MAC-ASD

Gene panel

Gene panel information

Gene panel	**MAC-ASD**
Version	4
Total genes	118
Activation date	Wednesday 04 december 2024
Publisher	Center for Medical Genetics, Ghent

Genes

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
ABCB6	99.29 %	605452	Dyschromatosis universalis hereditaria 3, 615402 (3), Autosomal dominant; [Blood group, Langereis system], 111600 (3); Pseudohyperkalemia, familial, 2, due to red cell leak, 609153 (3), Autosomal dominant; Microphthalmia/coloboma 7, 614497 (3), Autosomal dominant
ACTB	99.98 %	102630	Baraitser-Winter syndrome 1, 243310 (3), Autosomal dominant; Becker nevus, syndromic or isolated, somatic mosaic, 604919 (3); Thrombocytopenia 8, with dysmorphic features and developmental delay, 620475 (3), Autosomal dominant; Dystonia-deafness syndrome 1, 607371 (3), Autosomal dominant; Congenital smooth muscle hamartoma with or without hemihypertrophy, somatic mosaic, 620470 (3)
ACTG1	100 %	102560	Deafness, autosomal dominant 20/26, 604717 (3), Autosomal dominant; Baraitser-Winter syndrome 2, 614583 (3), Autosomal dominant
ADAMTS10	99.84 %	608990	Weill-Marchesani syndrome 1, recessive, 277600 (3), Autosomal recessive
ADAMTS17	99.86 %	607511	Weill-Marchesani 4 syndrome, recessive, 613195 (3), Autosomal recessive
ADAMTS18	99.86 %	607512	Microcornea, myopic chorioretinal atrophy, and telecanthus, 615458 (3), Autosomal recessive
ALDH1A3	99.57 %	600463	Microphthalmia, isolated 8, 615113 (3), Autosomal recessive
ALX1	98.39 %	601527	Frontonasal dysplasia 3, 613456 (3), Autosomal recessive
ANK3	99.75 %	600465	Intellectual developmental disorder, autosomal recessive 37, 615493 (3), Autosomal recessive
ARHGAP35	99.96 %	605277	*No OMIM phenotypes*
ASPH	99.83 %	600582	Traboulsi syndrome, 601552 (3), Autosomal recessive
ATOH7	99.55 %	609875	Persistent hyperplastic primary vitreous, autosomal recessive, 221900 (3), Autosomal recessive
B3GLCT	99.97 %	610308	Peters-plus syndrome, 261540 (3), Autosomal recessive
BCOR	99.92 %	300485	Microphthalmia, syndromic 2, 300166 (3), X-linked dominant
BMP4	99.99 %	112262	Orofacial cleft 11, 600625 (3), Autosomal dominant; Microphthalmia, syndromic 6, 607932 (3), Autosomal dominant
BMP7	99.94 %	112267	*No OMIM phenotypes*
BMPR1B	99.82 %	603248	Acromesomelic dysplasia 3, 609441 (3), Autosomal recessive; Brachydactyly, type A2, 112600 (3), Autosomal dominant; Brachydactyly, type A1, D, 616849 (3), Autosomal dominant
C12orf57	99.96 %	615140	Temtamy syndrome, 218340 (3), Autosomal recessive
CAPN15	99.95 %	603267	Oculogastrointestinal neurodevelopmental syndrome, 619318 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
CC2D2A	99.85 %	612013	COACH syndrome 2, 619111 (3), Autosomal recessive; Retinitis pigmentosa 93, 619845 (3), Autosomal recessive; Meckel syndrome 6, 612284 (3), Autosomal recessive; Joubert syndrome 9, 612285 (3), Autosomal recessive
CDH2	99.82 %	114020	Arrhythmogenic right ventricular dysplasia 14, 618920 (3), Autosomal dominant; ?Attention deficit-hyperactivity disorder 8, 619957 (3), Autosomal recessive; Agenesis of corpus callosum, cardiac, ocular, and genital syndrome, 618929 (3), Autosomal dominant
CDON	99.9 %	608707	Holoprosencephaly 11, 614226 (3), Autosomal dominant
CENPF	99.88 %	600236	Stromme syndrome, 243605 (3), Autosomal recessive
CHD7	99.9 %	608892	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 (3), Autosomal dominant; CHARGE syndrome, 214800 (3), Autosomal dominant
CHRDL1	99.67 %	300350	Megalocornea 1, X-linked, 309300 (3), X-linked recessive
CLDN19	98.74 %	610036	Hypomagnesemia 5, renal, with ocular involvement, 248190 (3), Autosomal recessive
