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seeing the invisible

DETECTION OF ALBINISM

OERs ADAPTED TO STUDENTS WITH LOW VISION.

redREE making projects

ALBA Asociación de ayuda a personas con albinismo

Aniridia Italiana

ANIRIDI NORGE

ANIRIDIA EUROPE

smallcodes

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I. INTRODUCTION

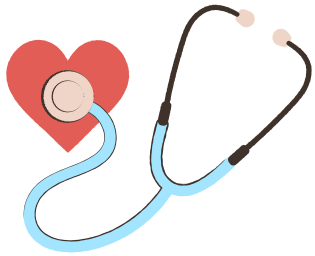
Albinism is a rare genetic disease with a very low incidence, of about 1 in 17000 people. It is characterized especially by visual anomalies that share all its variants, and in the vast majority of cases by pigmentation problems related to little or no melanin production.

There are many types of the disease, classified into two families: Oculocutaneous Albinism (OCA, with 7 varieties) and Ocular Albinism (OA). The vast majority of cases are OCA type I (40% of cases) and OCA type II (50% of cases). There are also several very rare added syndromes: Hermansky-Pudlak (with short life prognosis due to the possibility of pulmonary fibrosis), Chediak-Higashi (which causes severe immunodeficiency and progressive neurological degeneration), FHONDA (of more recent discovery, and doesn't involve pigmentary alterations), Nettleship-Falls (which can cause delayed sensorineural deafness), or Forsius-Eriksson.

These conditions begin in the formation of the fetus, in which they cause alterations in the development of the visual system. Albinism has no cure.

Albinism is autosomal recessive (there are very few dominant variants), so in order to develop it, the genetic alteration must be inherited from both parents. This is linked to the X chromosome, so some varieties have differences between sexes in the development of the disease and its inheritance.

II. SYMPTOMS



The extent and severity of symptoms varies greatly from type to type and from individual to individual, affecting both skin and vision:

- Lack of pigmentation in the iris and retina → photophobia, night blindness.
- Foveal hypoplasia and unusual optic nerve trajectory → decreased visual acuity, nystagmus, strabismus, amblyopia, lack of 3D vision (stereopsis).
- Refractive problems → myopia, hyperopia, astigmatism.
- Lack of pigmentation in the skin → hypersensitivity to UV radiation, propensity to develop keratosis, dermatological damage and skin cancer.
- All people with Albinism always have vision problems due to developmental problems of the retina and the ocular nerve, with varying degrees of impairment ranging from legal blindness to mild cases of visual problems, but most people with Albinism can use their remaining sight to carry out most day-to-day tasks.
- The phenotypes of the various types of Albinism involve quite different characteristics: skin and hair color can range from milky-white with blue-gray eyes (AOC1A), through brown skin, reddish hair, and blue or brown eyes (type III), white skin and golden hair (type V), or brown hair (type VII).

III. DETECTION

The vast majority of albino people are of white or very pale rosy complexion, which is almost already a diagnosis of their own, so the initial detection of Albinism usually requires just a visual examination noticing the very low or lack of pigmentation of the skin, hair, and eyes. This initial examination shouldn't be carried out in isolation, but considering the history and examples of pigmentation of the relatives, comparing the cases individually.

All its variants are detected with a thorough eye examination, especially of the retina, to recognize the problems related to malformations of the visual apparatus shared by all variants of Albinism. Thus, phenomena like iris translucency, decreased retinal pigmentation, fovea hypoplasia, and such consequences like decreased visual acuity, strabismus, or nystagmus can be verified.

