OCULOCUTANEOUS ALBINISM

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Summary

A case of partial albinism involving eyes and skin has been reported. It is a rare disease entity and is congenital in nature. No satisfactory treatment is advocated in this disease.

Oculocutaneous albinism is a rare disease entity and is characterised by the absence or paucity of melanin in the eyes and an unpatterned hypomelanosis in the skin and hair. It is believed to be transmitted as an autosomal recessive trait[1] with tendencies for partial or complete forms.

Albinos suffer from photophobia, refraction disorders and are usually highly myopic. Nystagmus as well as astigmatism are usually present. In such subjects, mental or physical retardation are not always observed, although an increased incidence of the latter is sometimes seen.

This condition is due to an enzymatic defect in the melanocytes, the latter being present in normal numbers in these cases. Enzyme tyrosinase which is synthesized by melanocytes is considered to be functionally defective and unable to catalyse the oxidation of tyrosine to melanin.

This disease entity is diagnosed on the basis of ocular as well as cutaneous findings, and is the first report from this institution during the last 18 years of professional service of the senior author.

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Case Report

A 15 years old male patient presented with low grade fever and mucopurulent expectoration for two months. Breathlessness and palpitation were also present. His mother had observed to and fro movements of the eye balls as well as pigmentary changes on the skin since birth. Examination revealed a poorly built and nourished individual, with ill-developed secondary sex characters. Testes were small, pubic and axillary hair were not present.

Chest had few basal crepitations with fixed splitting of 2nd heart sound in the pulmonary area. Rest of systemic examination was normal.

Skin : Fig. 1 Page No. 222). Hypo as well as hyperpigmented patches were seen distributed mainly on the chest and abdomen. These were smooth with sharply demarcated border and varied in size from 2 to 5 mm.

Eyes : Patient was photophobic and had blue sclera. Nystagmus was present in all directions. Fundus examination revealed myopic fundus with

partial albinism. Disc was bigger in size and was pale with narrowed arteries. Fundus was tasselated and thinned out, macula not being properly defined or pigmented. Macular or foveal reflex was absent. Patient could not focus the light as no fixation had developed.

Slit lamp examination showed normal cornea while atrophic pattern of the iris was detected. Some degenerative changes of vitreous humor were present. Retinoscopy revealed a high degree of myopia.

E. N. T. Examintion showed chronic suppurative otitis media with central perforation.

Investigations: Histopathology of the skin (Fig. 2 Page 222). showed marked hyperkaratosis with thinning of the strautum malpighii. Occasional areas revealed epithelial cleft formation. The upper dermis showed the presence of round and spindle shaped cells in relation to the blood vessels. Some brownish pigment was also present in the upper dermis.

Sex chromatin: Buccal smear for sex chromatin was negative.

Skiagram of the chest: Showed a patch of consolidation on right paracardiac area while electrocardiogram was suggestive of nodal extrasystoles.

Sputum examination for AFB and pyogenic organism was negative.

Routine investigations: Showed Hb. 9.12 g%, TLC-8700/c. mm, DLC-P69, L39, E 1. B 1% with ESR of 39 mm. in 1st hour (Westergren) urine and stools examinations were normal. Urine was negative for porphyrins,

Routine Liver Function Tests and Serum Alkaline Phosphatase were normal. Hair and urinary tyrosinase levels were inconclusive.

Discussion

Albinism can occur in total or partial forms. Partial albinism occurs as a congenital absence of pigment in different parts of the body and occurs as three main varieties, each representing a different mutation. The three types, according to Andrews[2], are the bald albinism, white forelock and white spotting of the skin. Ocular albinism cases showed hypoplasia of the macula, head nodding, nystagmus and amblyopia. When associated with skin changes, of whitish and brownish spots; in addition to rotatory nystagmus photophobia and a high degree of myopia, were also seen.

This disease is characterised by local absence of pigment in affected areas where hairs are blond, silky and brownish in colour. The lesions often do not increase in size and are sometimes surrounded by a zone of hyperpigmentation[3]. In our case also the lesions did not increase and were present since birth. He also showed ill-developed secondary sex characters. However, his mental status was not significantly affected, though mental and physical retardation may occur in some cases[3].

This case presented with cough and expectoration. Repeated sputum examinations, and skiagram of the chest, ruled out tuberculosis or other chronic lung lesions. Lack of resistance to such infections has been reported[4]. A patch of pneumonitis which was present disappeared with treatment. In Chediak-Higashi syndrome, a rare autosomal recessive disorder; in addition to other usual features of albinism, an increased susceptibility to infections and development of lymphomas, caused by virus are seen[5].

This condition is incurable and treatment is ineffective. It is important to avoid exposure to sunlight in these cases because of the danger of developing skin convers as a result of the absence of its protective pigment[3].

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FALSE

In 1978 Cleaver reported that excision repair of DNA was absent or markedly reduced in fibroblasts from patients with Xeroderma Pigmentosum (XP). This was the first report of a mammalian cell line with an abnormality in this type of DNA repair. Although the majority of patients with XP show this abnormality there are some who have what appears to be the same disease clinically but in whom excision repair is normal. Horkay et al also reported very low levels of excision repair after UVR in lymphocytes of six patients with polymorphic light eruption, as well as in patients with XP.

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