



Oculocutaneous Albinism, Type I

Alternative Names

OCA1
Albinism, Oculocutaneous, Type IA
OCA1A
Albinism I
Oculocutaneous Albinism, Tyrosinase-Negative
ATN

WHO International Classification of Diseases

Endocrine, nutritional and metabolic diseases

OMIM Number

203100

Mode of Inheritance

Autosomal recessive

Gene Map Locus

11q14-q21

Description

Oculocutaneous albinism (OCA) is manifested by reduced synthesis of melanin, which may result from mutations in a variety of genes. In OCA, hypopigmentation is both cutaneous and ocular, and in the most common types of OCA, other tissues and organs are not significantly involved. Hypopigmentation of the skin is associated with psychological and social problems, decreases natural protection against sunburn, and results in predisposition to skin cancer. In the absence of melanin in the eye, development of the visual pathways is abnormal, resulting in decreased visual acuity, strabismus, nystagmus and photophobia.

Molecular Genetics

One of the most common types of OCA is OCA1, an autosomal recessive disorder of reduced pigmentation in the hair, skin and eyes, resulting from mutations in the tyrosinase gene (TYR), a five exon gene spanning approximately 60 kb on chromosome 11q. Two different types of OCA1, A and B, can be distinguished in humans, based mainly on clinical observation, whereby OCA1A is the more severe type with absence of tyrosinase activity and pigmentation throughout life. The molecular basis of OCA1 is quite heterogeneous, and more than 190 mutations in the TYR gene have been identified so far in different population groups.

Epidemiology in the Arab World

Lebanon

Zahed et al (2005) performed ophthalmic and dermatological examinations on 30 Lebanese subjects with OCA, then screened for mutations in the tyrosinase gene in an effort to establish the molecular basis of the disorder in Lebanon.

References

Zahed L, Zahreddine H, Noureddine B, Rebeiz N, Shakar N, Zalloua P, Haddad F. Molecular basis of oculocutaneous albinism type 1 in Lebanese patients. *J Hum Genet.* 2005; 50(6):317-9.

Contributors

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