COL4A1	99.93 %	120130	?Retinal arteries, tortuosity of, 180000 (3), Autosomal dominant; {Hemorrhage, intracerebral, susceptibility to}, 614519 (3); Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 (3), Autosomal dominant; Microangiopathy and leukoencephalopathy, pontine, autosomal dominant, 618564 (3), Autosomal dominant; Brain small vessel disease with or without ocular anomalies, 175780 (3), Autosomal dominant
COX7B	99.97 %	300885	Linear skin defects with multiple congenital anomalies 2, 300887 (3), X-linked dominant
CPAMD8	99.58 %	608841	Anterior segment dysgenesis 8, 617319 (3), Autosomal recessive
CREBBP	99.79 %	600140	Menke-Hennekam syndrome 1, 618332 (3), Autosomal dominant; Rubinstein-Taybi syndrome 1, 180849 (3), Autosomal dominant
CRIM1	99.87 %	606189	*No OMIM phenotypes*
CRYAA	17.8 %	123580	Cataract 9, multiple types, 604219 (3), Autosomal recessive, Autosomal dominant
CRYBA4	99.98 %	123631	Cataract 23, 610425 (3), Autosomal dominant
CYP1B1	99.86 %	601771	Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300 (3), Autosomal recessive; Anterior segment dysgenesis 6, multiple subtypes, 617315 (3), Autosomal recessive
DYRK1A	99.92 %	600855	Intellectual developmental disorder, autosomal dominant 7, 614104 (3), Autosomal dominant
ERCC1	99.38 %	126380	Cerebrooculofacioskeletal syndrome 4, 610758 (3), Autosomal recessive
ERCC2	99.66 %	126340	Xeroderma pigmentosum, group D, 278730 (3), Autosomal recessive; Trichothiodystrophy 1, photosensitive, 601675 (3), Autosomal recessive; ?Cerebrooculofacioskeletal syndrome 2, 610756 (3), Autosomal recessive
ERCC5	99.95 %	133530	Xeroderma pigmentosum, group G, 278780 (3), Autosomal recessive; Cerebrooculofacioskeletal syndrome 3, 616570 (3), Autosomal recessive; Xeroderma pigmentosum, group G/Cockayne syndrome, 278780 (3), Autosomal recessive
ERCC6	99.56 %	609413	UV-sensitive syndrome 1, 600630 (3), Autosomal recessive; Cerebrooculofacioskeletal syndrome 1, 214150 (3), Autosomal recessive; ?De Sanctis-Cacchione syndrome, 278800 (3), Autosomal recessive; Cockayne syndrome, type B, 133540 (3), Autosomal recessive; {Macular degeneration, age-related, susceptibility to, 5}, 613761 (3); Premature ovarian failure 11, 616946 (3), Autosomal dominant; {Lung cancer, susceptibility to}, 211980 (3), Somatic mutation, Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
EYA1	99.41 %	601653	Branchiootic syndrome 1, 602588 (3), Autosomal dominant; Branchiootorenal syndrome 1, with or without cataracts, 113650 (3), Autosomal dominant; Anterior segment anomalies with or without cataract, 602588 (3), Autosomal dominant; ?Otofaciocervical syndrome, 166780 (3), Autosomal dominant
FAT1	99.97 %	600976	*No OMIM phenotypes*
FOXC1	99.97 %	601090	Axenfeld-Rieger syndrome, type 3, 602482 (3), Autosomal dominant; Anterior segment dysgenesis 3, multiple subtypes, 601631 (3), Autosomal dominant
FOXE3	99.91 %	601094	Anterior segment dysgenesis 2, multiple subtypes, 610256 (3), Autosomal recessive; {Aortic aneurysm, familial thoracic 11, susceptibility to}, 617349 (3), Autosomal dominant; Cataract 34, multiple types, 612968 (3), Autosomal recessive
FOXL2	98.7 %	605597	Premature ovarian failure 3, 608996 (3), Autosomal dominant; Blepharophimosis, epicanthus inversus, and ptosis, types 1 and 2, 110100 (3), Autosomal recessive, Autosomal dominant
FRAS1	99.75 %	607830	Fraser syndrome 1, 219000 (3), Autosomal recessive
FREM1	99.89 %	608944	Manitoba oculotrichoanal syndrome, 248450 (3), Autosomal recessive; Bifid nose with or without anorectal and renal anomalies, 608980 (3), Autosomal recessive; Trigonocephaly 2, 614485 (3), Autosomal dominant
