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CASE REPORT

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Epidermal Nevus Syndrome with Internal Carotid Artery Occlusion and Intracranial and Orbital Lipomas

SUMMARY: We report a case of epidermal nevus syndrome involving the brain in which there is chronic occlusion of the left distal internal carotid artery resulting in ipsilateral atrophy. Orbital and cerebellopontine angle cistern lipomas and a wide cortical developmental malformation are associated with the condition. We present MR imaging findings of a patient and discuss features in the context of other neurocutaneous diseases.

The epidermal nevus syndrome (ENS) constitutes a group of congenital neurocutaneous disorders characterized by epidermal nevi in association with cerebral, ocular, skeletal, and, sometimes, cardiac and renal abnormalities. Developmental central nervous system problems such as neuronal migration abnormalities, vascular malformations, agenesis of the corpus callosum, Dandy-Walker syndrome, myelomeningocele, Arnold-Chiari malformation, and tumors have been reported in association with this syndrome. Mental retardation, seizures, and focal neurologic deficits are the most common neurologic clinical features.

Case Report

A 27-month-old boy presented with a left-sided epidermal nevus involving his left torso, extremities, neck, and face. The nevus was a brown maculopapular hyperkeratotic plaque and was diagnosed as epidermal nevus by a skin biopsy. The boy was the product of an uncomplicated term pregnancy born by vaginal delivery. The mother and father of the patient, as well as the rest of the family, were free of this syndrome. Findings of a physical examination showed severe failure to thrive (weight and height both less than the third percentile), left frontoparietal alopecia, colobomas of the eyelid, a membrane on the conjunctiva, high soft palate, hyperkeratosis of the soft tissue of left palate, and hypertrophy of the gum. He could not sit and had poor head control and hypotonicity in the lower extremities. A patent ductus arteriosus was present. Findings of abdominal sonographic examination were normal.

A multilobulated circumferential peribulbar intraorbital mass with supraorbital extension was observed on MR imaging with a signal intensity similar to that of subcutaneous fat on all pulse sequences. The eyeball had a bizarre shape with an inferiorly dislocated lens (Fig 1A, -B). Ipsilaterally, there was a lipoma in the cerebellopontine angle cistern, and the subcutaneous fat over the left frontotemporoparietal region was diffusely thicker than that of the right side (Figs 1C and 2C). Left cerebral hemiatrophy was present with dilated lateral ventricle and hemispheric sulci. There was a wide cortical developmental malformation over the left temporo-occipital area (Fig 2A-C).

The left internal carotid artery (ICA) was very thin in caliber in all intracranial segments, tapered at the supraclinoid segment, and occluded at the bifurcation into the left middle and anterior cerebral

artery (Fig 3). Thin basal collaterals in the left sylvian fissure and basal cisterns were observed on T2-weighted turbo spin-echo (TSE) imaging (Fig 2B).

Discussion

ENS was first described in 1968 by Solomon et al.⁶ Many authors believe that it is not a distinct clinical entity but includes several different syndromes and that Proteus syndrome, congenital hemidysplasia with ichthyosiform erythroderma and limb defect (CHILD) syndrome, Schimmelpenning syndrome, nevus comedonicus syndrome, pigmented hairy ENS, and phacomatosis pigmentokeratolica can be grouped under this designation.^{7,8}

ENS is a rare sporadic neurocutaneous overgrowth syndrome characterized by the association of epidermal nevi with central nervous system (CNS) and/or skeletal abnormalities. CNS involvement in ENS is estimated to occur in 50% to 70% of patients. Ocular, cardiovascular, genitourinary, oral cavity, and skin anomalies, other than epidermal nevi, have likewise been observed, including various cutaneous or noncutaneous tumors.^{1,4,8}

