Case Report

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Linear nevus sebaceous syndrome with lissencephaly

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ABSTRACT

Linear nevus sebaceous syndrome is a congenital neurocutaneous syndrome comprising of nevus sebaceous, seizures and mental retardation. We report a 6 month old female child with a linear nevus sebaceous over the face and scalp, whose brain imaging revealed ipsilateral lissencephaly with hemimegalencephaly. Lissencephaly is rarely reported in this syndrome.

Keywords: Lissencephaly, Hemimegalencephaly, Linear nevus sebaceous syndrome

INTRODUCTION

Linear nevus sebaceous syndrome is a rare neurocutaneous disease characterized by a midline facial skin lesion (linear nevus sebaceous), seizures and mental retardation. Here we report a case of female child with linear nevus sebaceous syndrome. This diagnosis was confirmed by the presence of typical cutaneous lesions, seizures, and brain images 6 months after birth.

CASE REPORT

A 6 month female child presented with progressive increase in head size since birth, multiple episode of seizure since 2 months of age and a pigmented nevus on the right face and neck. There was no family history of skin disorder, craniofacial anomalies or neurological disease. Child was a term vaginal delivery born to a primigravida mother whose pregnancy was unbooked and unsupervised with no antenatal ultrasounds. Baby did not cried immediately after birth and was hospitalized for 4 days in a neonatal unit. Ultrasound brain at that time revealed dilated right ventricle suggestive of obstruction at level of foramen of monro. Patient did not undergo any further investigation and left the hospital against medical

advice. At 2 months, child started having multiple episodes of seizure for which medications were given only for 15 days though the seizures continued.

On examination child weighed 7kg, had a head circumference of 46cm (>2SD). A cutaneous abnormality of the face in the form of linear, well-demarcated, raised, brownish and hairless plaques located on the right side of the forehead, the face, chin, upper lip, and right ear was noted. A similar lesion was present over the midline of the neck and the anterior chest wall (Figure 1). There was one cafe-au-lait spot was present over left thigh, no other hypo or hyper pigmentation lesions were present. Ocular examination revealed sunset sign and bilateral total cataract with posterior synechie in right eye. Otherwise, no limbal mass or colobomas were apparent in the eyes. The child was developmental delayed in all spheres with complete head lag, no social smile, persistent grasp reflex and only cooing sounds at 6 months (motor, social, cognitive and language). Power and reflexes were similar in both upper limbs and lower limbs, without signs of hemiparesis.

A brain sonogram was suggestive of obstruction at foramen of monro with dilatation of lateral ventricles. No

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other congenital anomalies could be identified on ultrasound abdomen. Contrast enhanced computed tomography revealed asymmetrical overgrowth of the right cerebral hemisphere, with precedence of the occipital lobe pushing the midline toward the left side. These observations were compatible with hemimegalencephaly, along with unilateral lissencephaly on the right cerebral hemisphere. Left sided cerebral atrophy was also noted.



Figure 1: Linear nevus on right half.

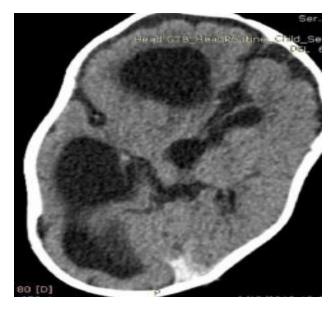


Figure 2: Hemimegencephaly and lissencellaly of the right cerebral hemisphere.

DISCUSSION

The syndrome was first described by Schimmelpenning in 1957, and Feuerstein and Mims in 1962. 2,3 Numerous subsequent reports have confirmed that linear nevus

sebaceous syndrome is frequently associated with ocular, hearing, cardiovascular, skeletal, hepatic, and urinary tract anomalies.⁴ No definite genetic or chromosomal abnormality has been found for this syndrome, except one mosaic dominant lethal gene mutation.⁵ Nevus sebaceous is estimated to occur in 1 per 1000 of newborns and is usually present at birth or in early childhood.⁶ Though the syndromic associations are seen in 1 in 10,000.

The hallmark of this syndrome is nevus which is large, often linear, sharply demarcated and slightly raised plaques. It may have a waxy or pebbly surface on the skin of the head, face and neck. The plaques may extend to the trunk and extremities following the midline of the body, with either a yellow to tan velvety appearance or a darker verrucous look. During infancy and in early childhood, sebaceous glands within the nevus are under developed and no hair follicles are present. Our patient had nevus sebaceous but because of his young age, sebaceous glands within the nevus were undeveloped and no hair follicles were present.

Associated neurological abnormalities, including seizures and mental retardation, are common features of linear nevus sebaceous syndrome. Seizures occur in 44%-90% of patients during their first year of life, with mental retardation or developmental delay occurring in 40%-80% of patients. The head may be enlarged and other associated neurological manifestations include progressive hemiparesis, quadriparesis, hypotonia, reflex abnormalities, gait disorders, diencephalic syndrome, microcephaly and macrocephaly. 1.6

A major finding with MRI is hemimegalencephaly (or unilateral megalencephaly), which is characterized by congenital overgrowth of one cerebral hemisphere ipsilateral to the sebaceous nevus with an increased volume of white matter and dilatation of the lateral ventricle on the affected side. The affected brain has essentially no function and is frequently clinically associated with hemiparesis, early-onset seizures, mental retardation, hemimacrocephaly and severe encephalopathy.6,7 The consistent association hemimegalencephaly is seen in up to 50% of the cases. Other common neurological imaging findings including focal pachygyria, cortical atrophy, hydrocephalus, porencephaly and cerebral neoplasm have been reported.

This syndrome has been reported at birth by Chen et al in which hemimegencephaly has been well defined. There were abnormal findings on sonogram prenatally though the diagnosis was confirmed after birth. Fetal MRI and ultrasound may be required for prenatal diagnosis. Hager BC et al reported a case of 3 month male with facial deformities, cortical blindness whose neuroimaging revealed unilateral lissencephaly, heterotrophic grey matter and hemimegalencephaly. Association with unilateral lissencephaly is rare.

Since nevus sebaceous is commonly seen in new-born period, linear nevus sebaceous syndrome should be considered in these children and search for associated anomalies should be made.

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