FREM2	99.94 %	608945	Fraser syndrome 2, 617666 (3), Autosomal recessive; Cryptophthalmos, unilateral or bilateral, isolated, 123570 (3), Autosomal recessive
FZD5	99.98 %	601723	Microphthalmia/coloboma 11, 620731 (3), Autosomal dominant
GDF3	99.99 %	606522	Klippel-Feil syndrome 3, autosomal dominant, 613702 (3); Microphthalmia, isolated, with coloboma 6, 613703 (3), Autosomal dominant; Microphthalmia, isolated 7, 613704 (3), Autosomal dominant
GDF6	99.99 %	601147	Microphthalmia with coloboma 6, digenic, 613703 (3), Autosomal dominant; Microphthalmia, isolated 4, 613094 (3); Leber congenital amaurosis 17, 615360 (3), Autosomal recessive; Multiple synostoses syndrome 4, 617898 (3), Autosomal dominant; Klippel-Feil syndrome 1, autosomal dominant, 118100 (3), Autosomal dominant
GJA1	100 %	121014	Erythrokeratodermia variabilis et progressiva 3, 617525 (3), Autosomal dominant; Craniometaphyseal dysplasia, autosomal recessive, 218400 (3), Autosomal recessive; Oculodentodigital dysplasia, 164200 (3), Autosomal dominant; Palmoplantar keratoderma with congenital alopecia, 104100 (3), Autosomal dominant; Syndactyly, type III, 186100 (3), Autosomal dominant; Oculodentodigital dysplasia, autosomal recessive, 257850 (3), Autosomal recessive
GRIP1	99.39 %	604597	Fraser syndrome 3, 617667 (3), Autosomal recessive
HCCS	99.85 %	300056	Linear skin defects with multiple congenital anomalies 1, 309801 (3), X-linked dominant
HESX1	99.81 %	601802	Pituitary hormone deficiency, combined, 5, 182230 (3), Autosomal recessive, Autosomal dominant; Septooptic dysplasia, 182230 (3), Autosomal recessive, Autosomal dominant; Growth hormone deficiency with pituitary anomalies, 182230 (3), Autosomal recessive, Autosomal dominant
HMX1	99.9 %	142992	Oculoauricular syndrome, 612109 (3), Autosomal recessive
IGBP1	99.89 %	300139	?Corpus callosum, agenesis of, with impaired intellectual development, ocular coloboma and micrognathia, 300472 (3), X-linked recessive
KDM6A	99.7 %	300128	Kabuki syndrome 2, 300867 (3), X-linked dominant
KIF26B	99.94 %	614026	*No OMIM phenotypes*
KMT2D	99.72 %	602113	Branchial arch abnormalities, choanal atresia, athelia, hearing loss, and hypothyroidism syndrome, 620186 (3), Autosomal dominant; Kabuki syndrome 1, 147920 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
LAMB2	99.88 %	150325	Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199 (3), Autosomal recessive; Pierson syndrome, 609049 (3), Autosomal recessive
LRP2	99.52 %	600073	Donnai-Barrow syndrome, 222448 (3), Autosomal recessive
LRP5	99.56 %	603506	Osteopetrosis, autosomal dominant 1, 607634 (3), Autosomal dominant; Polycystic liver disease 4 with or without kidney cysts, 617875 (3), Autosomal dominant; Endosteal hyperostosis, 144750 (3), Autosomal dominant; Osteoporosis-pseudoglioma syndrome, 259770 (3), Autosomal recessive; [Bone mineral density variability 1, high bone mass], 601884 (3), Autosomal dominant; Exudative vitreoretinopathy 4, 601813 (3), Autosomal recessive, Autosomal dominant
LTBP2	99.69 %	602091	Glaucoma 3, primary congenital, D, 613086 (3); Microspherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma, 251750 (3), Autosomal recessive; ?Weill-Marchesani syndrome 3, recessive, 614819 (3), Autosomal recessive
MAB21L1	100 %	601280	Cerebellar, ocular, craniofacial, and genital syndrome, 618479 (3), Autosomal recessive
MAB21L2	99.99 %	604357	Microphthalmia/coloboma and skeletal dysplasia syndrome, 615877 (3), Autosomal recessive, Autosomal dominant
MFRP	99.6 %	606227	Microphthalmia, isolated 5, 611040 (3), Autosomal recessive; Nanophthalmos 2, 609549 (3), Autosomal recessive
MIR204	99.89 %	610942	Retinal dystrophy and iris coloboma with or without cataract, 616722 (3), Autosomal dominant
