Megalencephaly Due to Impaired Cerebral Venous Return in a Sturge-Weber Variant Syndrome

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Abstract

An infant with a Sturge-Weber variant syndrome developed progressive megalencephaly and eventual hydrocephalus, which required shunting. Cerebral angiography revealed absence of the deep cerebral venous system and the development of abnormal drainage channels via the periorbital veins. It is postulated that the abnormal enlargement of the brain was due to the impaired venous return. Resistance of the brain to continued expansion may have caused an increase in hydrostatic pressure and the development of hydrocephalus. (*J Child Neurol* 1986; 1:115–118)

The classic Sturge-Weber syndrome consists of a unilateral facial hemangioma, ipsilateral leptomeningeal angiomatosis, and contralateral hemiparesis and seizures. Many patients manifest variations of these three findings and may have additional features (such as glaucoma) or occasionally lack one of the three classic features.[1] However, hydrocephalus and megalencephaly have not been described in Sturge-Weber syndrome. The postulated defect underlying the neurological manifestations of Sturge-Weber syndrome is developmental aberrations of the cerebral vascular system with major anomalies prominent in the cerebral venous system. Studies of the angiographic findings in patients with Sturge-Weber syndrome[2–4] have revealed capillary-venous angioma, lack of superficial cortical veins, non-filling of dural sinuses, tortuosity and enlargement of deep subependymal and medullary veins, and veins that course through the brain in unusual locations. Bentson et al[2] suggested that the abnormalities were due in part to the absence or nonfunctioning of cortical veins with collateral flow centrally to the subependymal veins.

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Case Report

We have followed a 26-month-old child with features of Sturge-Weber syndrome, including a nevus involving face and trunk, unilateral glaucoma, and mild developmental delay. Seizures, hemiparesis, and calcifications on computed tomographic (CT) scan have not been noted. Progressive macrocephaly has been present since 2 months of age. The head circumference has increased from the 90th percentile until its present size of 56.5 cm, which is well above the 98th percentile. Prominence and progressive distension of the periorbital veins, bilaterally, have been noted. CT scan of the head initially revealed megalencephaly, but serial examinations have demonstrated a slow but definite increase in both the intra- and extra-axial cerebrospinal fluid (CSF) spaces as well as the brain parenchyma (see Figure 1). Cerebral angiography revealed markedly dilated superficial cortical veins with absence of the deep Galenic venous system (see Figure 2). Drainage was centrifugally and anteriorly to the cavernous sinus and hence to the ophthalmic veins and by anastomosis to the facial veins. A ventriculoperitoneal shunt was inserted at 15 months of age because of progressive hydrocephalus. In the 10 months since shunting, the head size has not changed. The periorbital venous distension has decreased. His development is at the 18 to 20 month level. A CT scan performed at 25 months of age revealed normal-size ventricles, extra-axial fluid collections over the hemispheres, and slight increased prominence of the superficial cortices of the hemi-

