Leptomeningeal angiomatosis and aplasia congenita of the scalp

Case report

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This 9-year-old boy had two circular areas of aplasia congenita of the scalp in the territory of the first division of the trigeminal nerve and an ipsilateral parieto-occipital leptomeningeal capillary venous angioma. He had an associated giant aneurysm of the distal posterior cerebral artery on the same side as the vascular malformation. The patient had a long history of seizures and presented with intracerebral bleeding. This case may be considered as a variant of Sturge-Weber syndrome. Further observations must be added to consider this entity as a separate neurocutaneous disease.

KEY WORDS: aplasia cutis congenita, cerebral aneurysm, congenital scalp defect, intracranial hemorrhage, neurocutaneous syndrome, Sturge-Weber syndrome

Five entities are usually grouped under the category of neurocutaneous syndromes: 1) multiple neurofibromatosis (von Recklinghausen's disease); 2) encephalotrigeminal angiomatosis (Sturge-Weber disease); 3) tuberous sclerosis; 4) von Hippel-Lindau disease; and 5) ataxia telangiectasia. Another entity (the linear nevus sebaceous syndrome), characterized by the triad of midline linear nevi, mental retardation, and seizures, and, in addition, nonfunctioning major venous sinuses and leptomeningeal hemangioma, exhibits important similarities to the Sturge-Weber syndrome. It is now considered as a separate neurocutaneous disease.

We report the case of a patient who had an intracranial malformation typical of an encephalotrigeminal angiomatosis (leptomeningeal venous-capillary angiomatosis) without the vascular anomaly of the skin (port-wine nevus) usually found in this disease. In our case, the skin defect was represented by two circular areas of scalp aplasia on the left side of the forehead. Aplasia congenita of the scalp is the absence of epidermis, dermis, and sometimes subcutis, dura, and bone from one or more circumscribed areas. This subject was recently reviewed by Muakkassa, et al., and will not be discussed in detail.

Our patient belongs to Group 1 of congenital defects of the scalp (partial thickness defects) described by Kosnik and Sayers and Lynch and Kahn. This group includes those patients with small and sharply demarcated circular defects of the scalp, 2 cm or less in diameter. We do not know if the entity we have observed represents a variant of the Sturge-Weber disease or a separate neurocutaneous syndrome. Although further observations must be described to warrant classifying this association as a separate neurocutaneous disease, the presence of congenital aplasia of the scalp should suggest the possibility of a concomitant leptomeningeal angiomatosis.

Case Report

This 9-year-old boy was admitted to our hospital because of the sudden onset of headache, vomiting, and right hemiparesis. The medical history revealed that he had been born with two small alopecic areas on the left side of the forehead. Seizures were present since infancy. Gestation and delivery were reported as normal. No family history of unusual skin abnormalities was elicited.

Examination. Neurological examination disclosed...
a right-sided hypotonic hemiparesis. Homonymous hemianopsia was present on the right. There were two bald slightly depressed circular areas, about 2 cm in diameter, on the left side of the forehead in the territory of the first division of the trigeminal nerve (Fig. 1). Ophthalmological examination was normal. Skull x-ray films revealed “tram-line” calcifications in the left parieto-occipital region (Fig. 2). The bone underlying the scalp defect was normal.

Operation. A left parieto-occipital craniotomy was performed. The leptomeninges were thickened, opacified, and contained a mass of blood vessels. The left occipital lobe and a small portion of the left parietal and temporal lobes were resected. The giant aneurysm of the posterior cerebral artery was clipped and excised.

Pathological Examination. Microscopic examination revealed extensive venous and capillary angiomatosis of the leptomeninges. The underlying cortex showed several zones of calcification. Biopsy of the scalp defect revealed total absence of the skin appendages (such as hair follicles and sebaceous glands), with a layer of flat epithelial cells covering the dermis.

Postoperative Course. The postoperative course was uneventful. At dismissal 3 weeks after surgery, the right hemiparesis was unchanged. The patient has had no seizures since the operation.

Discussion

The most frequent cerebrovascular anomaly in encephalotrigeminal angiomatosis is represented by a capillary venous angioma of the leptomeninges, often located in the posterior parietal and occipital re-
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Fig. 4. Left: Left carotid angiogram, lateral view, showing a segmental dysplasia of a peripheral branch of the left middle cerebral artery (arrow). Right: Vertebral angiogram, anteroposterior view, showing the giant aneurysm of the left posterior cerebral artery (distal portion). The vessel is diffusely narrowed.

Poser and Taveras grouped under atypical Sturge-Weber syndrome those patients with some type of vascular anomaly of the skin of the face other than the typical port-wine nevus. The facial anomalies in these cases consisted of raised hemangioma, enlarged veins involving the trigeminal area, and hyperpigmentation of the skin in the distribution of the trigeminal nerve.

Besides leptomeningeal venous angioma, cerebrovascular anomalies in patients with Sturge-Weber syndrome include: true arteriovenous malformations, arterial thrombosis, anomalies of veins and venous sinuses, abnormalities within the territory of the external carotid artery, and miscellaneous abnormalities such as subdural hematoma. Aneurysms were never found in association with cerebral leptomeningeal angiomatosis. Extensive dysplasia with segmental narrowing and dilatation of the peripheral leptomeningeal arteries was observed by Hilal, et al., in patients with Sturge-Weber syndrome. Although intracranial bleeding “appears to be a fairly unusual complication” of Sturge-Weber syndrome, the presence of such an arterial dysplasia may constitute a basis for aneurysm formation and possible intracranial hemorrhage in the affected patients, as our case demonstrates.

Several factors seem to indicate a close relationship between the Sturge-Weber disease and the disorder we have observed. These factors include the pathological findings, the typical parieto-occipital location of the cerebrovascular malformation, the intracranial lesion on the same side as the anomaly, and the scalp defect in an area supplied by the trigeminal nerve.

Kjaer believes that some scalp defects might result from spontaneous intrauterine involution of a hemangioma. It is possible that the skin defect in our case represents the healing of a “vascular lesion” similar to lesions found in cases of Sturge-Weber syndrome. Thus, aplasia of the scalp may represent the “equivalent” of the typical facial nevus. Another hypothesis may be postulated. Since the skin near the vertex is supplied by blood from the primitive meninges, a maldevelopment of the vascular area of the superficial bed normally supplying the meninges, as occurs in Sturge-Weber disease, might lead to concomitant leptomeningeal angiomatosis and primary scalp defect.

Associated cerebral anomalies reported with congenital scalp aplasia include hydrocephalus, meningocele and encephalocele, arhinencephaly, hydranencephaly, and unconfirmed defects in early development of the telencephalon. The simultaneous occurrence of leptomeningeal angiomatosis with congenital scalp defects has not been described previously. Due to the fact that deposition of calcium within a leptomeningeal angioma may not be demonstrated in the very young, but may increase with time, control skull x-ray films and/or CT scanning should be performed as patients showing a congenital scalp defect grow older.
References


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