



phthalmic findings in albinism

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Introduction and Purpose:

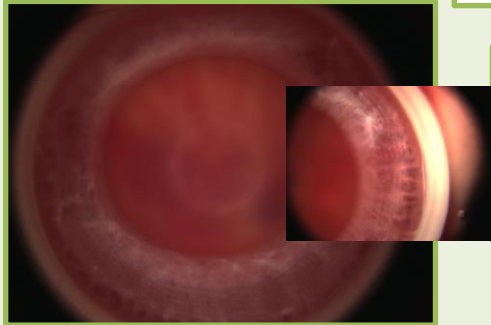
- Albinism is a rare **genetic disorder of melanin production**, which can affect only eyes or simultaneously eyes and skin/hair, resulting respectively in ocular (OA) or oculocutaneous albinism (OCA).
- Of all forms of albinism, OCA tyrosinase-negative is the most severe.
- OA is inherited most frequently as an x-linked recessive trait and OCA inheritance is autosomal recessive.
- Through of a case report of a child with OCA **we pretend review ophthalmological manifestations of albinism.**

Case Report:

A girl of West African descent was referenced to our appointment for ophthalmological evaluation of oculocutaneous albinism .

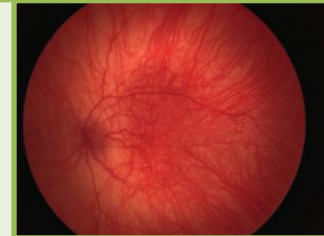


Visual acuity was 20/310 OD e 20/630 OS by teller cards.



In biomicroscopy, iris transillumination and hypopigmentation was visible , allowing to see spiral vessels and other iris details

Fundoscopy showed a dense and complex choroidal circulation due to lack of pigment in retinal pigment epithelium. Foveal hypoplasia was assumed because foveal pit is not apparent and vessels become less respectful of normal arcade and transverse the macula.



Results and Conclusion:

Melanin plays an important role in the development of the optic system and its absence leads to **diverse ocular manifestations** such as:

- iris hypopigmentation and transillumination
- reduced pigmentation of retinal pigment epithelium cells
- photoreceptor rod cell deficits
- foveal hypoplasia
- optic nerve hypoplasia
- misrouting of optic nerve at the chiasm, with temporal retina fibers inappropriately routed contralaterally instead of ipsilaterally.

Photophobia, nystagmus, reduced visual acuity, color impairment and strabismus are other manifestations usually seen in albinism.

Results and Conclusion:

- Ophthalmologists must be familiar with the specific visual manifestations and needs of albin patients.
- It is essential to correct refractive error to optimize visual acuity.
- Patients should be advised to wear tinted glasses and sunblock.
- In more severely affected children, they may benefit of low vision consultation and specialized low vision aids like telescopes.