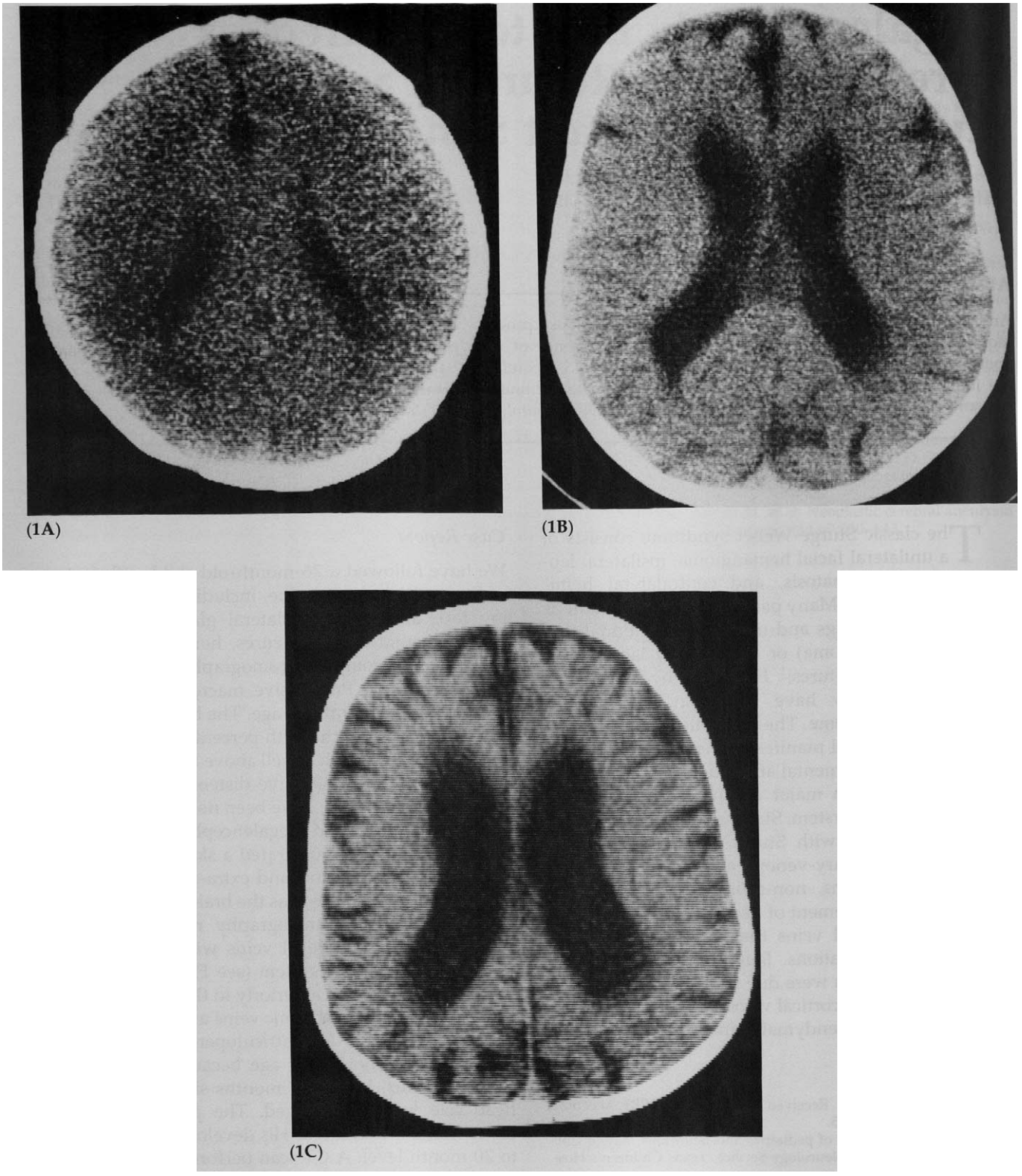

FIGURE 1
Progressive dilatation of CSF spaces at various ages; (**A**) 1 week, (**B**) 5 months, and (**C**) 14 months.

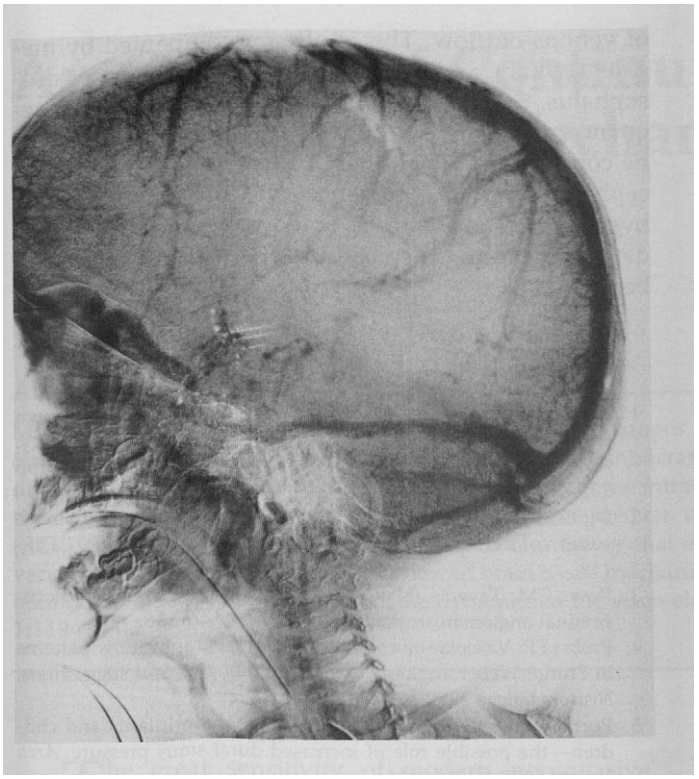

FIGURE 2
Cerebral angiography performed at 15 months of age demonstrating absence of deep venous system and development of large collateral veins about the orbit.

spheres noted on the contrast-enhanced examination. The latter finding was of unknown significance.

