266. ARTHROGRYPOSIS MULTIPLEX CONGENITA DUE TO CONGENITAL MYASTHENIC SYNDROME

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Two children, 6 and 5.5 years, presented in the neonatal period with hypotonia, multiple joint contractures, ptosis, extraocular weakness, bulbar symptoms, and respiratory distress. Symptoms were characterized by fluctuations and episodic exacerbations of weakness necessitating respiratory support. Development in both children has been delayed and independent walking has not been achieved, although 1 child underwent bilateral Achilles tendons lengthening procedures. Biochemical and metabolic investigations, EMG, and nerve conductions, including slow rate repetitive nerve stimulation, were normal. Acetylcholine receptor antibodies in serum were absent. Single-fiber EMG and axonal stimulation revealed prolonged mean jitter in the tibialis anterior and extensor digitorum muscles, with more than 2 abnormal individual jitter values in each muscle. Muscle biopsy demonstrated normal pattern and morphology of muscle fibers, and present histochemical staining for cholinesterase. However, electron microscopy revealed abnormalities in motor endplates. There was atrophy, flattening of the primary synaptic clefts, and paucity of side branches. Both children improved clinically on pyridostigmine therapy. The findings most likely represent one of the postsynaptic abnormalities (i.e., acetylcholine receptor deficiency, or paucity of synaptic folds). Arthrogryposis due to congenital myasthenic syndrome, as diagnosed in our children, has been reported in one previous patient. The diagnosis can be established by the characteristic clinical history, neurologic examination, electrophysiologic and pathologic findings. Clinical improvement can be achieved with anticholinesterase therapy.

267. BRAIN ABSCESS IN CHILDREN AT CHULALONGKORN HOSPITAL

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This study retrospectively reviewed 45 pediatric patients with brain abscesses admitted to Chulalongkorn Hospital between January, 1984 to December, 1993. There is a male to female ratio of 1.5:1. A peak in the age distribution was seen at 4 to 10 years of age. The majority of patients had prolonged headaches, fever, and clinical symptoms and signs of increased intracranial pressure. Abnormal neurologic signs were detected in 80% of patients. Congenital cyanotic heart disease was the most common predisposing factor. The most common organisms isolated from brain abscesses included anaerobic *Streptococci, Staphylococcus aureus,* and Gram-negative bacilli depending on the predisposing factors. Frontal and parietal lobes were the 2 most common sites of abscesses. Investigation by CT yielded 100% accuracy. Treatment included simple aspiration and/or primary excision together with intravenous antibiotics. Mortality rate in this study was 8.8%, which declined significantly compared with the past decade (1974-1983), in which the rate was 30% $P < .05$ (chi square). Brain herniation was the most common cause of death. Although neither the site nor the source of infection was a consistent prognostic factor, the patient's level of consciousness at admission was considered significant.

268. DO DEVELOPMENTAL ABNORMALITIES IN DOWN SYNDROME CONTRIBUTE TO EARLY-ONSET DEMENTIA?

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Anatomic and biochemical characteristics of the brain in Down syndrome (DS) may promote the early development of dementia. It is critical to document these changes from childhood to adulthood. A key feature of the mature DS brain is the prevalence of amyloid plaques and neurofibrillary tangles characteristic of Alzheimer disease (AD). Deposition of amyloid has been observed as early as 12 years of age, but was not detected in patients 2 and 5 years of age. At the earliest stages of senile plaque development, intact, morphologically normal, neurofilament positive neurons are present in amyloid deposits. In the early stages of plaque formation, β-amyloid is not thioflavine positive and there is little astrocytic response. As the DS brain ages and plaques become thioflavine positive, neurons may be lost and reactive astrocytosis is typically present. These postmortem results correspond to antemortem quantitative MRI findings that suggest an accelerated neural loss compared to age-matched controls. In addition to a relatively smaller frontal cortex and hippocampus, and a larger parahippocampal gyrus, DS MRIs had a sharper rate of increased ventricular area and decreased area of temporal lobe structures compared to age-matched volunteers. Examination of factors that contribute to the early onset of pathologic conditions in DS may contribute to an understanding of early maturational changes associated with the disorder as well as AD.

269. STROKE IN WILLIAM SYNDROME

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William syndrome (WS) is a genetic disorder featuring infantile hypercalcemia, elfin-like features, "cocktail party" personality, and cardiovascular abnormalities including congenital heart disease, supravalvular aortic stenosis, supravalvular pulmonic stenosis, ventricular septal defects, patent ductus arteriosus, and hypertension. Despite the frequency and severity of the reported cardiovascular features of WS, the literature does not contain any reports of cerebral infarction. We describe a 19-year-old adolescent with WS who presented with acute stroke. She was the product of a normal pregnancy and delivery who had feeding difficulties as an infant, and hypercalcemia was documented at age 2 years. At age 14 her serum cholesterol was 200-230 and she was placed on a modified diet. At age 16, her blood pressure was 170/105, and an echocardiogram revealed left atrial and ventricular dilatation, mitral valve prolapse, and mitral regurgitation. She was begun on an ACE inhibitor. At age 19, she awakened with weakness which progressed during the day to right hemiparesis, hemianesthesia, and dysarthria. Initial CT showed a 1 cm^2 hypodensity lateral to the left internal capsule and adjacent to the left putamen. No evidence of a coagulopathy was found. Three days later, her examination disclosed right arm plegia. Cranial MRI revealed a 3 × 1.5 cm area consistent with infarct in the left internal capsule and putamen. Transesophageal echocardiography failed to show an embolic source. Cerebral and renal angiograms were performed. Left carotid injection resulted in severe vasospasm requiring treatment with intra-arterial