

<b>MAC-ASD panel (v2)</b>
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<b>versie</b>	2019-09-03 (79 genen)	Centrum voor Medische Genetica Gent
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Gene	OMIM gene ID	Associated phenotype, OMIM phenotype ID, phenotype mapping key and inheritance pattern
<i>ABCB6</i>	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
<i>ADAMTS18</i>	607512	Microcornea, myopic chorioretinal atrophy, and telecanthus, 615458 (3), Autosomal recessive
<i>ALDH1A3</i>	600463	Microphthalmia, isolated 8, 615113 (3), Autosomal recessive
<i>ASPH</i>	600582	Traboulsi syndrome, 601552 (3), Autosomal recessive
<i>ATOH7</i>	609875	Persistent hyperplastic primary vitreous, autosomal recessive, 221900 (3), Autosomal recessive
<i>B3GLCT</i>	610308	Peters-plus syndrome, 261540 (3), Autosomal recessive
<i>BCOR</i>	300485	Microphthalmia, syndromic 2, 300166 (3), X-linked dominant
<i>BMP4</i>	112262	Microphthalmia, syndromic 6, 607932 (3), Autosomal dominant; Orofacial cleft 11, 600625 (3)
<i>BMP7</i>	112267	No OMIM phenotype
<i>C12orf57</i>	615140	Temtamy syndrome, 218340 (3), Autosomal recessive
<i>CC2D2A</i>	612013	COACH syndrome, 216360 (3), Autosomal recessive; Joubert syndrome 9, 612285 (3), Autosomal recessive; Meckel syndrome 6, 612284 (3), Autosomal recessive
<i>CHD7</i>	608892	CHARGE syndrome, 214800 (3), Autosomal dominant; Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 (3), Autosomal dominant
<i>CHRDL1</i>	300350	Megalocornea 1, X-linked, 309300 (3), X-linked recessive

<i>COL4A1</i>	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; Schizencephaly, 269160 (3)
<i>COX7B</i>	300885	Linear skin defects with multiple congenital anomalies 2, 300887 (3), X-linked dominant
<i>CRIM1</i>	606189	No OMIM phenotype
<i>CRYBA4</i>	123631	Cataract 23, 610425 (3)
<i>CYP1B1</i>	601771	Anterior segment dysgenesis 6, multiple subtypes, 617315 (3); Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300 (3), Autosomal recessive
<i>ERCC1</i>	126380	Cerebrooculofacioskeletal syndrome 4, 610758 (3), Autosomal recessive
<i>ERCC2</i>	126340	?Cerebrooculofacioskeletal syndrome 2, 610756 (3), Autosomal recessive; Trichothiodystrophy 1, photosensitive, 601675 (3), Autosomal recessive; Xeroderma pigmentosum, group D, 278730 (3), Autosomal recessive
<i>ERCC5</i>	133530	Cerebrooculofacioskeletal syndrome 3, 616570 (3), Autosomal recessive; Xeroderma pigmentosum, group G, 278780 (3), Autosomal recessive; Xeroderma pigmentosum, group G/Cockayne syndrome, 278780 (3), Autosomal recessive
<i>ERCC6</i>	609413	Cerebrooculofacioskeletal syndrome 1, 214150 (3), Autosomal recessive; Cockayne syndrome, type B, 133540 (3), Autosomal recessive; De Sanctis-Cacchione syndrome, 278800 (3), Autosomal recessive; {Lung cancer, susceptibility to}, 211980 (3), Autosomal recessive; {Macular degeneration, age-related, susceptibility to, 5}, 613761 (3); Premature ovarian failure 11, 616946 (3), Autosomal dominant; UV-sensitive syndrome 1, 600630 (3), Autosomal recessive
<i>EYA1</i>	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
<i>FOXC1</i>	601090	Anterior segment dysgenesis 3, multiple subtypes, 601631 (3), Autosomal dominant; Axenfeld-Rieger syndrome, type 3, 602482 (3), Autosomal dominant
<i>FOXE3</i>	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)
<i>FOXL2</i>	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
<i>FRAS1</i>	607830	Fraser syndrome 1, 219000 (3), Autosomal recessive
<i>FREM1</i>	608944	Bifid nose with or without anorectal and renal anomalies, 608980 (3); Manitoba oculotrichoanal syndrome, 248450 (3), Autosomal recessive; Trigenocephaly 2, 614485 (3), Autosomal dominant
<i>FREM2</i>	608945	Fraser syndrome 2, 617666 (3), Autosomal recessive
<i>GDF3</i>	606522	Klippel-Feil syndrome 3, autosomal dominant, 613702 (3); Microphthalmia with coloboma 6, 613703 (3), Autosomal dominant; Microphthalmia, isolated 7, 613704 (3), Autosomal dominant
<i>GDF6</i>	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); Multiple synostoses syndrome 4, 617898 (3), Autosomal dominant

