PORENCEPHALY, NASOFRONTAL MUCOCELES, HYPERTELORISM AND SEGMENTAL VITILIGO

— REPORT OF A NEW NEUROCUTANEOUS DISORDER

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INTRODUCTION

The purpose of this paper is to describe an interesting new syndrome consisting of porencephaly, nasofrontal mucoceles, ocular hypertelorism and segmental vitiligo.

The patient, a 15-year old Malay boy presented with recurrent epilepsies, oligophrenia, congenital hemiplegia, bilateral strabismus and ocular nystagmus, bilateral nasofrontal swellings, a segmental skin depigmentation on the right lower abdomen and the right lower limb.

The association of the congenital neurofacial malformations and the cutaneous lesion is discussed.

CASE REPORT

A Malay boy, aged 15, was first admitted to the Medical Unit on March 1968 for investigation of epileptic fits since childhood. The history obtained from his mother revealed that he was born normally without any complication in the antenatal period. There was no history of drugs intake, exposure to irradiation or to systemic infections. At birth, he was observed to have abnormal facial and cutaneous features. His milestones, according to his mother, were all delayed. Later, he was found to have mental retardation as well as emotional instability and fluctuation of moods. Because of the mental disturbance, he was not sent to school and was being looked after by his relatives. However, he was able to look after himself and obey simple orders or carry out simple tasks at home.

The epileptic attacks first noted at the age of three, were associated with progressive mental changes and enlargement of the cranium. Initially, the fits were focal in nature, starting in the right hand and spreading to the right half of the body, but a few years later, it became generalized. With recurrent grandmal epileptic attacks, the limbs on right half of the body became weak, spastic and underdeveloped. At first the epilepsy was controlled by small doses of phenobarbitones but over the past few months, there were increased number of fits in spite of the usual anticonvulsant medication.

Over these years, the nasal swellings and the left occipital portion of the skull had progressive grown in size, but the skin pigmentation remained unchanged since birth. Although he was born with the internal squints and bilateral shifting movements of the eyes, he had no visual complaints. He denied any headache, vomiting, auditory, speech or sphincteric symptoms.

The parents, apparently unrelated had no neurological or skin disorders. The father, aged 46, was a marine police, and the mother, aged 42, a housewife, had been recently treated for peptic ulcer.

The patient's only sister, aged 22, was married and well. There were no family history of neurocutaneous disorders or other major illnesses.

The patient on examination, was found to be conscious and well nourished, but he was unable to converse rationally. His I.Q. was 40 to 50, height 4 feet 6 inches and weight 87 lbs. Pulse rate was 80/min. Blood pressure 110/70. There were no abnormalities in the cardiovascular, respiratory, gastrointestinal or renal systems. Secondary sexual characteristics were present. There was posterior bulging of the left parieto-occipital area of the cranium and the affected bone was thinned. His face showed marked ocular hypertelorism with broadening of the nasal root, increased displacement of inter-pupillary distance, and a widened intercanthus distance of 42 cm. At the base of the nose, there were two nasofrontal mucoceles which could be compressed, inflated and deflated at will (Figs. 1 and 2). Both eyes showed bilateral internal strabismus with coarse horizontal nystagmus, but the visual acuity and fields were normal. The pupils, ocular fundi, and other cranial nerves were also normal. Motor power of the right upper and lower limbs was weaker (Grade 4) than the opposite normal limbs. The right limbs were spastic, underdeveloped and their tendon reflexes were all exaggerated. There