MR imaging is the preferred technique of structural imaging for ENS because of its high tissue contrast.² Among CNS abnormalities, unilateral hemimegalencephaly is the most striking finding and is typically ipsilateral to the skin lesions, which are distinctive and usually found on the scalp, face, and/or the neck.^{2,5,9} The classic neurologic variant consists of hemimegalencephaly, facial hemihypertrophy, and pachygyria or polymicrogyria. Hydrocephalus, heterotopias, cortical atrophy, and hamartomas or tumors such as astrocytoma and gliomatosis cerebri have been observed.^{3,4,9,10} Reported vascular abnormalities are rare and include infarction, cortical calcification, and vascular dysplasia.⁹

Approximately 35% to 70% of patients have ocular involvement. The most frequent abnormality is involvement of the eyelids and conjunctiva. Optic nerve hypoplasia, microphthalmia, macrophthalmia, corneal opacities, and cataracts are other abnormalities commonly reported.^{1,8,9} In our patient, lipomatous coloboma of the eye accompanied the CNS lesions.

The CNS lesions were all on the left side of the hemispheric, ipsilateral to the epidermal nevus and ocular abnormality in the present patient; however, there was atrophy most probably resulting from vascular insufficiency due to occlusion of ICA instead of hemimegalencephaly, which is the most frequently reported abnormality in the ENS and other related

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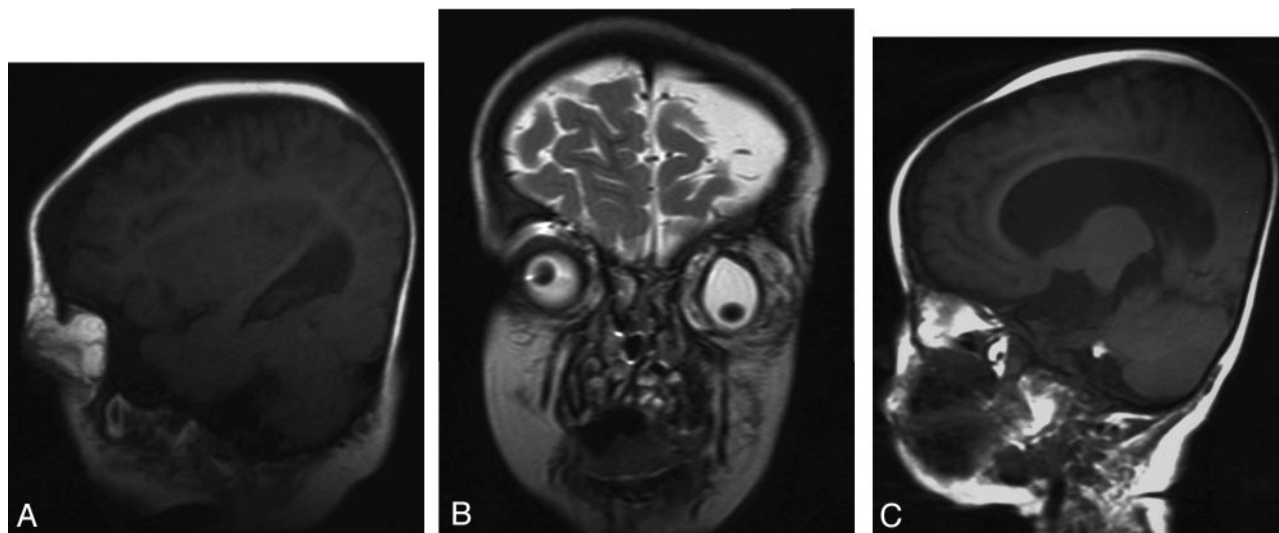


Fig 1. Sagittal T1-weighted spin-echo (TR/TE, 400/9.1 ms) (A) and coronal T2-weighted TSE (TR/TE, 3500/100 ms) images show lipomatous coloboma of the left eye (B). On the sagittal T1-weighted spin-echo (TR/TE, 400/9.1 ms) image of the medial brain, a cerebellopontine angle cistern lipoma is seen (C).

