

OCA-OA-Isolated nystagmus panel		
versie	v2 (30 genen)	Centrum voor Medische Genetica Gent
Gene	OMIM gene ID	Associated phenotype, OMIM phenotype ID, phenotype mapping key and inheritance pattern
<i>AHR</i>	600253	?Retinitis pigmentosa 85, 618345 (3), Autosomal recessive
<i>AP3B1</i>	603401	Hermansky-Pudlak syndrome 2, 608233 (3), Autosomal recessive
<i>AP3D1</i>	607246	?Hermansky-Pudlak syndrome 10, 617050 (3), Autosomal recessive
<i>BLOC1S3</i>	609762	Hermansky-Pudlak syndrome 8, 614077 (3), Autosomal recessive
<i>BLOC1S5</i>	607289	Hermansky-Pudlak syndrome 11, 619172 (3), Autosomal recessive
<i>BLOC1S6</i>	604310	?Hermansky-Pudlak syndrome 9, 614171 (3), Autosomal recessive
<i>DCT</i>	191275	Oculocutaneous albinism, type VIII, 619165 (3), Autosomal recessive
<i>DTNBP1</i>	607145	Hermansky-Pudlak syndrome 7, 614076 (3), Autosomal recessive
<i>EPG5</i>	615068	Vici syndrome, 242840 (3), Autosomal recessive
<i>FRMD7</i>	300628	Nystagmus, infantile periodic alternating, X-linked, 310700 (3), X-linked; Nystagmus 1, congenital, X-linked, 310700 (3), X-linked
<i>GPR143</i>	300808	Ocular albinism, type I, Nettleship-Falls type, 300500 (3), X-linked; Nystagmus 6, congenital, X-linked, 300814 (3), X-linked recessive
<i>HPS1</i>	604982	Hermansky-Pudlak syndrome 1, 203300 (3), Autosomal recessive
<i>HPS3</i>	606118	Hermansky-Pudlak syndrome 3, 614072 (3), Autosomal recessive
<i>HPS4</i>	606682	Hermansky-Pudlak syndrome 4, 614073 (3), Autosomal recessive
<i>HPS5</i>	607521	Hermansky-Pudlak syndrome 5, 614074 (3), Autosomal recessive
<i>HPS6</i>	607522	Hermansky-Pudlak syndrome 6, 614075 (3), Autosomal recessive
<i>LRMDA</i>	614537	Albinism, oculocutaneous, type VII, 615179 (3), Autosomal recessive
<i>LYST</i>	606897	Chediak-Higashi syndrome, 214500 (3), Autosomal recessive
<i>MC1R</i>	155555	[Analgesia from kappa-opioid receptor agonist, female-specific], 613098 (3); [Skin/hair/eye pigmentation 2, red hair/fair skin], 266300 (3), Autosomal recessive; [Skin/hair/eye pigmentation 2, blond hair/fair skin], 266300 (3), Autosomal recessive; {Melanoma, cutaneous malignant, 5}, 613099 (3); {Albinism, oculocutaneous, type II, modifier of}, 203200 (3), Autosomal recessive; {UV-induced skin damage}, 266300 (3), Autosomal recessive
<i>MITF</i>	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; Waardenburg syndrome/ocular albinism, digenic, 103470 (3); COMMAD syndrome, 617306 (3), Autosomal recessive
<i>MLPH</i>	606526	Griscelli syndrome, type 3, 609227 (3), Autosomal recessive
<i>MYO5A</i>	160777	Griscelli syndrome, type 1, 214450 (3), Autosomal recessive

<i>OCA2</i>	611409	[Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220 (3), Autosomal recessive; [Skin/hair/eye pigmentation 1, blond/brown hair], 227220 (3), Autosomal recessive; Albinism, brown oculocutaneous, 203200 (3), Autosomal recessive; Albinism, oculocutaneous, type II, 203200 (3), Autosomal recessive
<i>PAX6</i>	607108	Optic nerve hypoplasia, 165550 (3), Autosomal dominant; Cataract with late-onset corneal dystrophy, 106210 (3), Autosomal dominant; ?Coloboma, ocular, 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
<i>RAB27A</i>	603868	Griscelli syndrome, type 2, 607624 (3), Autosomal recessive
<i>SLC24A5</i>	609802	[Skin/hair/eye pigmentation 4, fair/dark skin], 113750 (3), Autosomal recessive; Albinism, oculocutaneous, type VI, 113750 (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>SLC45A2</i>	606202	[Skin/hair/eye pigmentation 5, dark/light eyes], 227240 (3), Autosomal recessive; [Skin/hair/eye pigmentation 5, black/nonblack hair], 227240 (3), Autosomal recessive; Albinism, oculocutaneous, type IV, 606574 (3), Autosomal recessive; [Skin/hair/eye pigmentation 5, dark/fair skin], 227240 (3), Autosomal recessive
<i>TYR</i>	606933	[Skin/hair/eye pigmentation 3, light/dark/freckling skin], 601800 (3), Autosomal dominant; [Skin/hair/eye pigmentation 3, blue/green eyes], 601800 (3), Autosomal dominant; {Melanoma, cutaneous malignant, susceptibility to, 8}, 601800 (3), Autosomal dominant; Albinism, oculocutaneous, type IB, 606952 (3), Autosomal recessive; Waardenburg syndrome/albinism, digenic, 103470 (3); Albinism, oculocutaneous, type IA, 203100 (3), Autosomal recessive
<i>TYRP1</i>	115501	[Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)], 612271 (3); Albinism, oculocutaneous, type III, 203290 (3), Autosomal recessive

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 26, 2021

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.