"Cutis marmorata" (that is, marble skin) is a descriptive term of a condition characterized by a red or pink reticulated covering of confluent areas of the skin created by a vascular pattern. Other significant congenital anomalies occur in 18.8% of affected patients.2 The distribution of cutis marmorata is equal between sexes. The skin condition tends to improve over time, and the overall prognosis is typically good.2 In the three cases presented in their paper in this issue of Journal of Neurosurgery: Pediatrics, Conway and co-workers describe a specific syndrome that includes cutis marmorata congenita, a very large head (macrocephaly), syndactyly of toes, hypotonia, and mental retardation. In terms of mental retardation, the prognosis associated with this manifestation is substantially worse than that associated with general cutis marmorata. A significant number of patients have severe and complex congenital anomalies, and sudden death is relatively common.

The nomenclature and classification of this and related conditions are complex and somewhat confusing. The term “phakomatosis pigmentovascularis” is an encompassing description of the conditions associated with cutis marmorata combined with a large number of other congenital anomalies and syndromic conditions.6,10 With the exception of a few patients, this form of phakomatosis is a sporadic condition. It has been thought to be related to a confusing condition called “twin-spotting.” This condition implies postzygotic mosaicism in which various cutaneous areas have distinctly different genetic bases.4 Most, if not all, of the abnormalities associated with these conditions are of vascular origin and include Sturge–Weber syndrome,1,7 moyamoya syndrome,32 arterial occlusion,11 and venous occlusion.8

Conway and coauthors describe three patients with macrocephaly–cutis marmorata telangiectatica congenita (M-CMTC) who developed an acquired form of the Chiari malformation Type I (CM-I). Concurrent with herniation of the cerebellar tonsils, the three patients developed hydrocephalus that required shunt treatment. These cases raise numerous questions about cause and effect. The authors postulate that this condition was part of the brain overgrowth that is known to be associated with M-CMTC, in which the cerebellum outgrows the volume of the posterior fossa. If the cerebellar overgrowth represented the primary event in the development of the hindbrain herniation, the occipital bone would be expected to distend to accommodate the overgrowth as it does for the increased volume of cerebrospinal fluid (CSF) associated with conditions such as mega cisterna magna.11

In other genetically determined conditions such as craniofacial disorders associated with hindbrain herniation, the Chiari malformation is acquired. That is, similar to the cases reported by Conway and colleagues, the malformation is not present at birth. In the context of craniofacial syndromes, the cause of the hindbrain herniation relates both to the inability of the occipital bone to distend due to bilateral lambdoid craniosynostosis and to venous hypertension related to constriction of the jugular foramina.3,5

As in the three cases presented here, these patients tend to have a history of hydrocephalus that requires shunt treatment. How does all of this information fit together? It is known that M-CMTC is associated with venous anomalies. It is also known that venous outflow obstruction during infancy or the prenatal period can lead to macrocephaly and hydrocephalus. The macrocephaly is related to an increase in cerebral venous volume as well as an increase in the pressure required to absorb CSF. Therefore, hydrocephalus is usually associated with some distension of the cortical subarachnoid spaces.

In my experience, one of the patients who presented to our institution with M-CMTC had a rapidly enlarging head. In terms of personal, social, and fine motor functions, the infant developed normally but showed delays in gross motor function at 7 months. Magnetic resonance (MR) venography (Fig. 1) showed a complete absence of the right transverse sinus and jugular vein and marked stenosis of the left transverse sigmoid junction. These features accounted for both the macrocephaly and the hydrocephalus. Analysis of our patient and the patients described by the authors raises the question of the value of shunt placement in patients with M-CMTC. Unfortunately, the developmental delays associated with M-CMTC rarely improve with shunt insertion.
is uncertain if a large head is always problematic enough to justify this type of treatment.

The word “malformation” in the term CM-I implies abnormal development of the cerebellar tonsils. In the cases described by the authors, as well as those in many other reports, this condition is considered an acquired phenomenon. Therefore, the term malformation is inappropriate. In the United Kingdom, this abnormality is usually described as hindbrain herniation, which seems more appropriate than the term malformation. In the cases described here, the venous hypertension presumably caused the increase in volume and turgor of the cerebellum as the skull of the occipital bone thickened. The loss of distensibility then led to chronic tonsillar herniation.

What are the important points that neurosurgeons should glean from this study? First, this condition is in the process of being described and studied. Therefore it is unlikely to be recognized by neurosurgeons first confronted with these infants. Recognition of M-CMTC improves counseling for families and allows a treatment decision to be made more rationally. The families can be warned about the possible need for shunt placement, the probability of the infant developing hindbrain herniation, and the possibility of sudden death despite appropriate treatment. The correct diagnosis will also help determine whether patients’ cognitive outcome might improve with shunt insertion or whether this treatment is best avoided.

References


RESPONSE: We would like to thank Dr. Rekate for his thoughtful comments and review of various clinical aspects of M-CMTC, as well as sharing his experience in treating a patient with this syndrome. Dr. Rekate emphasized numerous interesting points, and we thank the editor for allowing us to respond.

We appreciate Dr. Rekate’s analysis of the literature pertaining to the classification of this condition. First, we thoroughly agree with his observation that the nomenclature and classification of this vascular anomaly and related disorders are difficult. We have the same opinion that isolated CMTC and M-CMTC represent very different clinical conditions despite the fact that both disorders have the same vascular anomaly. The skin in a patient with M-CMTC often has a doughy, thickened appearance resembling a genetic connective tissue disorder (which is not seen in isolated CMTC). Second, we emphasize that another key feature of M-CMTC is the presence of generalized overgrowth that is often disproportionate, affecting both the head and the body and distinguishing it from other overgrowth disorders and isolated CMTC (as well as Klippel–Trenaunay syndrome).