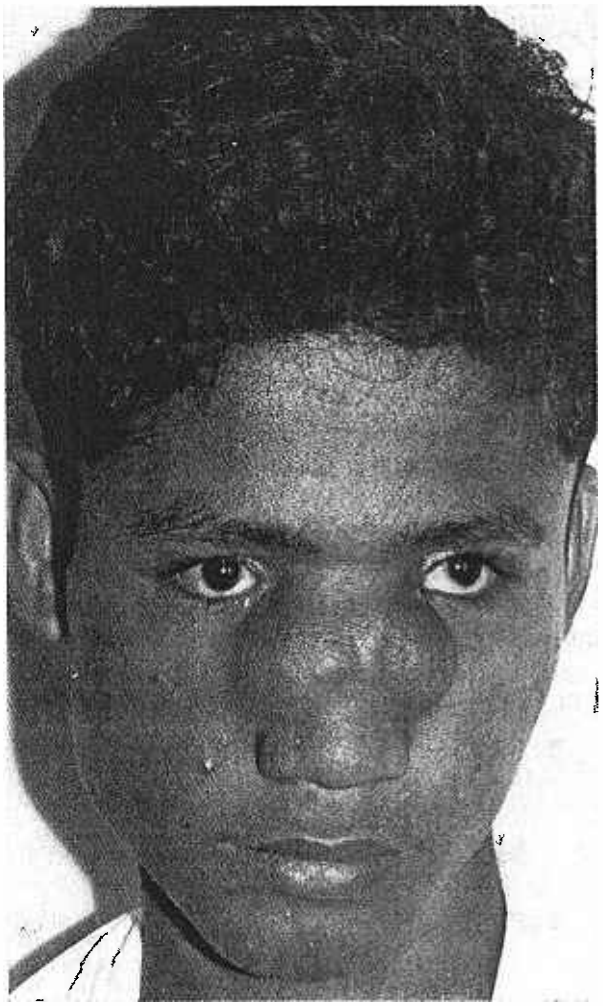

Fig. 1. Bilateral nasofrontal mucoceles — fully inflated by blowing the nose.

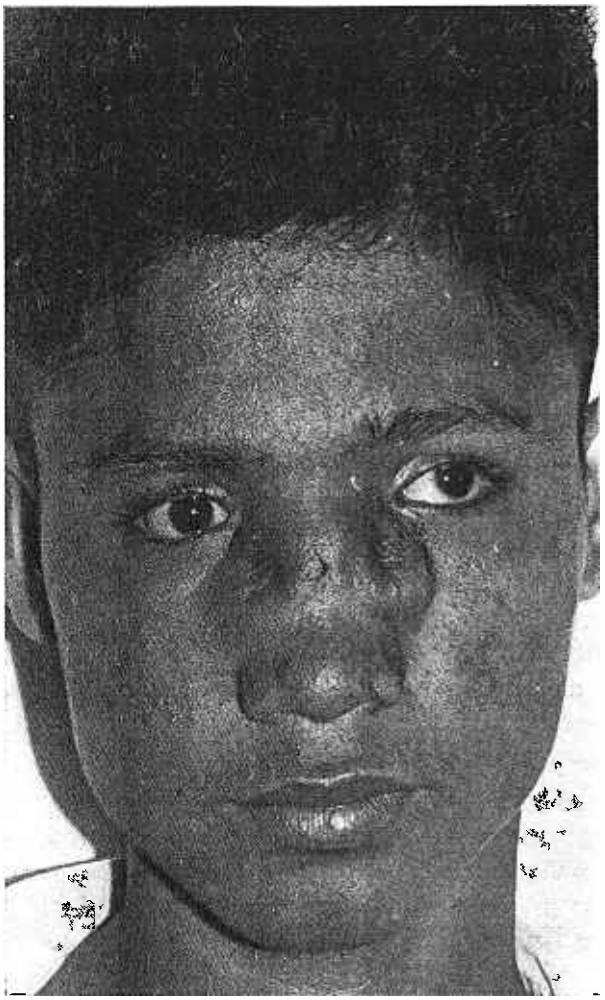

Fig. 2. As Fig. 1. Mucoceles both deflated. Note the ocular hypertelorism and the bilateral internal strabismus.

were bilateral ankle clonue but the plantar responses were flexors. His cerebration was slow, mental functions poor, and there were no parietal, cerebellar or sensory changes.

Segmental vitiligo extending from the right thoracic 10th to the right Lumbar 2nd dermatome were present (Fig. 3) and the lesion was sharply demarcated from the normal skin by a hyperpigmented border. Except for the depigmentation, the cutaneous texture of this lesion was normal, and there were no evidence of secondary diseases.

