenase deficiency	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
PHGDH	Phosphoglycerate dehydrogenase deficiency	AR	Ashkenazi Jewish	1 in 280	98%	1 in 13,951	<1 in 10 million
PKHD1	Polycystic kidney disease, PKHD1-related	AR	General	1 in 70	98%	1 in 3,451	1 in 966,280
PKHD1	Polycystic kidney disease, PKHD1-related	AR	Ashkenazi Jewish	1 in 107	98%	1 in 5,301	1 in 2,268,828
PLA2G6	Infantile neuroaxonal dystrophy	AR	General	1 in 500	97%	1 in 16,634	<1 in 10 million
PLOD	Ehlers-Danlos syndrome with kyphoscoliosis, PLOD1-related	AR	General	1 in 159	99%	1 in 15,801	<1 in 10 million
PMM2	Congenital disorder of glycosylation, type 1A	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
PMM2	Congenital disorder of glycosylation, type 1A	AR	Ashkenazi Jewish	1 in 57	99%	1 in 5,601	1 in 1,277,028
PMM2	Congenital disorder of glycosylation, type 1A	AR	Caucasian/European	1 in 71	99%	1 in 7,001	1 in 1,988,284
POLG	Alpers-Huttenlocher syndrome	AR	General	1 in 113	95%	1 in 2,241	1 in 1,012,932
POLG	Ataxia neuropathy spectrum	AR	General	1 in 113	95%	1 in 2,241	1 in 1,012,932
POLG	Myocerebrohepatopathy syndrome	AR	General	1 in 113	95%	1 in 2,241	1 in 1,012,932
POLG	POLG-related disorders	AR	General	1 in 113	95%	1 in 2,241	1 in 1,012,932
POLG	Progressive external ophthalmoplegia	AR	General	1 in 113	95%	1 in 2,241	1 in 1,012,932
POLR1C	Treacher Collins syndrome, POLR1C-related	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
POLR1C	Hypomyelinating leucodystrophy, POLR1C-related	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
POMGNT1	Muscular dystrophy-dystroglycanopathy	AR	General	1 in 462	98%	1 in 23,051	<1 in 10 million
POMGNT1	Muscular dystrophy-dystroglycanopathy	AR	Finnish	1 in 111	98%	1 in 5,501	1 in 2,442,444
POMGNT1	Retinitis pigmentosa 76	AR	General	1 in 462	98%	1 in 23,051	<1 in 10 million
POMGNT1	Retinitis pigmentosa 76	AR	Finnish	1 in 111	98%	1 in 5,501	1 in 2,442,444
POMT1	Muscular dystrophy-dystroglycanopathy, POMT1-related	AR	General	1 in 290	99%	1 in 28,901	<1 in 10 million
POMT2	Muscular dystrophy-dystroglycanopathy, POMT2-related	AR	General	1 in 371	99%	1 in 37,001	<1 in 10 million
POU3F4	X-linked hearing loss, POU3F4-related	XL	General	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
PPT1	Neuronal ceroid lipofuscinosis, PPT1-related	AR	General	1 in 368	98%	1 in 18,351	<1 in 10 million
PPT1	Neuronal ceroid lipofuscinosis, PPT1-related	AR	Caucasian/European	1 in 488	98%	1 in 24,351	<1 in 10 million
PPT1	Neuronal ceroid lipofuscinosis, PPT1-related	AR	Finnish	1 in 75	98%	1 in 3,701	1 in 1,110,300
PQBP1	Renpenning syndrome	XL	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
PROP1	Combined pituitary hormone deficiency 2	AR	General	1 in 45	98%	1 in 2,201	1 in 396,180
PRPS1	Arts syndrome	XL	General	<1 in 250,000	98%	1 in 12,499,951	<1 in 10 million
PRPS1	Rosenberg-Chutorian syndrome	XL	General	<1 in 250,000	98%	1 in 12,499,951	<1 in 10 million
PRPS1	Phosphoribosylpyrophosphate synthetase superactivity	XL	General	<1 in 250,000	98%	1 in 12,499,951	<1 in 10 million
PRPS1	Non-syndromic hearing loss, PRPS1-related	XL	General	<1 in 250,000	98%	1 in 12,499,951	<1 in 10 million
PSAP	Metachromatic leucodystrophy due to saposin B deficiency	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
PTS	Tetrahydrobiopterin deficiency	AR	General	1 in 354	96%	1 in 8,826	<1 in 10 million
PUS1	Mitochondrial myopathy and sideroblastic anaemia 1	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
PYGM	Glycogen storage disease, type 5	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
PYGM	Glycogen storage disease, type 5	AR	Caucasian/European	1 in 206	99%	1 in 20,501	<1 in 10 million
QDPR	Tetrahydrobiopterin deficiency, QDPR-related	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
RAB23	Carpenter syndrome	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
RAG1	Omenn syndrome, RAG1-related	AR	General	1 in 137	98%	1 in 6,801	1 in 3,726,948

