CONGENITAL MALFORMATIONS

CLINICAL FEATURES OF CORPUS CALLOSUM AGENESIS

The clinical and genetic characteristics of 63 patients with agenesis of the corpus callosum (ACC) are reported from the Scientific Institute "E Medea," Italy. Of 1753 patients admitted (1998-2001) and having neuroimaging, 63 (3.5%) showed ACC. Mean age was 2 years 7 months (range 1-25 yrs); 39 were male and 24 female. Thirty patients (47%) had complete ACC and 33 (53%) partial agenesis. Associated nervous system malformations were found in 10 (33%) patients with complete ACC (usually affecting the cortex) and in 14 (42%) with partial ACC (involving the posterior fossa). Non-CNS malformations, including craniofacial, cardiac and skeletal, were present in 41 (65%) patients; 21 (33%) with known syndromes (eg Aicardi (2), Sotos (3), tuberous sclerosis (1)). Seven patients had chromosomal abnormalities, and 3 had subtelomeric rearrangements. Mental retardation was present in 52 (83%), neuromotor inpairment in 58 (92%), and epilepsy (35%). ACC is manifested by a broad range of clinical manifestations. (Bedeschi MF, Bonaglia MC, Grasso R, et al. Agenesis of the corpus callosum: clinical and genetic study in 63 young patients. **Pediatr Neurol** March 2006;34:186-193). (Respond: Dr Borgatti, IRCCS "Eugenio Medea", La Nostra Famiglia, Via Don Luigi Monza, 20, 23842 Bosisio Parini (Lecco), Italy).

COMMENT. ACC has various manifestations, and the prognosis is also variable. ACC is an isolated finding in 17% cases. Multiple malformations, CNS and non-CNS, are present in more than two third of cases. A presentation with craniofacial abnormalities (macrocephaly, hypertelorism, depressed nasal bridge) is sufficiently frequent to warrant neuroimaging. A complete ACC is associated with malformations of cortical development whereas partial ACC is more frequently correlated with posterior fossa anomalies.

The autopsy case of a boy with arthrogryposis multiplex congenita, associated with complete ACC and dentato-olivary dysplasia is reported from Japan (Saito Y et al. **Brain & Dev** May 2006;28:261-264).

NEUROCUTANEOUS SYNDROMES AND THE NEURAL CREST

The role of the neural crest as an inducer of neurocutaneous syndromes and associated craniofacial abnormalities is proposed by researchers at the University of Calgary and Alberta Children's Hospital, AB, Canada. Abnormal neural crest differentiation results in the diverse features of these syndromes, including holoprosencephaly, anencephaly, facial hypoplasias, and hypertelorism associated with ACC. The classification of neurocristopathies should be expanded to include neurocutaneous syndromes other than neurofibromatosis, facilitating the identification of genetic mutations and interrelations with embryonic neural crest. (Sarnat HB, Flores-Sarnat L. Embryology of the neural crest: its inductive role in the neurocutaneous syndromes. **J Child Neurol** 2005;20:637-643). (Respond: Dr Harvey B Sarnat, Division of Pediatric Neurology, Alberta Children's Hospital, 1820 Richmond Road SW, Calgary, AB T2T 5C7, Canada).