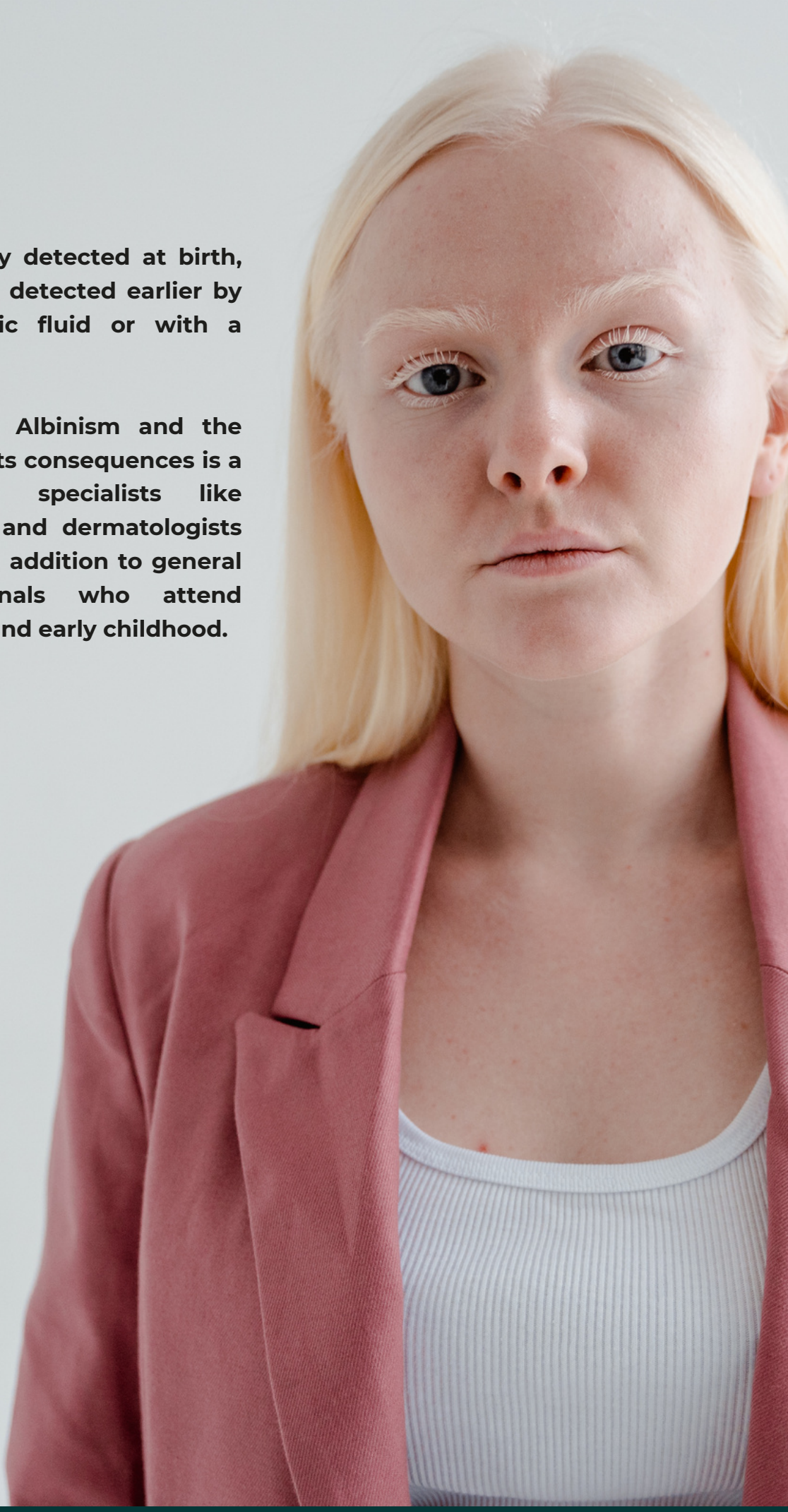
An electroretinogram can also be carried out to detect the alterations or confirm their scope: a non-invasive test in which electrodes are placed on the eyelid and cornea, to detect the response of the retina to light stimuli produced by electrical impulses and collected in a graph.

A skin biopsy may also be done to check for the presence or total absence of melanin.

The specific variety of the chromosomal alteration is determined by means of a specific genetic analysis, which also helps to understand the relationships of transmissibility through inheritance.

Albinism is usually detected at birth, but it can also be detected earlier by analyzing amniotic fluid or with a biopsy.

The detection of Albinism and the determination of its consequences is a task in which specialists like ophthalmologists and dermatologists must intervene, in addition to general health professionals who attend during childbirth and early childhood.



IV. TREATMENTS

There is no cure for Albinism. Its treatments are prophylactic or tend to improve the negative effects of the disease through proper eye care and monitoring of dermatological conditions.

The prevention of Albinism consists only of genetic counseling in family planning before having offspring. In the population as a whole, it is estimated that 1 in 50 to 70 people carries a genetic mutation related to Albinism, which means that 1 of every 2500 to 4900 random couples may have 25% of their offspring develop the disease.

The analysis of the genetic alteration helps not only to prevent the inheritance of the alteration, but also in guiding the medical action by identifying possible future complications.

The treatment of dermatological and visual conditions involves an annual specialized medical follow-up of the situation and evolution of the health of the affected person.

Surgical treatments can also be carried out to correct problems like strabismus or nystagmus, although these types of actions have a reduced scope.

People with low-incidence syndromic Albinism (Hermansky-Pudlak or Chediak-Higashi) should undergo specialized medical follow-up to avoid the development of other specific conditions related to the type of their genetic alteration.

V. RECOMMENDATIONS

Skin complications derived from the lack of pigmentation, like alterations, burns, keratosis, or skin cancer can and should be prevented by using protective creams against solar radiation with a 50+ factor and appropriate clothing to avoid prolonged exposure. People with Albinism should avoid being outside in hours and days of high UV radiation for a long time, at high altitudes, or near water. At least once a year, medical follow-up of existing dermatological alterations should be carried out.

Eye protection is equally important to avoid the consequences of photophobia, using glasses with special filters or colored contact lenses. In both cases they can also be used to correct refractive problems, thus helping to improve visual acuity. Other visual aids can also be used, such as convex lenses, which create a larger image on the retina, helping to it focus, and it can be adapted to any type of glasses, but aren't very aesthetic. On the other hand, magnifiers are a type of convex lens that can be used manually or using a support.

Telemicroscopes use several lenses, and there are lenses to see at all distances, although they are difficult to focus without prior learning, and reduce the visual field. Tele-magnifiers are electronic aids adapted to a closed-circuit TV that can reach up to 40 magnifications. The radio magnifiers incorporate a mini camera and a magnifying lens that send the signal to a conventional TV receiver being very useful and easy to transport.

Being a disease with well-determined phenotypic characteristics, Albinism can cause negative and undesirable reactions of rejection or misunderstanding by third parties. That is why it's important to teach young people with Albinism to cope with these reactions and the emotional and social problems they entail. To do this, we advise to encourage them to tell their feelings and experiences, and to learn to respond to possible embarrassing questions or even mockery.

The important thing is that they learn to face challenges and develop healthy communication and social interaction. In this sense, seeking support and help from social entities dedicated to Albinism is a very good initiative.

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