MAC-ASD panel

versie	16-Apr-2018 (59 genen)	Centrum voor Medische Genetica Gent

Gene	OMIM gene ID	Associated phenotype, OMIM phenotype ID, phenotype mapping key and inheritance pattern
ABCB6	605452	[Blood group, Langereis system], 111600 (3); Dyschromatosis universalis hereditaria 3, 615402 (3), Autosomal dominant; Microphthalmia, isolated, with coloboma 7, 614497 (3), Autosomal dominant; Pseudohyperkalemia, familial, 2, due to red cell leak, 609153 (3), Autosomal dominant
ADAMTS18	607512	Microcornea, myopic chorioretinal atrophy, and telecanthus, 615458 (3), Autosomal recessive
ALDH1A3	600463	Microphthalmia, isolated 8, 615113 (3), Autosomal recessive
ASPH	600582	Traboulsi syndrome, 601552 (3), Autosomal recessive
ATOH7	609875	Persistent hyperplastic primary vitreous, autosomal recessive, 221900 (3), Autosomal recessive
B3GLCT	610308	Peters-plus syndrome, 261540 (3), Autosomal recessive
BCOR	300485	Microphthalmia, syndromic 2, 300166 (3), X-linked dominant
BMP4	112262	Microphthalmia, syndromic 6, 607932 (3), Autosomal dominant; Orofacial cleft 11, 600625 (3)
BMP7	112267	No OMIM phenotype
C12orf57	615140	Temtamy syndrome, 218340 (3), Autosomal recessive
CHD7	608892	CHARGE syndrome, 214800 (3), Autosomal dominant; Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 (3), Autosomal dominant
COL4A1	120130	Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 (3), Autosomal dominant; Brain small vessel disease with or without ocular anomalies, 607595 (3), Autosomal dominant; {Hemorrhage, intracerebral, susceptibility to}, 614519 (3); Porencephaly 1, 175780 (3), Autosomal dominant; ?Retinal arteries, tortuosity of, 180000 (3), Autosomal dominant
COX7B	300885	Linear skin defects with multiple congenital anomalies 2, 300887 (3), X-linked dominant
CRYBA4	123631	Cataract 23, 610425 (3)
CYP1B1	601771	Anterior segment dysgenesis 6, multiple subtypes, 617315 (3); Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300 (3), Autosomal recessive

EYA1	601653	Anterior segment anomalies with or without cataract, 602588 (3), Autosomal dominant; Branchiootic syndrome 1, 602588 (3), Autosomal dominant; Branchiootorenal syndrome 1, with or without cataracts, 113650 (3), Autosomal dominant; ?Otofaciocervical syndrome, 166780 (3), Autosomal dominant
FKTN	607440	Cardiomyopathy, dilated, 1X, 611615 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588 (3), Autosomal recessive
FOXC1	601090	Anterior segment dysgenesis 3, multiple subtypes, 601631 (3), Autosomal dominant; Axenfeld-Rieger syndrome, type 3, 602482 (3), Autosomal dominant
FOXE3	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)
FOXL2	605597	Blepharophimosis, epicanthus inversus, and ptosis, type 1, 110100 (3), Autosomal dominant; Blepharophimosis, epicanthus inversus, and ptosis, type 2, 110100 (3), Autosomal dominant; Premature ovarian failure 3, 608996 (3), Autosomal dominant
GDF3	606522	Klippel-Feil syndrome 3, autosomal dominant, 613702 (3); Microphthalmia with coloboma 6, 613703 (3), Autosomal dominant; Microphthalmia, isolated 7, 613704 (3), Autosomal dominant
GDF6	601147	Klippel-Feil syndrome 1, autosomal dominant, 118100 (3), Autosomal dominant; Leber congenital amaurosis 17, 615360 (3), Autosomal recessive; Microphthalmia with coloboma 6, digenic, 613703 (3), Autosomal dominant; Microphthalmia, isolated 4, 613094 (3)
HCCS	300056	Linear skin defects with multiple congenital anomalies 1, 309801 (3), X-linked dominant
HESX1	601802	Growth hormone deficiency with pituitary anomalies, 182230 (3), Autosomal recessive, Autosomal dominant; Pituitary hormone deficiency, combined, 5, 182230 (3), Autosomal recessive, Autosomal dominant; Septooptic dysplasia, 182230 (3), Autosomal recessive, Autosomal dominant
IKBKG	300248	Ectodermal dysplasia, hypohidrotic, with immune deficiency, 300291 (3); Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency, 300301 (3); Immunodeficiency 33, 300636 (3), X-linked recessive; Immunodeficiency, isolated, 300584 (3); Incontinentia pigmenti, 308300 (3), X-linked dominant; Invasive pneumococcal disease, recurrent isolated, 2, 300640 (3)

