ALBINISM — A CASE REPORT

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Albinism means the absence of cutaneous pigments. This is a rare hereditary disorder of the skin found in all races. The basic defect is an inherent deficiency of the enzyme tyrosinase usually found in melanocytes. Albinism can be total or partial. The frequency of total albinism varies in different countries, being common in small communities, where there is a greater likelihood of intermarriage. In certain parts of the U.S.A. the incidence is about 1 in 20,000 whilst in France it is 1 in 100,000. The occurence of total albinism in Malta, in spite of the smallness of the country, is probably even rarer. A case of total albinism in a Maltese child is here reported.

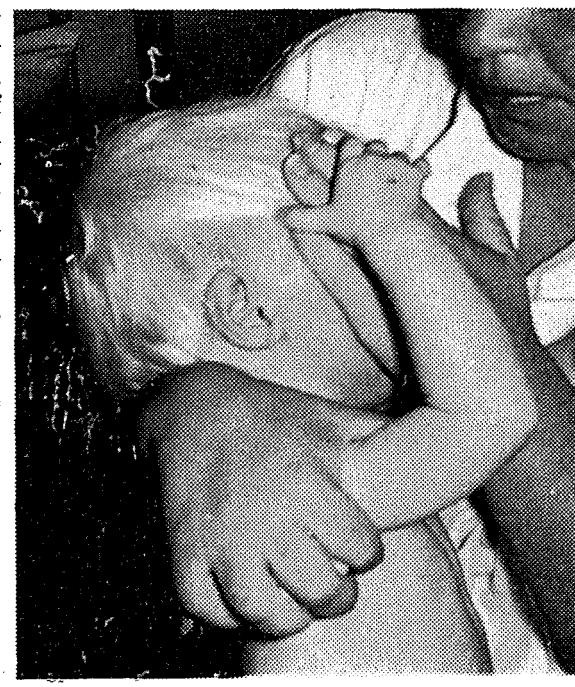
Case Report

Baby M.C. is the first child of healthy unrelated parents. She was born on the 16th May 1972 after a full term normal pregnancy. Delivery was normal. Her birth weight was 3 kg.

At birth, features of total albinism were present. She is now 18 months old, and her health has been quite satisfactory except for gastroenteritis at the age of 15 months, for which she required hospitalisation. Her mental development has been quite normal for her age, but her weight and height are in the 10th percentile range.

Photograph 1 shows some of the features of this child's pigment abnormality. Her skin is extremely white which can be seen by contrast with the normally pigmented skin of the nurse holding her. Her hair is fine, silky and whitish. Her eyebrows and eyelashes are also white.

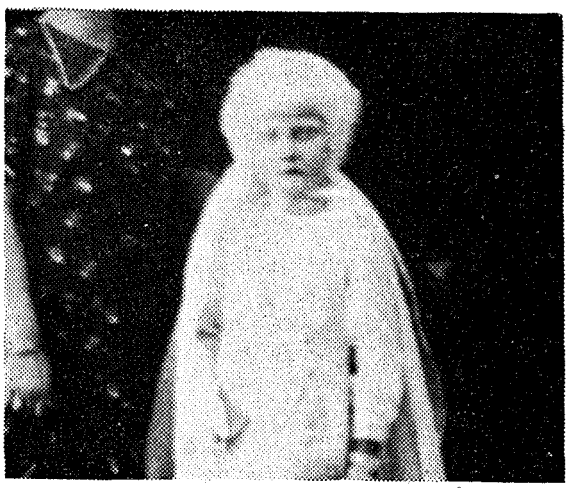
She has gross photophobia and nystagmus. In sunlight or bright artificial light, her eyes are kept tightly closed. Indoors, provided the light is not too bright, her eyes are open, showing bright red pupils. Because of this photophobia, and the harmful effect of sunlight on the unprotected unpigmented skin, the child is never taken out in the sunlight and is kept indoors most of the time. Her parents are Maltese, in their middle twenties, quite healthy, both having dark skin.



Photograph 1.

There is a family history of total albinism. The child's father has two cousins who are albinos. They are both females (Photographs 2 and 3) born and living in Australia of Maltese unrelated parents. These children are seen in a family group photograph. The father and mother and three siblings have normal coloured hair and skin. These photographs were

taken 12 years ago. Since that time these two girls have got married and now have two normal children each. These children are first cousins of the child's father, the respective fathers being brothers. The child's paternal grandfather had four brothers and seven sisters. Except one, they are all married and between them they had thirty children and up to now thirty-five grandchildren. Apart from the three members mentioned in this case report, the others all have normal pigmentation of hair and skin. There are no partial albinos.