INVESTIGATIONS

The following investigations were found to be normal:—

Hb., T.W. and D.C., E.S.R., Urine full and microscopic examination, Blood urea, serum electrolytes, Blood Kahn Test, urine phenylketones, sugar and acetones, cerebrospinal fluid, X-rays chest, abdomen, and spines, and chromsome karyotypes.

E.E.G. showed generalised slow waves and low voltages. Skull X-ray revealed a left parietal bulging with localised sutural diastasis. The frontal and nasal sinuses were radiologically normal. Lumbar air encephalogram showed a large porencephalic cyst of the left cerebral hemisphere projecting into the left occipital region so that there was only a thin layer or cortex remained posteriorly. The left ventricle was also grossly dilated (Figs. 4, 5 and 6).

DISCUSSION

The association of porencephaly, nasofrontal mucoceles, ocular hypertelorism and segmental vitiligo has not been previously documented although each condition is a well-known entity.

Porencephaly is a rare congenital disease (Moriarty and Klingman, 1966; Cohn and Neumann, 1946) and is characterized by atypical bilateral cavities in the cerebral hemisphere that communicate with the ventricles without entering into the sub-arachnoid space. Each cavity

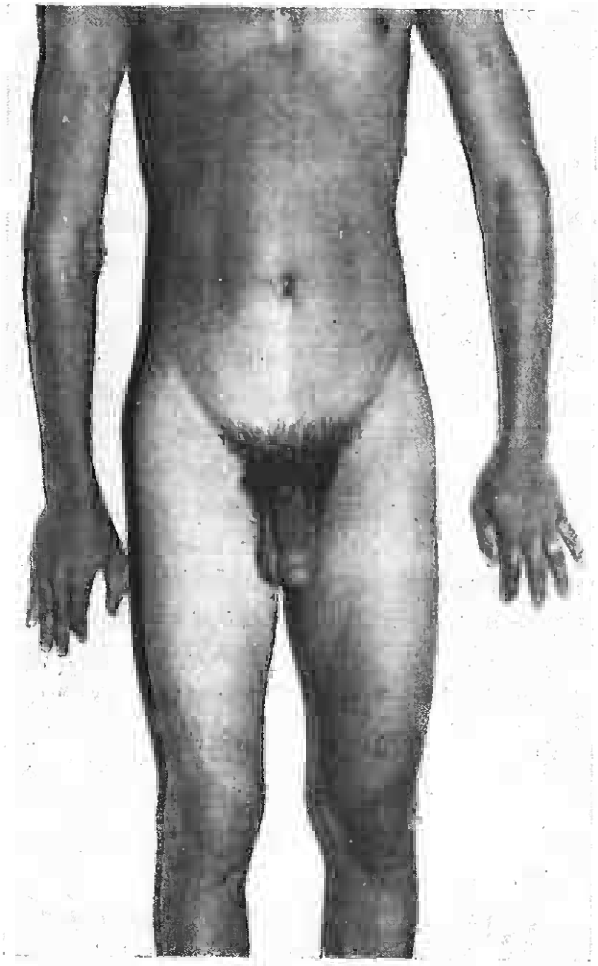

Fig. 3. Segmental vitiligo from right T10 to L2.

