melatonin secretion per se as melatonin administration has not been shown to duplicate the functional effects of the pineal. It is possible that other, as yet undefined, pineal hormones or metabolites may be implicated in causing some of the features of the syndrome.

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Collodion babies with Gaucher's disease

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SUMMARY Two neonates with acute infantile cerebral Gaucher's disease had prominent collodion skin. Ichthyosis has been described in some cases of metabolic lipid disorders, however, this is the first report of the association of lamellar desquamation of the newborn (collodion baby) with Gaucher's disease.

Gaucher's disease is characterised by an abnormal accumulation of cerebrosides primarily in cells of the reticuloendothelial system. The disease is due to a deficient activity of lysosomal glucocerebrosidase and is transmitted as an autosomal recessive trait. It is a clinically heretogeneous disorder, however, with at least three recognised types. In the acute infantile cerebral form (type II) the baby appears normal at birth but soon exhibits neurological symptoms and hepatosplenomegaly. Death usually occurs in the first year of life. The skin is not pigmented in the acute infantile form. Collodion skin and ichthyosis are not recognised skin manifestations in any of the forms of Gaucher's disease.[1] We report two siblings with generalised ichthyosis associated with infantile Gaucher's disease.

Case reports

A 27 year old Lebanese woman was admitted to this hospital in September 1986 for her second confinement. There was no consanguity with her Lebanese husband and there was no family history of Gaucher's disease or ichthyosis. Her first pregnancy in 1984 resulted in the delivery of a boy (case 1) in another hospital in Sydney.

Case 1. A 3500 g boy was delivered by caesarean section at 39 weeks' gestation. Generalised thick collodian like skin, gross hepatosplenomegaly, and apathy were noted at birth. The ichthyotic skin peeled over the next five days and there were no subsequent skin lesions. Soon after birth he had recurrent laryngospasms, generalised convulsions, and a persistent thrombocytopenia. Diagnosis of Gaucher's disease was established by leukocyte enzyme assays. He died at the age of 3 months after a respiratory arrest in hospital. The parents did not consent to a postmortem examination.

Case 2. The mother presented at 20 weeks' amenorrhoea but declined the offer of prenatal diagnosis. Polyhydramnios occurred in late gestation and a boy weighing 3400 g was delivered by caesarean section at 40 weeks' gestation. He was apnoeic at birth and remained dependent on assisted ventilation until death at 11 days of age. Generalised tight collodian like skin (figure), gross hepatosplenomegaly, and generalised joint contractures were noted. He was apathetic from birth and had frequent refractory convulsions and opisthotonos. The tight collodian like skin started to shed from the first day of life. At the time of death the hands, feet, and back were scaling in large sheets.

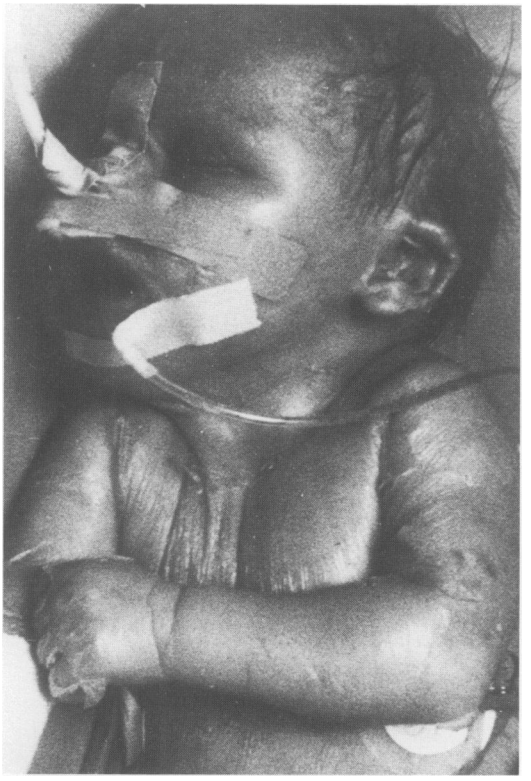

Figure *Case 2 at 3 days old showing desquamation of collodion skin.*

Results of laboratory investigations showed a persistent thrombocytopenia with platelets in the range $19–35 \times 10^9$/l and a mild conjugated hyperbilirubinaemia. His liver enzymes were normal but acid phosphatase was raised (28 U/l, normal 5·4 U/l). White cell enzymes measured (in pmol/minute/mg protein) showed deficiencies consistent with Gaucher's disease. The β-glucocerebrosidase was 35 pmol/minute/mg protein (normal range 600–3200), β-glucosidase was 17·1 pmol/minute/mg protein (normal range 60–220).

At postmortem examination Gaucher cells were found in lymph nodes, thymus, liver, spleen, adrenals, and bone marrow. The brain also showed changes consistent with cerebral Gaucher's disease. Section of skin of the back showed hyperkeratosis with follicular plugging. The underlying epidermis and dermis appeared normal. The skin was fixed in Millonig's buffered formalin and processed for transmission electron microscopy. The keratinocytes showed normal maturation and no abnormal storage products were seen. The keratinised layer was thick but otherwise normal. The granular layer was considerably reduced in thickness.

In January 1987 the parents returned for follow up counselling. The mother reported that she was pregnant at 11 weeks' amenorrhoea. A prenatal diagnosis by chorionic villi biopsy was performed, and this was followed by a first trimester abortion as the fetus was affected with Gaucher's disease.

Discussion

A striking clinical feature in these two infants was generalised collodion skin. Although scaling disorders have long been attributed to abnormalities of keratin protein, there has been increased awareness of the relation between ichthyosis and specific lipid metabolic disorders. A few examples include X linked ichthyosis with steroid sulphatase deficiency,[2] neutral lipid storage disease,[3] and Refsum's disease.[4]

Elias emphasised the importance of lipids in the integrity of the stratum corneum based on evidence from morphological studies by freeze fracture imaging and by histochemical and cytochemical studies.[5] He suggested abnormal lipid composition would affect the normal cohesion and orderly dissolution of corneocytes, which would be the basis of scaling skin disorders. In our infants the delicate balance of stratum corneum lipid composition may have been disturbed as a direct effect of the lipoidosis, resulting in the collodion membrane. As in the two cases we report, authors reporting other lipid diseases have not described any obvious abnormal storage products seen in ultrastructural examination of the ichthyotic skin that is associated with these diseases.[2–4] An alternative suggestion is that the ichthyosis and lipid disorder may represent allelic disorders with different mutations in the structure gene for the enzyme.[6] The range of clinical presentation including collodion baby in these two cases may be due to a single mutation at the glucocerebrosidase locus. Furthermore, it is also possible that the hyperkeratosis and glucocerebrosidase deficiency were two closely linked genetic conditions and that these two infants represented a variant of Gaucher's disease with neurocutaneous manifestations.

We thank Dr Andrew Berry of the Royal Alexandra Hospital for Children, Sydney, in providing information of the first reported case.

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