AR autosomal recessive; XL X-linked.
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Monash IVF

Expanded Carrier Screen

Gene list (by gene)

Gene	Disorder	Inheritance	Population	Carrier frequency	Detection rate	Residual risk of being a carrier*	Residual risk of having an affected child^
RAG2	Omenn syndrome, RAG2-related	AR	General	1 in 137	98%	1 in 6,801	1 in 3,726,948
RAPSN	Congenital myasthenic syndrome, RAPSN-related	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
RAPSN	Fetal akinesia deformation sequence	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
RARS2	Pontocerebellar hypoplasia, type 6	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
RAX	Microphthalmia, isolated 3	AR	General	1 in 289	99%	1 in 28,801	<1 in 10 million
RDH12	Leber congenital amaurosis, type 13	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
RDH12	Leber congenital amaurosis, type 13	AR	Caucasian/European	1 in 456	98%	1 in 22,751	<1 in 10 million
RMRP	Metaphyseal dysplasia without hypotrichosis	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
RMRP	Metaphyseal dysplasia without hypotrichosis	AR	Amish	1 in 16	99%	1 in 1,501	1 in 96,064
RMRP	Metaphyseal dysplasia without hypotrichosis	AR	Finnish	1 in 76	99%	1 in 7,501	1 in 2,280,304
RMRP	Anauxetic dysplasia	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
RMRP	Anauxetic dysplasia	AR	Amish	1 in 16	99%	1 in 1,501	1 in 96,064
RMRP	Anauxetic dysplasia	AR	Finnish	1 in 76	99%	1 in 7,501	1 in 2,280,304
RMRP	Cartilage-hair hypoplasia	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
RMRP	Cartilage-hair hypoplasia	AR	Amish	1 in 16	99%	1 in 1,501	1 in 96,064
RMRP	Cartilage-hair hypoplasia	AR	Finnish	1 in 76	99%	1 in 7,501	1 in 2,280,304
RP2	X-linked retinitis pigmentosa, RP2-related	XL	General	1 in 4,000	99%	1 in 399,901	1 in 1,600,000
RPE65	Leber congenital amaurosis 2	AR	General	1 in 228	98%	1 in 11,351	<1 in 10 million
RPE65	Retinitis pigmentosa 20	AR	General	1 in 228	98%	1 in 11,351	<1 in 10 million
RPGR	X-linked retinitis pigmentosa, RPGR-related	XL	General	1 in 3,000	75%	1 in 11,997	1 in 48,000
RPGRIP1L	Joubert syndrome 7	AR	General	1 in 259	98%	1 in 12,901	<1 in 10 million
RPGRIP1L	COACH syndrome	AR	General	1 in 259	98%	1 in 12,901	<1 in 10 million
RPGRIP1L	Meckel syndrome 5	AR	General	1 in 259	98%	1 in 12,901	<1 in 10 million
RS1	Juvenile retinoschisis, X-linked	XL	General	1 in 2,500	96%	1 in 62,476	1 in 249,956
RTEL1	Dyskeratosis congenita, type 5	AR	General	1 in 500	99%	1 in 49,901	<1 in 10 million
RTEL1	Dyskeratosis congenita, type 5	AR	Ashkenazi Jewish	1 in 203	99%	1 in 20,201	<1 in 10 million
SACS	Autosomal recessive spastic ataxia of Charlevoix-Saguenay	AR	General	<1 in 500	95%	1 in 9,981	<1 in 10 million
SACS	Autosomal recessive spastic ataxia of Charlevoix-Saguenay	AR	French Canadian	1 in 19	95%	1 in 361	1 in 27,436
SAMHD1	Aicardi–Goutières syndrome	AR	General	<1 in 500	95%	1 in 9,981	<1 in 10 million
SEPSECS	Pontocerebellar hypoplasia, type 2D	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
SERPINA1	Alpha-1 antitrypsin deficiency	AR	General	1 in 33	95%	1 in 641	1 in 84,612
SERPINA1	Alpha-1 antitrypsin deficiency	AR	Caucasian/European	1 in 19	95%	1 in 361	1 in 27,436
SGCA	Limb-girdle muscular dystrophy, type 2D	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
SGCA	Limb-girdle muscular dystrophy, type 2D	AR	Caucasian/European	1 in 288	98%	1 in 14,351	<1 in 10 million
