

OCA-OA panel (v1)

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(24 genen)

Centrum voor Medische Genetica Gent

GitHub commit: 669b7314f99860d45c0caa32b8c1a94bcabf2eee

Gene	OMIM gene ID	Associated phenotype, OMIM phenotype ID, phenotype mapping key and inheritance pattern
<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>BLOC1S6</i>	604310	?Hermansky-pudlak syndrome 9, 614171 (3), Autosomal recessive
<i>DTNBP1</i>	607145	Hermansky-Pudlak syndrome 7, 614076 (3), Autosomal recessive
<i>GPR143</i>	300808	Nystagmus 6, congenital, X-linked, 300814 (3); Ocular albinism, type I, Nettleship-Falls type, 300500 (3), X-linked
<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 {Albinism, oculocutaneous, type II, modifier of}, 203200 (3), Autosomal recessive; [Analgesia from kappa-opioid receptor agonist, female-specific], 613098 (3); {Melanoma, cutaneous malignant, 5}, 613099 (3); [Skin/hair/eye pigmentation 2, blond hair/fair skin], 266300 (3), Autosomal recessive; [Skin/hair/eye pigmentation 2, red hair/fair skin], 266300 (3), Autosomal recessive; {UV-induced skin damage}, 266300 (3), Autosomal recessive
<i>MC1R</i>	155555	COMMAD syndrome, 617306 (3), Autosomal recessive; {Melanoma, cutaneous malignant, susceptibility to, 8}, 614456 (3); Tietz albinism-deafness syndrome, 103500 (3), Autosomal dominant; Waardenburg syndrome, type 2A, 193510 (3), Autosomal dominant; Waardenburg syndrome/ocular albinism, digenic, 103470 (3), Autosomal dominant
<i>MITF</i>	156845	
<i>MLPH</i>	606526	Griscelli syndrome, type 3, 609227 (3), Autosomal recessive
<i>MYO5A</i>	160777	Griscelli syndrome, type 1, 214450 (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.

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

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.