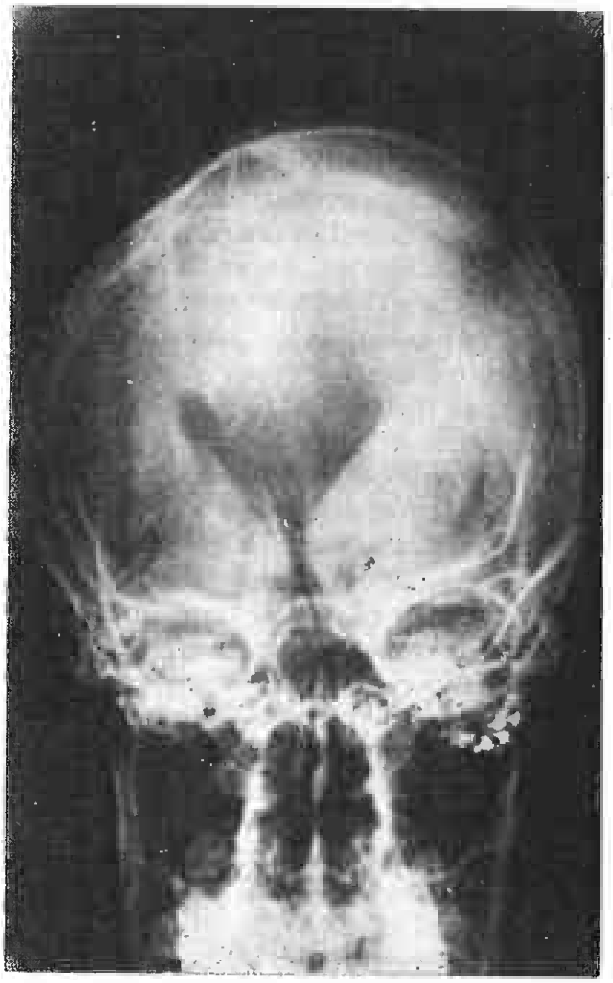

Fig. 4. Air encephalogram showing a large left porencephalic cyst communicating with the left ventricle.

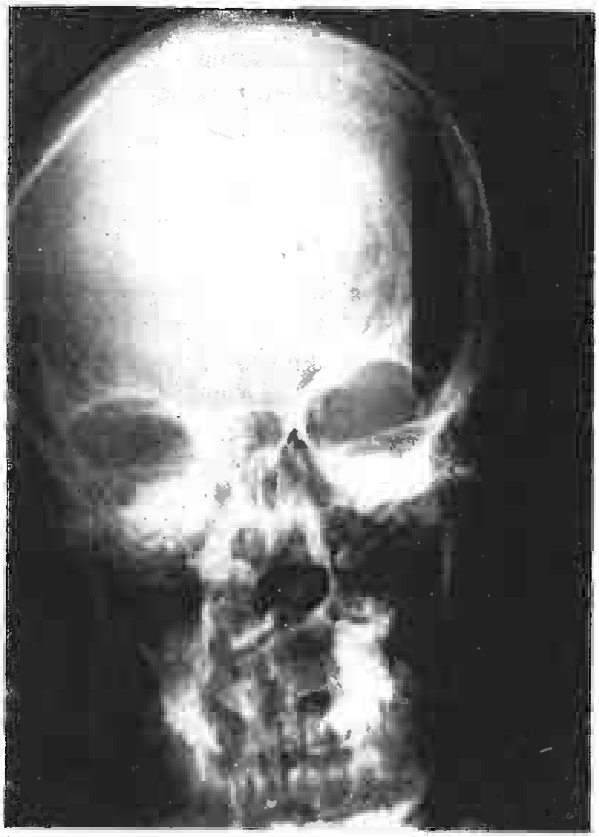

Fig. 5. As Fig. 4. Showing the extent and size of the porencephalic cyst towards the left lateral cerebral hemisphere.

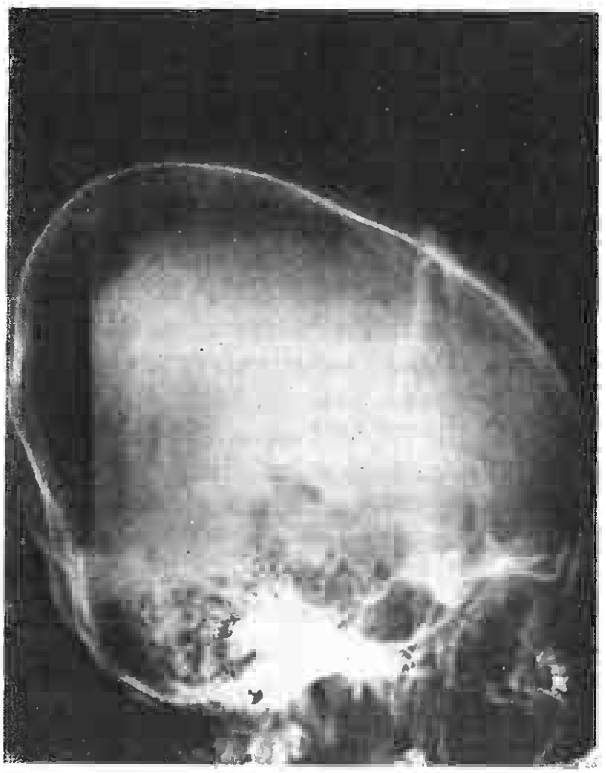

Fig. 6. As Fig. 4. Showing the posterior extent of the cyst and the bulging and thinning of the parieto-occipital part of the cranium.