SGCA	Limb-girdle muscular dystrophy, type 2D	AR	Finnish	1 in 150	98%	1 in 7,451	1 in 4,470,600
SGCB	Limb-girdle muscular dystrophy, type 2E	AR	General	1 in 500	98%	1 in 24,951	<1 in 10 million
SGCB	Limb-girdle muscular dystrophy, type 2E	AR	Caucasian/European	1 in 406	98%	1 in 20,251	<1 in 10 million
SGCD	Limb-girdle muscular dystrophy, type 2F	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
SGCG	Limb-girdle muscular dystrophy, type 2C	AR	General	1 in 381	98%	1 in 19,001	<1 in 10 million
SGCG	Limb-girdle muscular dystrophy, type 2C	AR	Moroccan	1 in 250	98%	1 in 12,451	<1 in 10 million
SGCG	Limb-girdle muscular dystrophy, type 2C	AR	Roma/Gypsy	1 in 96	98%	1 in 4,751	1 in 1,824,384
SGSH	Mucopolysaccharidosis, type 3A (Sanfilippo syndrome A)	AR	General	1 in 454	98%	1 in 22,651	<1 in 10 million
SGSH	Mucopolysaccharidosis, type 3A (Sanfilippo syndrome A)	AR	Caucasian/European	1 in 253	98%	1 in 12,601	<1 in 10 million
SH3TC2	Charcot-Marie-Tooth disease, SH3TC2-related	AR	General	1 in 69	99%	1 in 6,801	1 in 1,877,076
SLC12A3	Gitelman syndrome	AR	General	1 in 100	98%	1 in 4,951	1 in 1,980,400
SLC12A6	Andermann syndrome	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
SLC12A6	Andermann syndrome	AR	French Canadian	1 in 23	99%	1 in 2,201	1 in 202,492
SLC16A2	Allan-Herndon-Dudley syndrome	XL	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
SLC17A5	Sialic acid storage disorder	AR	General	<1 in 500	91%	1 in 5,545	<1 in 10 million
SLC17A5	Sialic acid storage disorder	AR	Finnish	1 in 100	91%	1 in 1,101	1 in 440,400
SLC22A5	Systemic primary carnitine deficiency	AR	General	1 in 129	76%	1 in 534	1 in 275,544
SLC22A5	Systemic primary carnitine deficiency	AR	African/African American	1 in 86	76%	1 in 355	1 in 122,120
SLC22A5	Systemic primary carnitine deficiency	AR	East Asian	1 in 77	76%	1 in 318	1 in 97,944
SLC22A5	Systemic primary carnitine deficiency	AR	Faroese	1 in 9	76%	1 in 34	1 in 1,224
SLC22A5	Systemic primary carnitine deficiency	AR	Pacific Islander	1 in 37	76%	1 in 151	1 in 22,348
SLC22A5	Systemic primary carnitine deficiency	AR	South Asian/Indian	1 in 51	76%	1 in 209	1 in 42,636
SLC25A13	Citrin deficiency	AR	General	<1 in 500	95%	1 in 9,981	<1 in 10 million
SLC25A13	Citrin deficiency	AR	East Asian	1 in 65	95%	1 in 1,281	1 in 333,060
SLC25A15	Hyperornithinaemia hyperammonaemia homocitrullinuria syndrome (Triple H syndrome)	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
SLC25A15	Hyperornithinaemia hyperammonaemia homocitrullinuria syndrome (Triple H syndrome)	AR	French Canadian	1 in 37	99%	1 in 3,601	1 in 532,948
SLC25A20	Carnitine-acylcarnitine translocase deficiency	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
SLC26A2	Achondrogenesis, type 1B	AR	General	1 in 158	90%	1 in 1,571	1 in 992,872
SLC26A2	Achondrogenesis, type 1B	AR	Finnish	1 in 50	90%	1 in 491	1 in 98,200
SLC26A2	Atelosteogenesis 2	AR	General	1 in 158	90%	1 in 1,571	1 in 992,872
SLC26A2	Atelosteogenesis 2	AR	Finnish	1 in 50	90%	1 in 491	1 in 98,200
SLC26A2	Diastrophic dysplasia	AR	General	1 in 158	90%	1 in 1,571	1 in 992,872
SLC26A2	Diastrophic dysplasia	AR	Finnish	1 in 50	90%	1 in 491	1 in 98,200
SLC26A2	Multiple epiphyseal dysplasia	AR	General	1 in 158	90%	1 in 1,571	1 in 992,872
SLC26A2	Multiple epiphyseal dysplasia	AR	Finnish	1 in 50	90%	1 in 491	1 in 98,200
SLC26A3	Congenital secretory chloride diarrhoea	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million