<i>GJA1</i>	121014	Atrioventricular septal defect 3, 600309 (3), Autosomal dominant; Craniometaphyseal dysplasia, autosomal recessive, 218400 (3), Autosomal recessive; Erythrokeratoderma variabilis et progressiva 3, 617525 (3), Autosomal dominant; Hypoplastic left heart syndrome 1, 241550 (3), Autosomal recessive; Oculodentodigital dysplasia, 164200 (3), Autosomal dominant; Oculodentodigital dysplasia, autosomal recessive, 257850 (3), Autosomal recessive; Palmoplantar keratoderma with congenital alopecia, 104100 (3), Autosomal dominant; Syndactyly, type III, 186100 (3), Autosomal dominant
<i>GRIP1</i>	604597	Fraser syndrome 3, 617667 (3), Autosomal recessive
<i>HCCS</i>	300056	Linear skin defects with multiple congenital anomalies 1, 309801 (3), X-linked dominant
<i>HESX1</i>	601802	Growth hormone deficiency with pituitary anomalies, 182230 (3), Autosomal recessive, Autosomal dominant; Pituitary hormone deficiency, combined, 5, 182230 (3), Autosomal recessive, Autosomal dominant; Septo-optic dysplasia, 182230 (3), Autosomal recessive, Autosomal dominant
<i>HMGB3</i>	300193	?Microphthalmia, syndromic 13, 300915 (3), X-linked
<i>HMX1</i>	142992	Oculoauricular syndrome, 612109 (3), Autosomal recessive
<i>IGBP1</i>	300139	Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia, 300472 (3), X-linked recessive
<i>LTBP2</i>	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
<i>MAB21L2</i>	604357	Microphthalmia/coloboma and skeletal dysplasia syndrome, 615877 (3), Autosomal recessive, Autosomal dominant
<i>MFRP</i>	606227	Microphthalmia, isolated 5, 611040 (3), Autosomal recessive; Nanophthalmos 2, 609549 (3)
<i>NAA10</i>	300013	?Microphthalmia, syndromic 1, 309800 (3), X-linked; Ogden syndrome, 300855 (3), X-linked recessive, X-linked dominant

<i>NHS</i>	300457	Cataract 40, X-linked, 302200 (3), X-linked; Nance-Horan syndrome, 302350 (3), X-linked dominant
<i>OTX2</i>	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
<i>PAX2</i>	167409	Glomerulosclerosis, focal segmental, 7, 616002 (3), Autosomal dominant; Papillorenal syndrome, 120330 (3), Autosomal dominant
<i>PAX6</i>	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
<i>PIGL</i>	605947	CHIME syndrome, 280000 (3), Autosomal recessive
<i>PITX2</i>	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
<i>PITX3</i>	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
<i>PORCN</i>	300651	Focal dermal hypoplasia, 305600 (3), X-linked dominant
<i>PQBP1</i>	300463	Renpenning syndrome, 309500 (3), X-linked recessive
<i>PRSS56</i>	613858	Microphthalmia, isolated 6, 613517 (3), Autosomal recessive
<i>PTCH1</i>	601309	Basal cell carcinoma, somatic, 605462 (3); Basal cell nevus syndrome, 109400 (3), Autosomal dominant; Holoprosencephaly 7, 610828 (3), Autosomal dominant
<i>PXDN</i>	605158	Anterior segment dysgenesis 7, with sclerocornea, 269400 (3), Autosomal recessive

