



Albinism, Oculocutaneous, Type III

Alternative Names

OCA3
Oculocutaneous Albinism, Type III
Albinism III
Rufous Oculocutaneous Albinism
Roca
Xanthism

Record Category

Disease phenotype

WHO-ICD

Endocrine, nutritional and metabolic diseases >
Metabolic disorders

Incidence per 100,000 Live Births

Unknown

OMIM Number

203290

Mode of Inheritance

Autosomal recessive

Gene Map Locus

9p23

Description

Oculocutaneous Albinism, type III (OCA3) is a form of brown albinism that was first discovered in people from Southern Africa. It causes mild hypopigmentation in light-skinned individuals, while in dark-skinned individuals, the condition results in bright copper-red skin coloration with freckles as well as bright copper-red hair. Affected patients also exhibit red reflex on transillumination of the iris, color dilution of the iris, nystagmus and strabismus.

Unlike other forms of albinism, OCA3 is considered a mild manifestation of the disorder.

Affected individuals have less severe vision abnormalities compared to OCA1 and OCA2. While the exact incidence rate of OCA3 is not known, it is a rare condition, mainly affecting dark-skinned people.

Diagnosis of the disorder depends on molecular testing of the TYRP1 gene. Affected individuals are advised to limit their exposure to the sun and to use broad-spectrum sunscreen to limit sun damage. They also require periodic ophthalmological evaluations and may benefit from low-vision aids.

Molecular Genetics

The disorder follows an autosomal recessive pattern of inheritance and is caused by mutations in the TYRP1 gene. This gene encodes an oxidoreductase enzyme found specifically in melanocyte cells that is involved in the biosynthesis of melanin. Homozygous TYRP1 mutations associated with OCA3 include deletions, missense and nonsense variants.

Epidemiology in the Arab World

Saudi Arabia

Monies et al. (2017) described the genomic landscape of Saudi Arabia based on the findings of 1000 diagnostic panels and exomes. One patient, an 11-year-old male, suffered from hemimegalencephaly, developmental delay and ADHD. He also had abnormal pigmentation all over his body. Whole exome sequencing helped identify a dual molecular diagnosis in this patient. A heterozygous mutation (c.1557T>G, p.Y519X) was found in exon 8 of the patient's TYRP1 gene, associated with oculocutaneous albinism type 3, and a heterozygous variant (c.90G>A, p.W30X) was uncovered in exon 1 of the TGIF1 gene, associated with holoprosencephaly 4. Such dual molecular diagnoses were rare and only occurred in 1.5% of the cohort.

References

Monies D, Abouelhoda M, AlSayed M, Alhassnan Z, Alotaibi M, Kayyali H, Al-Owain M, Shah A, Rahbeeni Z, Al-Muhaizea MA, Alzaidan HI, Cupler E, Bohlega S, Faqeih E, Faden M, Alyounes B, Jaroudi D, Goljan E, Elbardisy H, Akilan A, Albar R, Aldhalaan H, Gulab S, Chedrawi A, Al Saud BK, Kurdi W, Makhseed N, Alqasim T, El Khashab HY, Al-Mousa H, Alhashem A, Kanaan I, Algoufi T, Alsaleem K, Basha TA, Al-Murshedi F, Khan S, Al-Kindy A, Alnemer M, Al-Hajjar S, Alyamani S, Aldhekri H, Al-Mehaidib A, Arnaout R, Dabbagh O, Shagrani M, Broering D, Tulbah M, Alqassmi A, Almugbel M, AlQuaiz M, Alsaman A, Al-Thihli K, Sulaiman RA, Al-Dekhail W, Alsaegh A, Bashiri FA, Qari A, Alhomadi S, Alkuraya H, Alsebayel M, Hamad MH, Szonyi L, Abaalkhail F, Al-Mayouf SM, Almojalli H, Alqadi KS, Elsiesy H, Shuaib TM, Seidahmed MZ, Abosoudah I, Akleh H, AlGhonaium A, Alkharfy TM, Al Mutairi F, Eyaid W, Alsharbary A, Sheikh FR, Alsohaibani FI, Alsonbul A, Al Tala S, Balkhy S, Bassiouni R, Alenizi AS, Hussein MH, Hassan S, Khalil M, Tabarki B, Alshahwan S, Oshi A, Sabr Y, Alsaadoun S, Salih MA, Mohamed S, Sultana H, Tamim A, El-Haj M, Alshahrani S, Bubshait DK, Alfadhel M, Faquih T, El-Kalioby M, Subhani S,

Shah Z, Moghrabi N, Meyer BF, Alkuraya FS. The landscape of genetic diseases in Saudi Arabia based on the first 1000 diagnostic panels and exomes. *Hum Genet.* 2017 Aug;136(8):921-939. PMID: 28600779.

Related CTGA Records

Tyrosinase-Related Protein 1

External Links

<https://rarediseases.org/rare-diseases/oculocutaneous-albinism/>

<https://ghr.nlm.nih.gov/condition/oculocutaneous-albinism#inheritance>

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=79433

<https://rarediseases.info.nih.gov/diseases/4039/disease>

Contributors

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18.09.2017