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Monash IVF

Expanded Carrier Screen

Gene list (by gene)

Gene	Disorder	Inheritance	Population	Carrier frequency	Detection rate	Residual risk of being a carrier*	Residual risk of having an affected child^
SLC26A3	Congenital secretory chloride diarrhoea	AR	Middle-Eastern	1 in 57	98%	1 in 2,801	1 in 638,628
SLC26A4	Pendred syndrome	AR	General	1 in 80	98%	1 in 3,951	1 in 1,264,320
SLC26A4	Pendred syndrome	AR	African/African American	1 in 76	98%	1 in 3,751	1 in 1,140,304
SLC26A4	Pendred syndrome	AR	Caucasian/European	1 in 88	98%	1 in 4,351	1 in 1,531,552
SLC26A4	Pendred syndrome	AR	East Asian	1 in 74	98%	1 in 3,651	1 in 1,080,696
SLC35A3	Arthrogryposis, intellectual disability and seizures	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
SLC35A3	Arthrogryposis, intellectual disability and seizures	AR	Ashkenazi Jewish	1 in 453	98%	1 in 22,601	<1 in 10 million
SLC37A4	Glycogen storage disease, type 1B	AR	General	1 in 158	95%	1 in 3,141	1 in 1,985,112
SLC37A4	Glycogen storage disease, type 1B	AR	Ashkenazi Jewish	1 in 71	95%	1 in 1,401	1 in 397,884
SLC39A4	Acrodermatitis enteropathica	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
SLC46A1	Hereditary folate malabsorption	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
SLC46A1	Hereditary folate malabsorption	AR	Puerto Rican	1 in 500	99%	1 in 49,901	<1 in 10 million
SLC4A11	Corneal endothelial dystrophy	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
SLC5A5	Thyroid dyshormonogenesis, SLC5A5-related	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
SLC6A19	Hartnup disorder	AR	General	1 in 87	99%	1 in 8,601	1 in 2,993,148
SLC6A8	Creatine deficiency syndrome	XL	General	1 in 3,434	98%	1 in 171,651	1 in 686,716
SLC7A7	Lysinuric protein intolerance	AR	General	<1 in 500	95%	1 in 9,981	<1 in 10 million
SLC7A7	Lysinuric protein intolerance	AR	Finnish	1 in 122	95%	1 in 2,421	1 in 1,181,448
SLC7A7	Lysinuric protein intolerance	AR	Japanese	1 in 119	95%	1 in 2,361	1 in 1,123,836
SMARCAL1	Schimke immunoosseous dysplasia	AR	General	1 in 500	90%	1 in 4,991	1 in 9,982,000
SMN1	Spinal muscular atrophy	AR	General	1 in 54	91%	1 in 590	1 in 127,440
SMN1	Spinal muscular atrophy	AR	African/African American	1 in 72	71%	1 in 246	1 in 70,848
SMN1	Spinal muscular atrophy	AR	Ashkenazi Jewish	1 in 67	91%	1 in 734	1 in 196,712
SMN1	Spinal muscular atrophy	AR	Caucasian/European	1 in 47	95%	1 in 921	1 in 173,148
SMN1	Spinal muscular atrophy	AR	East Asian	1 in 59	93%	1 in 830	1 in 195,880
SMN1	Spinal muscular atrophy	AR	Latino	1 in 68	90%	1 in 671	1 in 182,512
SMPD1	Niemann-Pick disease, type A/B	AR	General	1 in 250	95%	1 in 4,981	1 in 4,981,000
SMPD1	Niemann-Pick disease, type A/B	AR	Ashkenazi Jewish	1 in 115	95%	1 in 2,281	1 in 1,049,260
SMPD1	Niemann-Pick disease, type A/B	AR	Latino	1 in 106	95%	1 in 2,101	1 in 890,824
SPG11	SPG11-related neuromuscular disorders	AR	General	1 in 159	99%	1 in 15,801	<1 in 10 million
SPG7	Spastic paraplegia, type 7	AR	General	1 in 159	99%	1 in 15,801	<1 in 10 million
STAR	Lipoid congenital adrenal hyperplasia	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
SUMF1	Multiple sulphatase deficiency	AR	General	1 in 500	98%	1 in 24,951	<1 in 10 million
SUMF1	Multiple sulphatase deficiency	AR	Ashkenazi Jewish	1 in 320	98%	1 in 15,951	<1 in 10 million
SURF1	Charcot-Marie-Tooth disease, SURF1-related	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