Discussion

The mechanism of the megalencephaly and eventual hydrocephalus may be related to the lack of functioning major intracranial venous channels. Portnoy and Croissant[5] speculated that mild elevation of the sagittal sinus pressure in the presence of open sutures results in the increased venous pressure being distributed equally to the CSF and parenchyma. The intracranial volume increases and the increased volume is distributed among the ventricles and subarachnoid, blood, and interstitial spaces. Thus, mild hydrocephalus occurs, as well as an apparent increase in brain parenchymal volume. The additional fluid load can be accommodated while the skull is expansile. With continued fluid overload from impaired venous outflow and eventual resistance to unimpeded expansion of the brain, the ventricles begin to enlarge. Closing of the sutures may change the transmission of the pressure to a hydrostatic situation, ie, equal forces in all directions, and limit further expansion of the ventricles resulting in compensated hydrocephalus.

The causal relationship between dysfunction of the major intracranial venous drainage system and hydrocephalus is supported by several other studies. Shapiro and Shulman[6] reported two infants with facial angiomata, macrocrania, and intracranial venous anomalies. In one 7-month-old infant, the jugular system was obstructed and venous blood flowed to the cavernous sinus, ophthalmic veins, and facial veins. This patient had marked communicating hydrocephalus. The second patient, also 7 months old, had mild ventricular dilatation and no filling of the jugular venous system during cerebral angiography. The dural sinuses communicated with scalp veins. Further support for the relationship between hydrocephalus and abnormalities of the cranial venous system is the presence of communicating hydrocephalus in several patients with achondroplasia associated with elevated sagittal sinus pressure and obstruction of venous outflow at the jugular foramen.[7,8]

Stephan et al[9] noted a group of ten patients with macrocephaly in association with unusual cutaneous angiomatosis, Klippel-Trenaunay-Weber syndrome alone or in combination with Sturge-Weber anomaly, and cutis marmorata telangiectasia congenita. The rate of postnatal head growth tended to be excessive in early infancy and then decreased to approximately parallel the normal growth curve. This clinical course is compatible with speculation by Portnoy and Croissant[5] that closing of the cranial sutures results in the increased pressure secondary to venous obstruction changing from a nonhydrostatic to a hydrostatic situation and thereby decreasing the rate of head growth. Thus impaired cerebral venous circulation may have been responsible for the macrocephaly noted in these patients.

Sainte-Rose et al[10] recently demonstrated that elevated sagittal sinus venous pressure can be the cause of hydrocephalus rather than secondary to increased intracranial pressure in infants with sinography-demonstrated anatomical interruption of the draining sinuses. They simultaneously measured the pressure in the sagittal sinus, ventricle, and jugular bulb before and after removing enough CSF to lower the intracranial pressure to zero. In infants without anatomical interruption of the draining venous system, both the sagittal sinus venous pressure and the intracranial pressure fell simultaneously. However, in infants with narrowing or occlusion of the sigmoid sinuses only the intracranial pressure fell. Thereby they proved that the sagittal sinus venous pressure was not elevated secondary to the increased intracranial pressure in these infants. A venous bypass graft was inserted between the transverse sinus and the jugular vein in an infant with craniostenosis and hydrocephalus. Both the intracra-

nial pressure and ventricular size decreased postoperatively. This strongly suggests that the impaired venous return was responsible for the hydrocephalus. Their patients also developed extensive venous collateral circulation in the occipital region and/or the scalp.

The findings in our patient demonstrate that hydrocephalus may occur secondary to impaired function of the deep venous draining system, as well as secondary to abnormalities in flow of the sagittal sinus. In the former situation, the site of collateral venous channels tends to be anterior about the orbit, as compared to the occipital region and scalp when the major superficial sinuses are involved.

The exact neurocutaneous syndrome affecting our patient may be questionable. We prefer to consider it an example of the Sturge-Weber variant because of the cutaneous hemangioma and the glaucoma. The possibility of this case being an example of the Klippel-Trenaunay-Weber syndrome should be considered; however, varices and osteohypertrophy, hallmarks of that disorder, are missing, although they may develop after infancy. Occasionally the two syndromes can occur simultaneously, as noted in the patients reported by Stephen et al.[9,11] The reduction in ventricular size after shunting and the cessation of abnormal head growth indicates that the macrocrania was not due only to megalencephaly, but that hydrocephalus was also present.

Our patient appears to have an extremely rare complication of Sturge-Weber syndrome—progressive megalencephaly with the subsequent development of hydrocephalus. The mechanism responsible for this sequence of events may involve disturbances in drainage of major intracranial venous systems. Infants with macrocephaly who have abnormally enlarged veins about the cranium should be suspected of having impaired intracranial venous drainage (through normal pathways) and the development of alternate pathways of venous outflow. This is often accompanied by megalencephaly and associated communicating hydrocephalus. Serial CT scans will help diagnose this sequence of events. Ventriculoperitoneal shunts should be considered for treatment of the progressive hydrocephalus. Further experience with the use of venous bypass grafts needs to be obtained before recommendations regarding the usefulness of this procedure can be made.

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