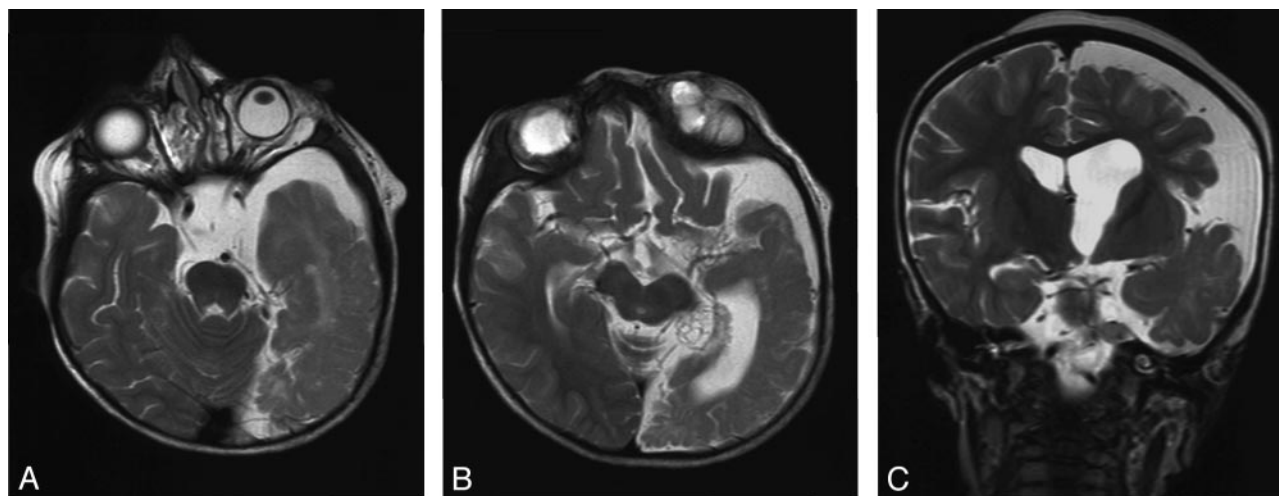


Fig 2. A, Left cerebral hemiatrophy with cortical developmental malformation is seen on the transverse T2-weighted TSE (TR/TE, 3500/100 ms) image. Thin caliber of the left ICA is also observed.

B, T2-weighted TSE (TR/TE, 3500/100 ms) transverse image from a higher section shows the collaterals in the basal cisterns.

C, Coronal T2-weighted TSE (TR/TE; 3500/100 ms) image shows atrophy of the left cerebral hemisphere with dilated lateral ventricles and temporal cortical dysplasia. Also, subcutaneous fat is diffusely thicker over the left frontotemporoparietal cranium.

neurocutaneous disorders. Hemiatrophy and abnormal cortical development on the same side may suggest intrauterine occlusion of the ICA, preventing normal neuronal migration, thus cortical organization and neuronal proliferation. Whereas seizures are one of the most frequently presenting symptoms in patients with ENS, they were absent in this patient. However, active bilateral epileptiform discharges were observed on electroencephalogram, and the patient was put on phenobarbital therapy.

To our knowledge, Moyamoya phenomenon has not been reported in ENS. Moyamoya phenomenon is a rare progressive cerebrovascular occlusive disease characterized by parenchymal collateralization. Many abnormalities may result, depending on the stage of the brain development at the time of vascular insult. The typical findings of Moyamoya phenomenon on brain MR imaging are stenosis or occlusion of the distal ICA and Moyamoya vessels with signal intensity voids in

the basal ganglia, collaterals around and distal to stenotic segments, and ischemia, infarction, atrophy, and ventriculomegaly. The phenomenon is associated with other underlying conditions such as neurofibromatosis, which is notorious for vascular stenoses; Down syndrome; Sjögren syndrome; and previous cranial irradiation.¹¹

Booth and Rollins⁹ have reported intraspinal lipoma in ENS; however, intracranial lipoma has not been reported. In our patient, left orbital and left cerebellopontine angle cistern lipomas were present. In the literature, ENS has been noted to have features that overlap with those of Proteus syndrome, encephalocraniocutaneous lipomatosis, congenital infiltrating lipomatosis regarding facial and intracranial lipomas, and neuronal migration anomalies, though hemimegalencephaly is usually associated with these cases.^{4,12,13} Encephalocraniocutaneous lipomatosis is a neurocutaneous syndrome characterized by unilateral lipomas of the cranium, face, and neck.

