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FACE ABNORMALITY PREDICTING BRAIN MALFORMATION

Poster

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The severe malformation of the brain known as holoprosencephaly (HPE) is more frequent among embryos than in newborn infants, suggesting that most embryos with HPE succumbed prenatally. Nowadays this malformation should be ruled out by routine prenatal ultrasonography. Nevertheless, HPE could be a postnatal finding and the relationship between facial anomalies and brain malformation might help to reach the diagnosis. It has been suggested that facial anomalies predict brain malformations, and a close correlation between facies and HPE was emphasized. The brain's findings have been correlated with facial anomalies ranging from anophthalmia or cyclopia to midline defects such as median cleft lip or single upper-midline incisor. The authors have had the opportunity to observe a newborn infant with a protuberance in the middle of the nose and a dimple in its centre. The presence of this unusual anomaly prompted ultrasonographic examination of the brain. The coronal scan through frontal lobes showed a relatively small central single ventricle, with a flat roof and a somewhat squared right frontal horn. The third ventricle had been incorporated into the single ventricle. A CT scan confirmed the fusion of frontal horns. This report justifies the performance of an ultrasonography examination in the presence of an even mild midline anomaly of the face.

DYNAMIC POSTUROGRAPHY IN A GROUP OF DYSLEXIC BOYS

Poster

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It is well documented that in a large number of adolescents with dyslexia, one of the frequent 'soft signs' is motor clumsiness. Two groups of dyslexic and non-dyslexic Israeli boys matched for age, gender and IQ underwent dynamic posturography. All participants had above-average IQ and were otherwise healthy. Language, arithmetical calculation, visuomotor perception and fine and gross motor co-ordination were scored according to criteria of Paine and Oppé. A self-assessment and self-appreciation questionnaire was completed by each subject. Posturography disclosed a significant impairment of sensory axial balance in the dyslexic group. Severity of the balance impairment was unrelated to the degree of dyslexia.

ASYMMETRY OF INTERNAL CAROTID BLOOD VELOCITY IN PRETERM NEWBORN INFANTS

Poster

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The object of this study was to decide whether the susceptibility of the left hemisphere to periventricular haemorrhage (PVH) is due to asymmetry of the cerebral blood flow (CBF) and if so, whether this asymmetry is due to patent ductus arteriosus (PDA). Left and right internal carotid CBF velocity (CBFV) and ductal shunting were measured by Doppler ultrasound in 33 preterm infants during their first five days of life. Seven infants developed PVH, 13 developed RDS without PVH and 13 remained healthy. In the infants without PVH, asymmetry was insignificant. The infants with PVH, on the other hand, exhibited asymmetry (to the disadvantage of the left side) which differed significantly from zero and from the infants without PVH. Inclusion of

PDA in the analyses reduced the differences in comparisons with healthy infants, but not with infants with RDS. Inclusion of behaviour, heart rate, global CBFV (right-left average) and capillary pCO₂ did not affect the differences at all. Since all haemorrhages were bilateral, a relationship with the side of haemorrhage could not be demonstrated. Susceptibility to PVH is related to CBFV asymmetry, to the disadvantage of the left side, and this relationship is partially dependent on PDA. To decide whether the susceptibility to PVH applies in particular to the left hemisphere requires a study on a larger scale.

EXTERNAL HYDROCEPHALUS SECONDARY TO SUPERIOR VENA CAVA HYPERTENSION DURING THE NEONATAL PERIOD

Poster

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Three preterm infants born at 29 to 30 weeks of gestational age, perfused with central catheters during the neonatal period, developed a total or partial thrombosis in the superior vena cava system with subsequent bilateral chylothorax in two cases. A progressive macrocephaly was observed at around seven to 10 months corrected age. After clinical neurological and neurodevelopmental evaluations, and further investigations (MRI, non-invasive intracranial pressure), two infants were shunted for external hydrocephalus with good stabilisation of the head circumference. A spontaneous compensated external hydrocephalus was observed in the third infant. The results of an angio-MRI recently performed at 2.5, four and 3.5 years, respectively, suggest a relationship between superior vena cava hypertension and hydrocephalus.

MUSCULAR DYSTROPHIES: WHEN AND HOW TO INVESTIGATE

Invited Lecture

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The term muscular dystrophy refers to a clinical phenotype of progressive muscle weakness, with specific morphological changes (such as variation of fibre size, the presence of degeneration and regeneration, increase in fibrous and adipose tissue) in the muscle biopsy. A variety of clinically and genetically heterogeneous conditions are all characterised by these changes in the muscle, and their distinction relies both on a careful physical examination and on specific diagnostic tests. Two major subgroups can be broadly defined according to the age at onset: congenital types, with onset at birth or within the first six months of age; and progressive, later-onset types, with an onset that varies according to the severity and subtype of the condition. An accurate history, which must include the family history, often provides the clue to diagnosis: information such as the presence of other affected family members and the pattern of inheritance of the condition will often be of considerable help during the diagnostic process. A careful clinical examination of the patient is also essential: take notice of the pattern of muscle involvement, presence of fixed deformities and involvement of organs other than the muscle. Investigations should be divided into screening tests and more sophisticated diagnostic tests. CPK levels (almost but not invariably elevated), muscle ultrasound and electromyography should be performed in all patients. Additional investigations, such as a muscle biopsy, will then be required to define the specific dystrophic subtype. A complete study of the muscle pathology should include immunocytochemistry: the detection of dystrophin, merosin and the dystrophin-associated proteins is an essential step for a precise diagnosis. Genetic analysis, such as the search of dystrophin gene deletions if a Duchenne or Becker muscular dystrophy is suspected, is also of value both for diagnosis and genetic counselling of families with these conditions.