LTBP2	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
MAB21L2	604357	Microphthalmia/coloboma and skeletal dysplasia syndrome, 615877 (3), Autosomal recessive, Autosomal dominant
MAF	177075	Ayme-Gripp syndrome, 601088 (3), Autosomal dominant; Cataract 21, multiple types, 610202 (3), Autosomal dominant
MFRP	606227	Microphthalmia, isolated 5, 611040 (3), Autosomal recessive; Nanophthalmos 2, 609549 (3)
NAA10	300013	?Microphthalmia, syndromic 1, 309800 (3), X-linked; Ogden syndrome, 300855 (3), X-linked recessive, X-linked dominant
NHS	300457	Cataract 40, X-linked, 302200 (3), X-linked; Nance-Horan syndrome, 302350 (3), X-linked dominant
OTX2	600037	Microphthalmia, syndromic 5, 610125 (3), Autosomal dominant; Pituitary hormone deficiency, combined, 6, 613986 (3), Autosomal dominant; Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125 (3), Autosomal dominant
PAX2	167409	Glomerulosclerosis, focal segmental, 7, 616002 (3), Autosomal dominant; Papillorenal syndrome, 120330 (3), Autosomal dominant
PAX6	607108	Aniridia, 106210 (3), Autosomal dominant; Anterior segment dysgenesis 5, multiple subtypes, 604229 (3); Cataract with late-onset corneal dystrophy, 106210 (3), Autosomal dominant; ?Coloboma of optic nerve, 120430 (3), Autosomal dominant; ?Coloboma, ocular, 120200 (3), Autosomal dominant; Foveal hypoplasia 1, 136520 (3), Autosomal dominant; Keratitis, 148190 (3), Autosomal dominant; ?Morning glory disc anomaly, 120430 (3), Autosomal dominant; Optic nerve hypoplasia, 165550 (3), Autosomal dominant
PITX2	601542	Anterior segment dysgenesis 4, 137600 (3), Autosomal dominant; Axenfeld-Rieger syndrome, type 1, 180500 (3), Autosomal dominant; Ring dermoid of cornea, 180550 (3), Autosomal dominant
PITX3	602669	Anterior segment dysgenesis 1, multiple subtypes, 107250 (3), Autosomal dominant; Cataract 11, multiple types, 610623 (3), Autosomal dominant; Cataract 11, syndromic, 610623 (3), Autosomal dominant
POMGNT1	606822	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, 613151 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157 (3), Autosomal recessive; Retinitis pigmentosa 76, 617123 (3), Autosomal recessive

POMT1	607423	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308 (3), Autosomal recessive
PORCN	300651	Focal dermal hypoplasia, 305600 (3), X-linked dominant
PRSS56	613858	Microphthalmia, isolated 6, 613517 (3), Autosomal recessive
PXDN	605158	Anterior segment dysgenesis 7, with sclerocornea, 269400 (3), Autosomal recessive
RAB3GAP1	602536	Warburg micro syndrome 1, 600118 (3), Autosomal recessive
RAB3GAP2	609275	Martsolf syndrome, 212720 (3), Autosomal recessive; Warburg micro syndrome 2, 614225 (3), Autosomal recessive
RARB	180220	Microphthalmia, syndromic 12, 615524 (3), Autosomal recessive, Autosomal dominant
RAX	601881	Microphthalmia, isolated 3, 611038 (3), Autosomal recessive
RBP4	180250	Microphthalmia, isolated, with coloboma 10, 616428 (3), Autosomal dominant; Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147 (3), Autosomal recessive
SALL2	602219	?Coloboma, ocular, autosomal recessive, 216820 (3), Autosomal recessive
SHH	600725	Holoprosencephaly 3, 142945 (3), Autosomal dominant; Microphthalmia with coloboma 5, 611638 (3), Autosomal dominant; Schizencephaly, 269160 (3); Single median maxillary central incisor, 147250 (3), Autosomal dominant
SIX6	606326	Optic disc anomalies with retinal and/or macular dystrophy, 212550 (3), Autosomal recessive
SLC38A8	615585	Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis, 609218 (3), Autosomal recessive
SMOC1	608488	Microphthalmia with limb anomalies, 206920 (3), Autosomal recessive
SOX2	184429	Microphthalmia, syndromic 3, 206900 (3), Autosomal dominant; Optic nerve hypoplasia and abnormalities of the central nervous system, 206900 (3), Autosomal dominant
STRA6	610745	Microphthalmia, isolated, with coloboma 8, 601186 (3), Autosomal recessive; Microphthalmia, syndromic 9, 601186 (3), Autosomal recessive
TENM3	610083	Microphthalmia, isolated, with coloboma 9, 615145 (3), Autosomal recessive
TFAP2A	107580	Branchiooculofacial syndrome, 113620 (3), Autosomal dominant
VAX1	604294	?Microphthalmia, syndromic 11, 614402 (3), Autosomal recessive
VSX1	605020	?Craniofacial anomalies and anterior segment dysgenesis syndrome, 614195 (3); Keratoconus 1, 148300 (3), Autosomal dominant

VSX2	142993	Microphthalmia with coloboma 3, 610092 (3); Microphthalmia, isolated 2, 610093 (3)
YAP1	606608	Coloboma, ocular, 120433 (3), Autosomal dominant; Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation, 120433 (3), Autosomal dominant

Gene symbols used are according to the HGNC guidelines. For some genes a previously HGNC-approved symbol is in brackets.
Each Phenotype is followed by its MIM number, phenotype mapping key and inheritance pattern.
OMIM release used for OMIM disease identifiers and descriptions: June 06, 2017

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.