OCA-OA-Isolated nystagmus

Gene panel

Gene panel information

Gene panel	**OCA-OA-Isolated nystagmus**
Version	3
Total genes	31
Activation date	Monday 02 september 2024
Publisher	Center for Medical Genetics, Ghent

Genes

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
AHR	99.79 %	600253	?Retinitis pigmentosa 85, 618345 (3), Autosomal recessive
AP3B1	99.89 %	603401	Hermansky-Pudlak syndrome 2, 608233 (3), Autosomal recessive
AP3D1	100 %	607246	?Hermansky-Pudlak syndrome 10, 617050 (3), Autosomal recessive
BLOC1S3	100 %	609762	Hermansky-Pudlak syndrome 8, 614077 (3), Autosomal recessive
BLOC1S5	99.8 %	607289	Hermansky-Pudlak syndrome 11, 619172 (3), Autosomal recessive
BLOC1S6	99.98 %	604310	?Hermansky-Pudlak syndrome 9, 614171 (3), Autosomal recessive
CACNA1A	98.16 %	601011	Spinocerebellar ataxia 6, 183086 (3), Autosomal dominant; Episodic ataxia, type 2, 108500 (3), Autosomal dominant; Developmental and epileptic encephalopathy 42, 617106 (3), Autosomal dominant; Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500 (3), Autosomal dominant; Migraine, familial hemiplegic, 1, 141500 (3), Autosomal dominant
CACNA1F	99.94 %	300110	Cone-rod dystrophy, X-linked, 3, 300476 (3), X-linked recessive; Night blindness, congenital stationary (incomplete), 2A, X-linked, 300071 (3), X-linked; Aland Island eye disease, 300600 (3), X-linked
DAGLA	99.93 %	614015	*No OMIM phenotypes*
DCT	99.98 %	191275	Oculocutaneous albinism, type VIII, 619165 (3), Autosomal recessive
DTNBP1	99.89 %	607145	Hermansky-Pudlak syndrome 7, 614076 (3), Autosomal recessive
EPG5	99.95 %	615068	Vici syndrome, 242840 (3), Autosomal recessive
FRMD7	99.97 %	300628	Nystagmus, infantile periodic alternating, X-linked, 310700 (3), X-linked; Nystagmus 1, congenital, X-linked, 310700 (3), X-linked
GPR143	99.6 %	300808	Ocular albinism, type I, Nettleship-Falls type, 300500 (3), X-linked; Nystagmus 6, congenital, X-linked, 300814 (3), X-linked recessive
HPS1	100 %	604982	Hermansky-Pudlak syndrome 1, 203300 (3), Autosomal recessive
HPS3	99.91 %	606118	Hermansky-Pudlak syndrome 3, 614072 (3), Autosomal recessive
HPS4	99.98 %	606682	Hermansky-Pudlak syndrome 4, 614073 (3), Autosomal recessive
HPS5	99.91 %	607521	Hermansky-Pudlak syndrome 5, 614074 (3), Autosomal recessive
HPS6	100 %	607522	Hermansky-Pudlak syndrome 6, 614075 (3), Autosomal recessive
LRMDA	99.87 %	614537	Albinism, oculocutaneous, type VII, 615179 (3), Autosomal recessive
LYST	99.87 %	606897	Chediak-Higashi syndrome, 214500 (3), Autosomal recessive
MLPH	100 %	606526	Griscelli syndrome, type 3, 609227 (3), Autosomal recessive
MYO5A	99.94 %	160777	Griscelli syndrome, type 1, 214450 (3), Autosomal recessive
OCA2	99.6 %	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

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PAX6	99.95 %	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
RAB27A	99.94 %	603868	Griscelli syndrome, type 2, 607624 (3), Autosomal recessive
SLC24A5	99.99 %	609802	[Skin/hair/eye pigmentation 4, fair/dark skin], 113750 (3), Autosomal recessive; Albinism, oculocutaneous, type VI, 113750 (3), Autosomal recessive
SLC38A8	99.99 %	615585	Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis, 609218 (3), Autosomal recessive
SLC45A2	100 %	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
TYR	100 %	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; Albinism, oculocutaneous, type IA, 203100 (3), Autosomal recessive
TYRP1	99.97 %	115501	[Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)], 612271 (3); Albinism, oculocutaneous, type III, 203290 (3), Autosomal recessive

Explanation

OMIM release used for OMIM disease identifiers and descriptions: **2023-07-31**

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.