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Case Report

A rare case of SCALP syndrome presenting with progressive hydrocephalus and refractory epilepsy successfully treated with ventriculoperitoneal shunting

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ABSTRACT

SCALP syndrome is an exceptionally rare neurocutaneous disorder characterized by sebaceous nevus, central nervous system malformations, Aplasia cutis congenita, limbaldermoid, and pigmented nevus. Neurological manifestations are variable, and severe hydrocephalus requiring neurosurgical intervention has rarely been reported. A 4-year-old girl with developmental delay, epilepsy, and progressive macrocephaly presented with status epilepticus. Seizures were controlled with intravenous levetiracetam and fosphenytoin. Examination revealed a sebaceous nevus over the nape of the neck, multiple areas of aplasia cutis congenita, a left limbaldermoid, and giant congenital melanocytic nevi involving the face and abdomen. Magnetic resonance imaging (MRI) of the brain demonstrated severe communicating hydrocephalus with marked dilatation of all ventricles and diffuse cerebral parenchymal thinning. The constellation of dermatological, ocular, and neurological findings was consistent with SCALP syndrome. The child underwent ventriculoperitoneal shunt placement, following which head circumference decreased by 2 cm at one-month follow-up, with no further seizures. This case highlights severe communicating hydrocephalus as a potentially reversible neurological manifestation of SCALP syndrome. Recognition of its characteristic cutaneous features should prompt early neuroimaging and multidisciplinary evaluation. Timely neurosurgical intervention may prevent neurological deterioration and improve clinical outcomes in affected children.

Keywords: SCALP syndrome, Hydrocephalus, Epilepsy, Aplasia cutis congenita, Sebaceous nevus, Giant congenital melanocytic nevus

INTRODUCTION

SCALP syndrome is rare neurocutaneous disorder characterized by the association of sebaceous nevus, central nervous system malformations, aplasia cutis congenita, limbaldermoid, and pigmented nevus.[1] It is regarded as a phenotypic variant within the epidermal nevus syndrome spectrum and is classified under type III aplasia cutis congenita according to the Frieden classification.[2,3]

The syndrome involves multiple organ systems, particularly the skin, central nervous system, and eyes. Reported neurological manifestations include epilepsy, developmental delay, neurocutaneous melanosis, cerebral infarction, intracranial aneurysms, and structural brain malformations. However, hydrocephalus requiring neurosurgical intervention has been only rarely described.[1-14]

We report a child with SCALP syndrome presenting with developmental delay, refractory epilepsy, and progressive communicating hydrocephalus requiring ventriculo-peritoneal shunting, highlighting the importance of early recognition and multidisciplinary management.

CASE REPORT

A 4-year-old girl presented to the emergency department with generalized tonic-clonic seizures involving all four limbs for approximately 30 minutes. She had a known history of epilepsy diagnosed at 7 months of age and was receiving two antiepileptic medications. On admission, seizures were terminated with intravenous levetiracetam (40 mg/kg) followed by fosphenytoin (20 mg PE/kg).

She was the second child born to a third-degree consanguineous couple. The perinatal period was significant for a 15-day neonatal intensive care unit stay for meconium aspiration syndrome. Following the diagnosis of epilepsy during infancy, the child was subsequently lost to neurological follow-up.

Developmental assessment using the trivandrum developmental screening chart demonstrated global developmental delay.

Physical examination revealed multiple congenital cutaneous and ocular abnormalities. A sebaceous nevus was present over the nape of the neck (Figure 1). A limbal dermoid involving the left eye was confirmed by ophthalmological evaluation (Figure 2). Giant congenital melanocytic nevi were noted over the left side of the face and abdomen (Figures 3). Multiple areas of congenital alopecia over the scalp were diagnosed as aplasia cutis congenita by dermatological assessment (Figure 4). All lesions had been present since birth without progression.

The child had marked macrocephaly with a head circumference of 62 cm (Figure 2), which had progressively increased since infancy. Neurological examination demonstrated generalized spasticity and exaggerated deep tendon reflexes.

Routine hematological and biochemical investigations were unremarkable.

Magnetic resonance imaging of the brain demonstrated gross dilatation of the lateral ventricles, third ventricle (transverse diameter 33 mm), and fourth ventricle (transverse diameter 26.1 mm), with diffuse thinning of the cerebral parenchyma and corpus callosum, consistent with severe communicating hydrocephalus (Figure 5).