Photograph 2.

Discussion

Pathology — Albinism is the total or partial absence of melanin in the affected parts of the body. The pigment-forming cells of the skin, the melanocytes are situated in the basal layer of the skin (Deutsch & Mexon — 1957). Melanocytes are also present in the eyes, (uveal tract and retinal pigment epithelium), C.N.S. (leptomeninges) and other sites such as mucosae, orbits and mesentery. Melanocytes are embryologically derived from the neural crest.

Melanocytes synthesise the copper-containing enzyme tyrosinase, a very important enzyme which catalyses the oxidation of tyrosine to melanin.

Tyrosine is derived by hydroxylation of the essential aminoacid phenylalanine. The metabolism of phenylalanine can be represented as follows:—

$$\text{Phenylalanine} \xrightarrow[\text{Hydroxylase}]{\text{Phenylalanine}} \text{Tyrosine}$$

The important enzyme tyrosinase, synthesised by the melaocytes then oxidises tyrosine through D O P A↓ to melanin.

$$\text{Tyrosine} \rightarrow \text{DOPA} \xrightarrow{\text{Tyrosinase}} \begin{matrix}\text{Melanin} \\ \text{Melanoprotein}\end{matrix}$$

What happens in Albinism? The Albino has the normal number of pigment-forming cells (melanocytes). But, there is an inherent defect in the enzyme tyrosinase, the oxidation of tyrosine through D O P A to melanin does not take place, and so the cells do not form melanin. This metabolic abnormality may affect all the melanocytes of the body or only those of the eye or the eye and skin.

Clinical Types—Albinism exists in 2 forms (Turpin 1941).

a) *Total Albinism:*— Oculocutaneous, characterised by unpigmented skin, hair, eyebrows, eyelashes and eyes, as in the case presented. There is a hypopigmented fundus oculi, translucent irises with diminished visual acuity, light intolerance and nystagmus. There is a tendency to malignancy in exposed areas of the skin (De Smeth 1956). This has been reported in tropical areas. Precancerous areas have been reported. These are irregular blotches of freckle-like pigmentation which may develop in areas exposed to light. Total Albinism is inherited via an autosomal recessive gene (Whitkop C.J. — 1970).

b) *Partial Albinism:*— Is more common. Here the hair is yellow or light brown. Errors of refraction, photophobia and nystagmus are common. The iris is light blue or pink. The partial form is often transmitted by a dominant gene, but in ocular albanisms where ocular finding only are present the inheritance is sex-linked recessive. Most albinos are of small stature and fertility and expectation of life are reduced. Mental retardation is sometimes associated.

Photograph 3.

Management of these Children — The most important problem in the management of these children is the protection of the unpigmented skin and eyes against the harmful effects of sunlight. The skin is unable to tan and so it is subject to sunburn. Direct sunlight has to be avoided at all costs especially so in summer, otherwise these children easily get severe burns. In the hot summer months even when they stay in the shade, these children are prone to burns from ultraviolet sunrays. So light screens like a 5% solution of para-amino benzoic acid in spirit should be used. This is an effective and cosmetically acceptable preparation. Besides, long-sleeved garments and head wear are advised to decrease as much as possible the skin areas exposed. Tinted glasses are advisable from an early age to protect the unpigmented iris and retina from the damaging effect of the sun.

Prognosis—In temperate climates the prognosis for the albino is good. The main disabilities are the visual defects and the susceptibility to sunburns. Constant supervision by physician and ophthalmologist are nessary. As happened in the case reported these children can marry and have normally pigmented children.

As a final comment one can add that albinism is one of a group of diseases known as hereditary hypomelanoses. It is due to defective biosynthesis of tyrosinase. There are other forms of hypomelanoses where the cause is different.

Hypomelanoses can be due to (i) abnormalities in the morphology (or absence of) the melanocyte as in-

Vitiligo
Piebaldism and white forelock
Waardenburg's syndrome, where there is premature greying of the hair associated with eye changes and congenital nerve deafness.

or to (2) Defective biosynthesis of tyrosine-melanin as in phenylketonuria. Here there is a metabolic block in the conversion of phenylaline to tyrosine, due to absence of the enzyme phenylalanine-hydroxylase.

Defects associated with albinism have also been reported, such as the Chediak-Higashi syndrome where oculocutaneous albinism is accompanied by defective leucocytes which are unable to phagocyte. These children get intractable infections in childhood.

Deaf mutism (Margolis 1962) and pseudohaemophilia (Larson 1972) have also been reported.

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