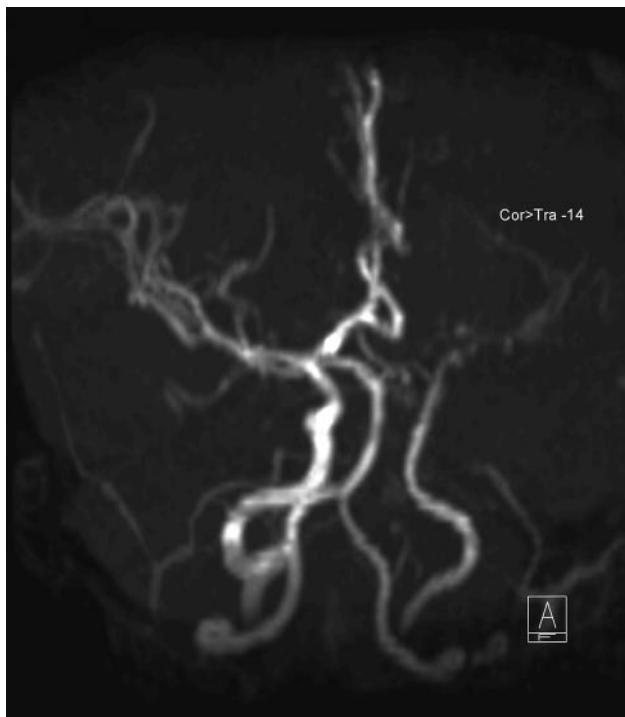


Fig 3. Time-of-flight MR angiogram (TR/TE: 39/3.26) shows thin-caliber left ICA in all intracranial segments, with a tapering end and occlusion of the artery at its bifurcation.

Ipsilateral ventriculomegaly; intracranial lipomas, which are generally believed to be congenital lesions of the meninx primitive rather than neoplasms; dysplastic cortex; and corticopial calcifications are characteristically associated with the syndrome.^{4,11} Similar findings have also been reported in ENS. The presence of intracranial and orbital lipomas may link encephalocraniocutaneous lipomatosis to our patient with ENS. In addition, corticopial calcifications and abnormal pial enhancement have been described in both syndromes but not observed in our patient.¹⁴

There is considerable clinical overlap among many of these syndromes, such as neurofibromatosis, Sturge-Weber syndrome, tuberous sclerosis, Von Hippel-Lindau syndrome, McCune-Albright syndrome, encephalocraniocutaneous lipomatosis, and congenital infiltrating lipomatosis. The pathogenesis of ENS is subject to speculation, yet several facts support the theory of lethal genes surviving by mosaicism. The clinical heterogeneity of the disorders can be explained by different mutations of the same gene or differences in the tem-

poral occurrence of mutations, leading to different patterns of chromosomal mosaicism. The theory of somatic mosaicism explains the sporadic occurrence of ENS, the variability of clinical presentation, and the fact that all ENS are mosaic phenotypes.⁴ The overlapping features of ENS and encephalocraniocutaneous lipomatosis in this patient may be the result of similar chromosomal aberrations.

Conclusion

Intracranial lipoma in ENS is reported for the first time with this patient. ENS shares clinical and imaging findings with other neurocutaneous disorders such as vascular stenoses, neuronal migration abnormalities, and lipomas of the CNS, face, and neck and should be included in differential diagnosis in the presence of these disorders.

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