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Research Letter

Intracranial Anomalies Detected by Imaging Studies in 30 Patients With Apert Syndrome

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To the Editor:

Brain imaging was reviewed in 30 patients with Apert syndrome (25 male and 5 female, newborn to 37 years) including 17 patients studied by computerized tomography (CT), 1 patient imaged with only magnetic resonance imaging (MRI), and 12 patients who had both CT and MRI. Agenesis of the corpus callosum was present in 2 patients and hypogenesis of the corpus callosum was found in another patient, ventriculomegaly in 27 (89%) patients, and complete or partial absence of the septum pellucidum in 12 (40%) patients. Defects of the septum pellucidum, attributed to erosion secondary to hydrocephalus, were documented in 3 (10%) additional patients. In 13 patients who had MRI, Chiari I malformation was detected in 5 (38%), cerebellar tonsillar ectopia in 5 (38%), and posterior fossae arachnoid cyst in 2 (15%). CT studies also showed abnormal semicircular canals in 21 patients (70%) and jugular foraminal stenosis in 28 (93%). Table I, summarizes all of our findings. Of the patients who had an MRI, five patients had Chiari I malformation and five patients had low-lying cerebellar tonsils. An arachnoid cyst in the posterior fossa was seen in two patients. Abnormal semicircular canals were noted in 21 patients and jugular foraminal stenosis in 28 patients.

Central nervous system anomalies in Apert syndrome have been documented by several authors and discussed by them [Cohen and Kreiborg, 1990; Cinalli et al., 1995, 1998; Renier et al., 1996; Cohen and MacLean, 2000].

TABLE I. Central Nervous System Abnormalities in Apert Syndrome

	Number of cases (n = 30)	Percentage (%)
Non-progressive ventriculomegaly	23	76
Hydrocephalus	4	13
Completely absent septum pellucidum	5	17
Partially absent septum pellucidum	7	23
Deficiency of septal leaflets	3	10
Agenesis of corpus callosum	2	7
Deficient corpus callosum[a]	1	3
Thinning of corpus callosum[b]	4	13

[a]The rostrum is missing.
[b]Secondary to ventriculomegaly.

It has been well established that megalencephaly is a characteristic of all newborns with Apert syndrome. Cohen and Kreiborg [1993] found that six brain weights at different ages were far in excess of the 95th centile. Combining the data of Hanieh

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and David [1993] with that of Cohen and Kreiborg [1990, 1993, 1994] and Cohen and MacLean [2000], distortion ventriculomegaly was present in 38 of 41 infants, increased intracranial pressure was not detected in any of the 41 infants, and progressive hydrocephalus occurred in only 3 of 41 infants (see Table 24-7 of Cohen and MacLean [2000]). Thus, progressive hydrocephalus is uncommon in Apert syndrome (7.3%, n = 41) compared to Crouzon syndrome (25.6%, n = 86), Pfeiffer syndrome (27.8%, n = 18), and cloverleaf skull (extremely common) [Cohen, 2005].

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