MITF	99.9 %	156845	Waardenburg syndrome, type 2A, 193510 (3), Autosomal dominant; {Melanoma, cutaneous malignant, susceptibility to, 8}, 614456 (3); Tietz albinism-deafness syndrome, 103500 (3), Autosomal dominant; COMMAD syndrome, 617306 (3), Autosomal recessive
MYRF	99.59 %	608329	Nanophthalmos 1, 600165 (3), Autosomal dominant; Encephalitis/encephalopathy, mild, with reversible myelin vacuolization, 618113 (3), Autosomal dominant; Cardiac-urogenital syndrome, 618280 (3), Autosomal dominant
NAA10	99.94 %	300013	Microphthalmia, syndromic 1, 309800 (3), X-linked; Ogden syndrome, 300855 (3), X-linked dominant, X-linked recessive
NDP	99.93 %	300658	Exudative vitreoretinopathy 2, X-linked, 305390 (3), X-linked dominant, X-linked recessive; Norrie disease, 310600 (3), X-linked recessive
NHS	99.91 %	300457	Cataract 40, X-linked, 302200 (3), X-linked; Nance-Horan syndrome, 302350 (3), X-linked dominant
OCRL	99.78 %	300535	Dent disease 2, 300555 (3), X-linked recessive; Lowe syndrome, 309000 (3), X-linked recessive
OTX2	99.98 %	600037	Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125 (3), Autosomal dominant; Pituitary hormone deficiency, combined, 6, 613986 (3), Autosomal dominant; Microphthalmia, syndromic 5, 610125 (3), Autosomal dominant
PACS1	99.57 %	607492	Schuurs-Hoeijmakers syndrome, 615009 (3), Autosomal dominant
PAX2	99.8 %	167409	Glomerulosclerosis, focal segmental, 7, 616002 (3), Autosomal dominant; Papillorenal syndrome, 120330 (3), Autosomal dominant
PAX6	99.91 %	607108	Optic nerve hypoplasia, 165550 (3), Autosomal dominant; Cataract with late-onset corneal dystrophy, 106210 (3), Autosomal dominant; Microphthalmia/coloboma 12, 120200 (3), Autosomal dominant; ?Coloboma of optic nerve, 120430 (3), Autosomal dominant; Aniridia, 106210 (3), Autosomal dominant; Anterior segment dysgenesis 5, multiple subtypes, 604229 (3), Autosomal dominant; ?Morning glory disc anomaly, 120430 (3), Autosomal dominant; Foveal hypoplasia 1, 136520 (3), Autosomal dominant; Keratitis, 148190 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
PIGL	99.79 %	605947	CHIME syndrome, 280000 (3), Autosomal recessive
PITX2	99.85 %	601542	Ring dermoid of cornea, 180550 (3), Autosomal dominant; Axenfeld-Rieger syndrome, type 1, 180500 (3), Autosomal dominant; Anterior segment dysgenesis 4, 137600 (3), Autosomal dominant
PITX3	99.89 %	602669	Cataract 11, multiple types, 610623 (3), Autosomal recessive, Autosomal dominant; Anterior segment dysgenesis 1, multiple subtypes, 107250 (3), Autosomal dominant; Cataract 11, syndromic, autosomal recessive, 610623 (3), Autosomal recessive, Autosomal dominant
PLK4	99.71 %	605031	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171 (3), Autosomal recessive
PORCN	99.81 %	300651	Focal dermal hypoplasia, 305600 (3), X-linked dominant
PQBP1	99.91 %	300463	Renpenning syndrome, 309500 (3), X-linked recessive
PRR12	99.55 %	616633	Neuroocular syndrome, 619539 (3), Autosomal dominant
PRSS56	99.72 %	613858	Microphthalmia, isolated 6, 613517 (3), Autosomal recessive
PTCH1	99.87 %	601309	Basal cell nevus syndrome 1, 109400 (3), Autosomal dominant; Basal cell carcinoma, somatic, 605462 (3); Holoprosencephaly 7, 610828 (3), Autosomal dominant
PUF60	99.86 %	604819	Verheij syndrome, 615583 (3), Autosomal dominant
PXDN	99.9 %	605158	Anterior segment dysgenesis 7, with sclerocornea, 269400 (3), Autosomal recessive
RAB18	99.77 %	602207	Warburg micro syndrome 3, 614222 (3), Autosomal recessive
RAB3GAP1	99.26 %	602536	Martsolf syndrome 2, 619420 (3), Autosomal recessive; Warburg micro syndrome 1, 600118 (3), Autosomal recessive