The differential diagnosis included sebaceous nevus syndrome and isolated aplasia cutis congenita. However, the coexistence of sebaceous nevus, aplasia cutis congenita, limbal dermoid, giant congenital melanocytic nevus, epilepsy, developmental delay, and hydrocephalus was consistent with SCALP syndrome.

The child underwent ventriculo peritoneal shunt placement. At one-month follow-up, head circumference had reduced by 2 cm (Figure 6), and no further seizures were reported. Surgical correction of the limbal dermoid and eyelid coloboma, along with genetic testing, was planned but could not be performed because of financial constraints.



Figure 1: Sebaceus nevus at the nape of the neck.



Figure 2: Macrocephaly due to hydrocephalus and limbal epidermoid of left eyelid.



Figure 3 (A and B): Pigmented nevus over abdomen and left side of face.



Figure 4: Aplasia cutis congenita over the scalp.

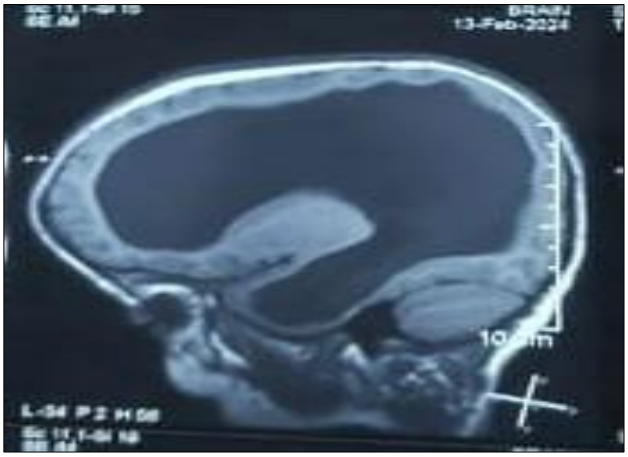


Figure 5: MRI suggestive of bilateral diffuse severe gross dilatation of lateral, 3rd (TD-33 mm) and 4th (TD-26.1 mm) ventricles causing diffuse cerebral parenchymal and corpus callosum thinning.
*TD-transverse diameter

DISCUSSION

SCALP syndrome is a rare and underreported condition, with only a limited number of cases documented to date.

Neurological manifestations reported include epilepsy, developmental delay, structural brain malformations, neurocutaneous melanosis, intracranial aneurysms, cerebral infarction, and status epilepticus.[1-7] Ocular manifestations have also been reported in a small subset of cases.[7,8] Dermatological features, particularly those within the spectrum of aplasia cutis congenita, have been more consistently documented.[1-14]

This patient presented with progressive macrocephaly secondary to severe communicating hydrocephalus. Following ventriculo peritoneal shunt placement, head circumference decreased and seizures remained controlled during follow-up, suggesting that hydrocephalus may represent an important and potentially reversible neurological manifestation of SCALP syndrome. This case highlights the importance of considering progressive hydrocephalus in children with SCALP syndrome presenting with neurological deterioration.

SCALP syndrome shares certain clinical features with sebaceous nevus syndrome but is considered a distinct clinical entity. Sebaceous nevus syndrome is commonly associated with neurological, ocular, and skeletal abnormalities, whereas SCALP syndrome is defined by the characteristic combination of sebaceous nevus, aplasia cutis congenita, limbal dermoid, and giant congenital melanocytic nevus.[4] Other syndromes associated with aplasia cutis congenita, including Adams-Oliver syndrome, trisomy 13, Wolf-Hirschhorn syndrome, Setleis syndrome, Johanson-Blizzard syndrome, and Goltz syndrome, were excluded clinically based on the absence of their characteristic systemic manifestations.[3-9]

An additional concern is the increased lifetime risk of malignant melanoma associated with giant congenital melanocytic nevi, necessitating long-term dermatological surveillance.[7-14] Nevertheless, neurological complications may represent the most urgent and potentially treatable aspect of the disorder.

CONCLUSION

This case expands the neurological spectrum of SCALP syndrome by demonstrating severe communicating hydrocephalus requiring ventriculo peritoneal shunting. Recognition of the syndrome's characteristic cutaneous manifestations should prompt early neuroimaging and multidisciplinary evaluation. Timely neurosurgical intervention may prevent irreversible neurological deterioration and improve long-term outcomes in affected children.

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