SURF1	Leigh syndrome, SURF1-related	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
SYN1	X-linked epilepsy with variable learning disabilities	XL	General	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
TAT	Tyrosinaemia, type 2	AR	General	1 in 250	98%	1 in 12,451	<1 in 10 million
TCIRG1	Osteopetrosis, TCIRG1-related	AR	General	1 in 250	98%	1 in 12,451	<1 in 10 million
TECPR2	Spastic paraplegia, type 49	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
TFR2	Haemochromatosis, type 3	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
TG	Thyroid dyshormonogenesis, TG-related	AR	General	1 in 241	99%	1 in 24,001	<1 in 10 million
TGM1	Congenital ichthyosis	AR	General	1 in 224	95%	1 in 4,461	1 in 3,997,056
TH	Segawa syndrome	AR	General	1 in 224	98%	1 in 11,151	1 in 9,991,296
THOC2	X-linked intellectual disability, THOC2-related	XL	General	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
TMEM216	Joubert syndrome 2	AR	General	1 in 141	98%	1 in 7,001	1 in 3,948,564
TMEM216	Joubert syndrome 2	AR	Ashkenazi Jewish	1 in 92	98%	1 in 4,551	1 in 1,674,768
TMEM216	Meckel syndrome 2	AR	General	1 in 141	98%	1 in 7,001	1 in 3,948,564
TMEM216	Meckel syndrome 2	AR	Ashkenazi Jewish	1 in 92	98%	1 in 4,551	1 in 1,674,768
TPO	Thyroid dyshormonogenesis, TPO-related	AR	General	1 in 373	99%	1 in 37,201	<1 in 10 million
TPP1	Neuronal ceroid lipofuscinosis, TPP1-related	AR	General	1 in 252	97%	1 in 8,368	1 in 8,434,944
TPP1	Neuronal ceroid lipofuscinosis, TPP1-related	AR	French Canadian	1 in 53	97%	1 in 1,734	1 in 367,608
TRDN	Catecholaminergic polymorphic ventricular tachycardia, TRDN-related	AR	General	1 in 354	98%	1 in 17,651	<1 in 10 million
TRIM32	Limb-girdle muscular dystrophy, type 2H	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
TRIM32	Limb-girdle muscular dystrophy, type 2H	AR	Hutterite	1 in 12	98%	1 in 551	1 in 26,448
TRIM32	Bardet-Biedl syndrome 11	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
TRIM32	Bardet-Biedl syndrome 11	AR	Hutterite	1 in 12	98%	1 in 551	1 in 26,448
TRMU	Liver failure, acute infantile	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
TRMU	Liver failure, acute infantile	AR	Yemeni Jewish	1 in 34	98%	1 in 1,651	1 in 224,536
TSFM	Combined oxidative phosphorylation deficiency, TSFMrelated	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
TSFM	Combined oxidative phosphorylation deficiency, TSFM-related	AR	Finnish	1 in 80	98%	1 in 3,951	1 in 1,264,320
TSHB	Congenital hypothyroidism, TSHB-related	AR	General	1 in 500	99%	1 in 49,901	<1 in 10 million
TTC37	Trichohepatoenteric syndrome	AR	General	1 in 500	98%	1 in 24,951	<1 in 10 million
TTPA	Ataxia with isolated vitamin E deficiency	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
TTPA	Ataxia with isolated vitamin E deficiency	AR	Caucasian/European	1 in 267	90%	1 in 2,661	1 in 2,841,948
TYMP	Mitochondrial neurogastrointestinal encephalopathy (MNGIE) disease	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
UGT1A1	Crigler-Najjar syndrome	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
UPF3B	Lujan-Fryns syndrome, UPF3B-related	XL	General	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
USH1C	Non-syndromic hearing loss, USH1C-related	AR	General	1 in 353	90%	1 in 3,521	1 in 4,971,652
USH1C	Non-syndromic hearing loss, USH1C-related	AR	French Canadian	1 in 227	90%	1 in 2,261	1 in 2,052,988
USH1C	Usher syndrome, type 1C	AR	General	1 in 353	90%	1 in 3,521	1 in 4,971,652