RAB3GAP2	99.66 %	609275	Martsolf syndrome 1, 212720 (3), Autosomal recessive; Warburg micro syndrome 2, 614225 (3), Autosomal recessive
RARB	99.84 %	180220	Microphthalmia, syndromic 12, 615524 (3), Autosomal recessive, Autosomal dominant
RAX	99.27 %	601881	Microphthalmia, syndromic 16, 611038 (3), Autosomal recessive
RBP4	99.69 %	180250	Microphthalmia/coloboma 10, 616428 (3), Autosomal dominant; Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147 (3), Autosomal recessive
RERE	99.62 %	605226	Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart, 616975 (3), Autosomal dominant
RPGRIP1L	96.51 %	610937	Joubert syndrome 7, 611560 (3), Autosomal recessive; Meckel syndrome 5, 611561 (3), Autosomal recessive; ?COACH syndrome 3, 619113 (3), Autosomal recessive
SALL2	99.99 %	602219	?Coloboma, ocular, autosomal recessive, 216820 (3), Autosomal recessive
SALL4	99.99 %	607343	?IVIC syndrome, 147750 (3), Autosomal dominant; Duane-radial ray syndrome, 607323 (3), Autosomal dominant
SHH	99.93 %	600725	Single median maxillary central incisor, 147250 (3), Autosomal dominant; Holoprosencephaly 3, 142945 (3), Autosomal dominant; Microphthalmia/coloboma 5, 611638 (3), Autosomal dominant
SIX3	99.84 %	603714	Schizencephaly, 269160 (3); Holoprosencephaly 2, 157170 (3), Autosomal dominant
SIX6	99.99 %	606326	Optic disc anomalies with retinal and/or macular dystrophy, 212550 (3), Autosomal recessive
SLC38A8	99.83 %	615585	Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis, 609218 (3), Autosomal recessive
SMCHD1	99.84 %	614982	Facioscapulohumeral muscular dystrophy 2, digenic, 158901 (3), Digenic dominant; Bosma arhinia microphthalmia syndrome, 603457 (3), Autosomal dominant
SMOC1	99.87 %	608488	Microphthalmia with limb anomalies, 206920 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
SOX2	99.98 %	184429	Optic nerve hypoplasia and abnormalities of the central nervous system, 206900 (3), Autosomal dominant; Microphthalmia, syndromic 3, 206900 (3), Autosomal dominant
STRA6	99.82 %	610745	Microphthalmia, syndromic 9, 601186 (3), Autosomal recessive; Microphthalmia, isolated, with coloboma 8, 601186 (3), Autosomal recessive
TBC1D20	99.94 %	611663	Warburg micro syndrome 4, 615663 (3), Autosomal recessive
TENM3	99.93 %	610083	Microphthalmia, syndromic 15, 615145 (3), Autosomal recessive; ?Microphthalmia/coloboma 9, 615145 (3), Autosomal recessive
TFAP2A	99.92 %	107580	Branchiooculofacial syndrome, 113620 (3), Autosomal dominant
TMEM67	99.79 %	609884	Nephronophthisis 11, 613550 (3), Autosomal recessive; {Bardet-Biedl syndrome 14, modifier of}, 615991 (3), Autosomal recessive; Joubert syndrome 6, 610688 (3), Autosomal recessive; Meckel syndrome 3, 607361 (3), Autosomal recessive; ?RHYNS syndrome, 602152 (3), Autosomal recessive; COACH syndrome 1, 216360 (3), Autosomal recessive
TMEM98	99.92 %	615949	Nanophthalmos 4, 615972 (3), Autosomal dominant
VAX1	99.96 %	604294	?Microphthalmia, syndromic 11, 614402 (3), Autosomal recessive
VPS35L	98.94 %	618981	Ritscher-Schinzel syndrome 3, 619135 (3), Autosomal recessive
VSX1	99.82 %	605020	?Craniofacial anomalies and anterior segment dysgenesis syndrome, 614195 (3), Autosomal dominant; Keratoconus 1, 148300 (3), Autosomal dominant
VSX2	99.99 %	142993	Microphthalmia, isolated 2, 610093 (3), Autosomal recessive; Microphthalmia/coloboma 3, 610092 (3), Autosomal recessive
WDR37	99.56 %	618586	Neurooculocardiogenitourinary syndrome, 618652 (3), Autosomal dominant
YAP1	99.85 %	606608	Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or impaired intellectual development, 120433 (3), Autosomal dominant
ZIC2	99.97 %	603073	Holoprosencephaly 5, 609637 (3), Autosomal dominant

