d Rehabilitation, Medical University of Bialystok, ul Waszyngtona 17, 15-274 Bialystok, Poland).

COMMENT. Spastic diplegia involves the legs more than the arms, and is frequently associated with premature birth. Spastic quadriplegia, the most severe form of CP, affects all limbs, but impairment of motor function is usually more severe in the upper limbs. In the classification of CP by Crothers and Paine (1959), 65% of cases were spastic (19% quadriplegic, 2.8% diplegic, 40.5% hemiplegic), 22% extrapyramidal, and 13% mixed types. Changes in the classification of CP in the years 1954-90 were related to increased survival rates of preterm infants (Hagberg B et al. Acta Paediatr Suppl 1996;416:48-52). Bilateral spastic forms (diplegias, quadriplegias, and extrapyramidal) were most prevalent, and 75% were in preterm and 45% in term infants. In the above Polish study (Kulak et al), 55% of both DCP and TCP cases were born prematurely. In the above Australian study (Badawi et al), the classification of CP in term infants (% of total cases; and with/without encephalopathy) was as follows: spastic quadriplegic (17%;31/12%), spastic diplegic (23%;19/24%), hemiplegic (34%;22/38%), athetotic dystonic (17%;25/14%), and ataxic hypotonic (10%;3/11%).

CONGENITAL MALFORMATIONS

GENETIC VARIETIES OF JOUBERTS SYNDROME

Four families with linkage to two loci for Jouberts syndrome (JS), JBTS1 or JBTS2, and clinical and radiographic correlations for 4 known genetic causes of JS-related disorders (JSRD) are described in a report from University of California-San Diego, La Jolla, CA, and centers in Europe and the Middle East. The clinical manifestations of JS are hypotonia, ataxia, mental retardation, oculomotor apraxia, and dysregulation of neonatal breathing. The molar tooth sign (MTS) is the diagnostic neuroradiological feature of JS, a complex midbrain-hindbrain malformation consisting of cerebellar vermis hypoplasia, deep interpeduncular fossa, and thickened, elongated, maloriented superior cerebellar peduncles. JSRD are characterized by the association of the MTS with abnormalities of other organs, including retinal dystrophy and polycystic and fibrotic kidneys, optic coloboma, polydactyly, liver fibrosis, and other CNS malformations (eg polymicrogyria). JBTS1 and -3 features are restricted to the CNS, whereas JBTS2 involves multiple organs, including kidney, retina, and liver as well as CNS. (Valente EM, Marsh SE, Castori M et al. Distinguishing the four genetic causes of Jouberts syndrome-related disorders. Ann Neurol April 2005;57:513-519). (Respond: Dr Gleeson, University of California-San Diego, Leichtag 332, 9500 Gilman Drive, La Jolla, CA 92093).

COMMENT. In addition to Jouberts, syndromes of cerebellar vermis agenesis or hypoplasia include Dandy-Walker malformation, Walker-Warburg (lissencephaldy, retinal ahnormalities and hydrocephalus), Meckel-Gruber (occipital encephalocele, polycystic kidneys, poydactyly and hydrocephalus), and atypical Dandy-Walker with facial angioma. Some cases of vermian agenesis are associated with urinary excretion of succinyl-purines and pipecolic acid (Bordarier C, Aicardi J. Dev Med Child Neurol 1990;32:285-294; See Progress in Pedeiatric Neurology I, 1991;312-313).