<i>RAB3GAP1</i>	602536	Warburg micro syndrome 1, 600118 (3), Autosomal recessive
<i>RAB3GAP2</i>	609275	Martsof syndrome, 212720 (3), Autosomal recessive; Warburg micro syndrome 2, 614225 (3), Autosomal recessive
<i>RARB</i>	180220	Microphthalmia, syndromic 12, 615524 (3), Autosomal recessive, Autosomal dominant
<i>RAX</i>	601881	Microphthalmia, isolated 3, 611038 (3), Autosomal recessive
<i>RBP4</i>	180250	Microphthalmia, isolated, with coloboma 10, 616428 (3), Autosomal dominant; Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147 (3), Autosomal recessive
<i>RPGRIP1L</i>	610937	COACH syndrome, 216360 (3), Autosomal recessive; Joubert syndrome 7, 611560 (3), Autosomal recessive; Meckel syndrome 5, 611561 (3), Autosomal recessive
<i>SALL2</i>	602219	?Coloboma, ocular, autosomal recessive, 216820 (3), Autosomal recessive
<i>SHH</i>	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
<i>SIX3</i>	603714	Holoprosencephaly 2, 157170 (3), Autosomal dominant; Schizencephaly, 269160 (3)
<i>SIX6</i>	606326	Optic disc anomalies with retinal and/or macular dystrophy, 212550 (3), Autosomal recessive
<i>SLC38A8</i>	615585	Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis, 609218 (3), Autosomal recessive
<i>SMOC1</i>	608488	Microphthalmia with limb anomalies, 206920 (3), Autosomal recessive
<i>SOX2</i>	184429	Microphthalmia, syndromic 3, 206900 (3), Autosomal dominant; Optic nerve hypoplasia and abnormalities of the central nervous system, 206900 (3), Autosomal dominant
<i>STRA6</i>	610745	Microphthalmia, isolated, with coloboma 8, 601186 (3), Autosomal recessive; Microphthalmia, syndromic 9, 601186 (3), Autosomal recessive
<i>TBC1D20</i>	611663	Warburg micro syndrome 4, 615663 (3), Autosomal recessive
<i>TENM3</i>	610083	Microphthalmia, isolated, with coloboma 9, 615145 (3), Autosomal recessive

<i>TFAP2A</i>	107580	Branchiooculofacial syndrome, 113620 (3), Autosomal dominant
<i>TMEM67</i>	609884	{Bardet-Biedl syndrome 14, modifier of}, 615991 (3), Autosomal recessive; COACH syndrome, 216360 (3), Autosomal recessive; Joubert syndrome 6, 610688 (3), Autosomal recessive; Meckel syndrome 3, 607361 (3), Autosomal recessive; Nephronophthisis 11, 613550 (3), Autosomal recessive
<i>TMEM98</i>	615949	Nanophthalmos 4, 615972 (3), Autosomal dominant
<i>VAX1</i>	604294	?Microphthalmia, syndromic 11, 614402 (3), Autosomal recessive
<i>VSX1</i>	605020	?Craniofacial anomalies and anterior segment dysgenesis syndrome, 614195 (3); Keratoconus 1, 148300 (3), Autosomal dominant
<i>VSX2</i>	142993	Microphthalmia with coloboma 3, 610092 (3); Microphthalmia, isolated 2, 610093 (3)
<i>WDR37</i>	No OMIM gene	No OMIM phenotype
<i>YAP1</i>	606608	Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation, 120433 (3), Autosomal dominant
<i>ZIC2</i>	603073	Holoprosencephaly 5, 609637 (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: July 04, 2018

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.