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Monash IVF

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USH1C	Usher syndrome, type 1C	AR	French Canadian	1 in 227	90%	1 in 2,261	1 in 2,052,988
USH1G	Usher syndrome, type 1G	AR	General	1 in 434	99%	1 in 43,301	<1 in 10 million
USH2A	Usher syndrome, type 2A	AR	General	1 in 126	96%	1 in 3,126	1 in 1,575,504
USH2A	Usher syndrome, type 2A	AR	Caucasian/European	1 in 73	96%	1 in 1,801	1 in 525,892
VPS13A	Choreoacanthocytosis	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
VPS13B	Cohen syndrome	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
VPS45	Severe congenital neutropaenia, VPS45-related	AR	General	1 in 224	98%	1 in 11,151	1 in 9,991,296
VRK1	Pontocerebellar hypoplasia, type 1A	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
VSX2	Microphthalmia with or without coloboma	AR	General	1 in 91	98%	1 in 4,501	1 in 1,638,364
WAS	Wiskott-Aldrich syndrome	XL	General	1 in 125,000	99%	1 in 12,499,901	<1 in 10 million
WAS	Thrombocytopaenia, X-linked	XL	General	1 in 125,000	99%	1 in 12,499,901	<1 in 10 million
WAS	Severe congenital neutropaenia, WAS-related	XL	General	1 in 125,000	99%	1 in 12,499,901	<1 in 10 million
WHRN	Usher syndrome, type 2D	AR	General	1 in 282	99%	1 in 28,101	<1 in 10 million
WNT10A	Schopf-Schulz-Passarge syndrome	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
WNT10A	Odontoonychodermal dysplasia	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
XPA	Xeroderma pigmentosum, group A	AR	General	1 in 500	99%	1 in 49,901	<1 in 10 million
XPA	Xeroderma pigmentosum, group A	AR	Japanese	1 in 74	99%	1 in 7,301	1 in 2,161,096
XPC	Xeroderma pigmentosum, group C	AR	General	1 in 500	99%	1 in 49,901	<1 in 10 million
ZDHHC9	Lujan-Fryns syndrome, ZDHHC9-related	XL	General	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
ZFYVE26	Spastic paraplegia, type 15	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
ZNF711	X-linked intellectual disability, ZNF711-related	XL	General	<1 in 50,000	93%	1 in 714,272	1 in 2,857,143

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