or cyst may gap or be covered by pia-arachnoid or by a normal dural and skull. Rarely, however, single porencephalic cyst as found in our patient occurs, and this condition should be distinguished from the false or pseudo-porencephalic cyst which does not communicate with the ventricle and is caused by destruction of cerebral tissue from other aetiologies rather than by abnormal cortical development. Large size porencephaly as seen in the patient may distort the brain, causing ventricular obstruction and may produce skull defects like cranium bifidum, craniofenestria or cranioschisis. The thinned area of the cerebral hemisphere is often a persistence of the foetal condition as evidenced by the hypoplastic structure. Thus depending upon the location and the extent of the porencephalic cyst, wide range of clinical manifestation may be presented. Thus in our patient, the huge left porencephalic cyst which extended from the frontal to occipital areas was responsible to his focal and generalized epilepsies, congenital hemiplegia, underdevelopment of the right limbs and the diffuse mental disturbances. Occasionally, associated maldevelopment of other neural tissues like basal ganglia, thalamus, brain stem and its nuclei may give rise to pyramidal and extrapyramidal signs, cranial nerves palsies and other neurological deficits. Bilateral congenital internal strabismus and ocular nystagmus in our case may be attributed to hypoplasia of the abducens and vestibular nuclei. Obstructive hydrocephalus and its attending symptoms were not found in this patient. Porencephaly may be found in association with various types of encephaloceles, but not with cutaneous changes such as vitiligo. (Cohn and Neumann, 1946). Sincipital encephaloceles are found in 15% of all encephaloceles and these malformations are found on the dorsum of the nose, the orbits and sometimes, the forehead. (Blumenfeld and Skolnik, 1965; Record and McKeown, 1949; McLaurin, 1964). However, in nasofrontal mucoceles, the herniated sacs of the nose do not communicate with the central nervous system as they are lined by nasal mucosa and could be inflated and deflated by will e.g. by blowing the nose as in our patient, whereas sincipital sacs with central connection may contain meninges (meningocele) or meninges and neural tissues (encephalocele), and lack the features of the inflatable mucoceles. Close to the nasal defect is another facial malformation, the ocular hypertelorism found in the present case. Increased growth of the lesser wings and decreased development of the greater wings of both sphenoidal bones are often the cause of the hypertelorism. This condition is also present with sincipital encephaloceles. Congenital cutaneous defects are rarely described with encephaloceles, mucoceles or hypertelorism. Orkin and Fisher (1966), in a survey of some of above conditions found congenital dermal sinus, dermoid cysts, congenital scalp defects such as bullae, erosions and ulcerations, abnormal hair-tufts, patchy alopecia and heterotropic brain tissues in the skin of their patients. There was no mention of any congenital pigmentary disorders.

The presence of the congenital segmental vitiligo on the paralysed limb of our patient may be a part of the generalised neurological maldevelopment since it is known that idiopathic vitiligo is often controlled by the nervous system (Lerner, 1955; Lerner *et al*, 1966). Vitiligo may develop at the level of transverse myelitis and on the hemiplegic limb of cerebrovascular accidents. It is also found in some neurological diseases such as Vogt-Koyanagi-Harada's syndrome and the Alezzandrini syndrome.

The primary defect of this neurocutaneous disorder must be the ectodermal (neural) tissue which would account for the congential malformations of the skull, brain, face and skin. The aetiology of this disease is not known. There are no familial disorder, chromosome abnormalities or any known causes. It is most likely a sporadic condition.

SUMMARY

A neurocutaneous syndrome characterized by porencephaly, nasofrontal mucoceles, ocular hypertelorism and segmental vitiligo is described. The multiple congenital malformations could be due to a ectodermal defect in the embryoloic stage of development. This disease is most likely a sporadic condition.

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