Explanation

OMIM release used for OMIM disease identifiers and descriptions: **2025-11-12**

Gene symbols used are according to the HGNC guidelines (corresponding to Ensembl database release 105).

Each Phenotype is followed by its MIM number, phenotype mapping key and inheritance pattern.

Possible phenotype mapping keys

(1) the disorder is placed on the map based on its association with a gene, but the underlying defect is not known

(2) the disorder has been placed on the map by linkage; no mutation has been found

(3) the molecular basis for the disorder is known; a mutation has been found in the gene

(4) a contiguous gene deletion or duplication syndrome, multiple genes are deleted or duplicated causing the phenotype

Brackets, "[ ]", indicate "nondiseases," mainly genetic variations that lead to apparently abnormal laboratory test values (e.g., dysalbuminemic euthyroidal hyperthyroxinemia).

Braces, "{ }", indicate mutations that contribute to susceptibility to multifactorial disorders (e.g., diabetes, asthma) or to susceptibility to infection (e.g., malaria).

A question mark, "?", before the phenotype name indicates that the relationship between the phenotype and gene is provisional. More details about this relationship are provided in the comment field of the map and in the gene and phenotype OMIM entries.

* The column '% at least 20 x covered' shows the percentage of the coding sequence (+/-20 nucleotides of the flanking introns) of that gene that is on average at least 20 x covered. This according to the experience with exome sequencing in our laboratory and based on the current method.