OCA-OA panel (v1)

versie	2019-08-30 (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
AP3B1	603401	Hermansky-Pudlak syndrome 2, 608233 (3), Autosomal recessive
AP3D1	607246	?Hermansky-Pudlak syndrome 10, 617050 (3), Autosomal recessive
BLOC1S3	609762	Hermansky-Pudlak syndrome 8, 614077 (3), Autosomal recessive
BLOC1S6	604310	?Hermansky-pudlak syndrome 9, 614171 (3), Autosomal recessive
DTNBP1	607145	Hermansky-Pudlak syndrome 7, 614076 (3), Autosomal recessive
GPR143	300808	Nystagmus 6, congenital, X-linked, 300814 (3); Ocular albinism, type I, Nettleship-Falls type, 300500 (3), X-linked
HPS1	604982	Hermansky-Pudlak syndrome 1, 203300 (3), Autosomal recessive
HPS3	606118	Hermansky-Pudlak syndrome 3, 614072 (3), Autosomal recessive
HPS4	606682	Hermansky-Pudlak syndrome 4, 614073 (3), Autosomal recessive
HPS5	607521	Hermansky-Pudlak syndrome 5, 614074 (3), Autosomal recessive
HPS6	607522	Hermansky-Pudlak syndrome 6, 614075 (3), Autosomal recessive
LRMDA	614537	Albinism, oculocutaneous, type VII, 615179 (3), Autosomal recessive
LYST	606897	Chediak-Higashi syndrome, 214500 (3), Autosomal recessive
MC1R	155555	{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
MITF	156845	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
MLPH	606526	Griscelli syndrome, type 3, 609227 (3), Autosomal recessive
MYO5A	160777	Griscelli syndrome, type 1, 214450 (3), Autosomal recessive

OCA2	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
RAB27A	603868	Griscelli syndrome, type 2, 607624 (3), Autosomal recessive
SLC24A5	609802	Albinism, oculocutaneous, type VI, 113750 (3), Autosomal recessive; [Skin/hair/eye pigmentation 4, fair/dark skin], 113750 (3), Autosomal recessive
SLC38A8	615585	Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis, 609218 (3), Autosomal recessive
SLC45A2	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
TYR	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
TYRP1	115501	Albinism, oculocutaneous, type III, 203290 (3), Autosomal recessive; [Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)], 612271 (3)

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.