ECTODERMAL DYSPLASIA WITH HYDROCEPHALUS: A RARE ASSOCIATION

Dear Sir,

Ectodermal dysplasia (ED) comprises a large, heterogeneous group of inherited, non-progressive disorders that are defined by primary defects in the development of two or more tissues derived from the embryonic ectoderm. The tissues involved are primarily the skin, hair, nails, eccrine glands and teeth.[1] Pure EDs are manifested by defects in ectodermal structures alone, while ED syndromes are defined by a combination of ectodermal defects in association with other anomalies. We present a nine-month-old girl who had a rare association of ED with developmental delay and hydrocephalus.

Our patient was a nine-month-old Indian girl who presented with a gradually increasing head size that was noticed since two months of age. Global developmental delay was present. She had partial head control, was able to reach out for objects and laugh aloud when excited, which puts her developmental age at four months. The patient had a history of failure to thrive but no history of convulsion, decreased vision or hearing loss, heat intolerance, decreased sweating, lacrimation or any feeding difficulties. She was the third child born of a non-consanguineous marriage, with a normal birth history. Examination of her parents and two older brothers revealed no sweating deficiency and hair, nail or teeth abnormalities.

On examination, the patient was conscious, and had a heart rate of 100 beats per minute, respiratory rate of 28 breaths per minute and blood pressure of 90/64 mm Hg. Her weight and length were below the fifth percentile, and her head circumference was 47 cm (more than 95th percentile for age and gender). Dysmorphic facies were present in the form of low-set deformed ears, long philtrum, thin upper lip and thin vermillion borders. Hair abnormalities included sparse eyelashes, and absent eyebrows and scalp hair (Fig. 1). The patient also had anodontia (congenital absence of teeth). Her nails and skin were normal. She had post-axial polydactyly (six) in both her upper and lower limbs. Ultrasonography of the abdomen was normal. Computed tomography (CT) imaging of the brain revealed mild dilatation of the frontal and body of the lateral ventricle, third ventricle and fourth ventricle, suggestive of communicating hydrocephalus (Fig. 2). Conservative management with regular follow-up was advised by the neurosurgeons. The patient was discharged and is well on follow-up.

Fig. 1 Photograph shows dysmorphic facies in the form of low-set deformed ears, long philtrum, thin upper lip and thin vermillion borders. Also seen are sparse eyelashes, absent eyebrows and scalp hair, and anodontia.

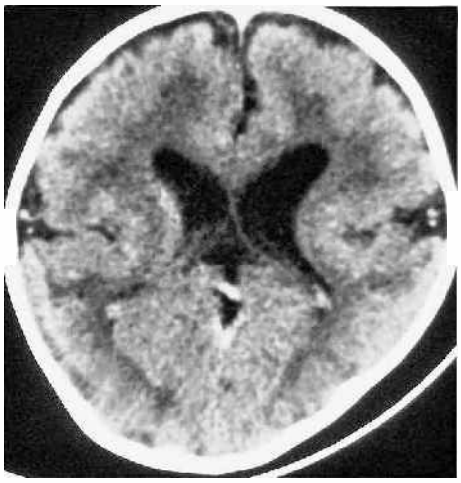

Fig. 2 CT image of the brain shows mild dilatation of the frontal and body of lateral ventricle, third ventricle and fourth ventricle, suggestive of communicating hydrocephalus.

Thurnam published the first report of a patient with ED in 1848,[2] and the term 'ectodermal dysplasia' was coined in 1929 by Weech.[3] Diagnosis of ED is often difficult since any ectodermal derivative may be involved in varying degrees. The number of ectodermal dysplasia syndromes has increased to more than 170.[1] Despite the different genetic causes, their presentations may be very similar. Diagnosis is usually made by clinical observation, often with the assistance of family medical histories. Numerous types of ED syndromes have been described and several classifications exist.

Freire-Maia and Pinheiro proposed the first classification system of ectodermal dysplasia in 1982,[4] with additional updates in 2001.[5] In this classification, "1" indicates hair dysplasia, "2" dental dysplasia, "3" nail dysplasia and "4" sweat gland dysplasia. Based on this classification, more than ten subgroups exist in Freire-Maia and Pinheiro's classification. Since our patient had hair and teeth abnormalities, her ectodermal dysplasia fits into the "1–2" subgroup. Of specific interest is the central nervous system (CNS) malformation found in our case. Although the CNS originates from the embryological ectoderm, reports of associated CNS malformations are rare. Moreover, a comparison of such reports with our case shows little similarity in phenotype and clinical characteristics. For example, Soekarman and Fryns reported internal hydrocephalus with partial hypoplasia of the cerebellum in a severely mentally retarded boy who showed signs of ectodermal dysplasia.[6] Cortes and Lacassie reported a boy with hypoplastic nails, malformed hands and feet, curly hair, small lower teeth and seizures, and the CT imaging showed asymmetry of the cerebral hemispheres.[7] Rushton and Genel described a brother and sister with mental retardation, short stature, hypodontia, and abnormalities of the neurological and endocrine system, with CT imaging showing cerebellar atrophy.[8]

Reports of the triad of developmental delay-hydrocephalus-ectodermal dysplasia are rare. Our observation also highlights the fact that any patient who shows signs of ectodermal dysplasia with developmental delay should be investigated further for CNS malformations.

Yours sincerely,

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