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Infantile Huntington's disease in Mexico's National Institute of Neurology and Neurosurgery. Rasmussen A[1], Alonso ME[1], Macías R[1], Yescas P[1], Ochoa A[1], Dávila G[2]. [1] Neurogenetics Department, Instituto Nacional de Neurología y Neurocirugía. [2] Neurology Department, Instituto Nacional de Pediatría. Mexico City, D.F. Mexico.

Objective: Analysis of the clinical expression and molecular behavior of the CAG repeat expansion responsible for Huntington's disease (HD) in children attending a tertiary care center in Mexico.

Background: Clinical manifestations of Huntington's disease usually appear between the ages of 35 and 45, but there is a number of patients with an early onset of disease: Juvenile HD is defined by an onset before 20 years of age, while Infantile HD is characterized by appearance of the first symptoms prior to the patient's 10th. birthday. This last group constitutes less than 1% of all HD patients, and shows a distinct clinical behavior with large CAG repeat expansions in the IT 15 gene.

Methods: Clinical files from 145 families with HD who attend the National Institute of Neurology and Neurosurgery were reviewed in order to identify individuals with an age of onset before 10 years of age. Molecular detection of the CAG repeat expansion was carried out in 4 out of 5 patients identified, and clinical and molecular behavior were analyzed.

Results: The PCR assay demonstrated the CAG repeat expansion in the children tested, with number of copies ranging from 54 to more than 120. All patients had paternal inheritance, and had a clinically aggressive illness, with prominent rigidity, dystonia, seizures, and behavioral disorders. Two of these children have already died.

Conclusions: Huntington's disease should be considered as a differential diagnosis of a variety of progressive motor, cognitive and/or affective disorders even in the absence of a clearly affected parent.

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The value of using gestational age by ultrasonography in Down syndrome and neural tube defects screening

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The optimal gestational age for prenatal screening for neural tube defects and Down syndrome is usually between 15 and 18 weeks. Elevated concentrations of maternal serum alpha-fetoprotein are a diagnostic indication of neural tubal defects where as low levels of alpha-fetoprotein, unconjugated estriol and elevated human chorionic gonadotropin are indication of Down syndrome. Accurate assessment of gestational age at the time of testing for these analytes is very important for the interpretation of the test results. The present report is designed to determine whether gestational age should be based on last menstrual period (LMP) data or ultrasonographic evaluation in the interpretation of maternal serum screening. The present study population consisted of 4,200 women at 15 to 19 weeks gestation. Seventy-seven percent of test interpretations were based on ultrasonographic evaluation of gestational ages. For 56 Down syndrome pregnancies both ultrasonography and LMP dates were available. Whenever the test results were positive for Down syndrome or neural tube defects, the serum samples were collected and again assayed for results. We found both initial and revised screen positive rates for Down syndrome were significantly lower and detection rates for Down syndrome seemed to be higher with ultrasonography (68% vs. 49% for LMP dates). The opposite trend was seen for neural tube defects. Serum-positive rates for neural tube defects were higher for women when gestational age was based on ultrasonographic examination than those women referred with LMP dates. The detection rates for neural tube defects by ultrasonography was slightly lower than those by LMP dates. Our data indicate that ultrasonography is preferred for the effectiveness of maternal serum screening. The ultrasonographic biometric measurement for estimating gestational age for both neural tube defects and Down syndrome screening by biparietal diameter measurement was disassumed.

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Carpal tunnel syndrome: familial autosomal inheritance

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It has been known that carpal tunnel syndrome which is common in adults is usually sporadic and idiopathic. When the inheritance is familial various disorders such as collagen vascular disease, mucopolysaccharidosis, amyloidosis are frequently manifested. It has also been shown in the literature that the carpal tunnel syndrome is always associated with the manifestation of systemic disorder. There are only a few reports which describe the absence of any disorder in patients with carpal tunnel syndrome. The present report deals with a case of 42-year old woman married to a 51-year old man and their marriage was consanguineous one. The patient had bilateral median nerve entrapment at the wrist. Various physical and laboratory tests were conducted to see any evidence of systemic disorder but there was none. Genetic counseling revealed that there were other family members had symptoms consistent with carpal tunnel syndrome. All cases were confirmed electrophysiologically. The mother of propositus, younger sister of propositus, and her 14-year old son and 19-year old daughter of propositus were also confirmed with carpal tunnel syndrome. The plausible modes of inheritance of this genetic disease were discussed.

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Severe amniotic band syndrome with limb body wall complex: Ultrasonographic-clinicopathologic correlations. J.L. Roberts, L. Csury, F. Schneider, H. Cohen and V.M. Anderson. SUNY Health Science Center at Brooklyn-Kings County Hospital, Brooklyn, NY.

A 25 year-old primigravida first presented with preterm rupture of membranes at 28 weeks gestation. She had prenatal care in Antigua. Our sonogram showed a complete encephalocele and dorsally malpositioned eyes; the rest of the face was not visualized. A beating two chambered heart was seen at the dorsal aspect of the twisted spine. The limbs could not be followed past the femur, which measured 26 weeks. Dilated bowel loops occupied the area around the fetus and there was no amniotic fluid. Augmentation of labor was initiated because of ruptured membranes and poor fetal prognosis. A 360gm female stillborn was attached to the placenta by a 6 cm umbilical cord. A dense amniotic band from the palate, amputated the nose, bisected the face and skull and adhered to an umbilical cord adjacent to a large abdominal wall defect. The brain was evacuated through a wide sagittal bone defect. Heterotopic liver with separate dorsal and ventral pancreas and apple-peel atresia of the large bowel was also identified. Meconium-filled small bowel loops were remarkably dilated. The head and trunk tethered by the amniotic band resulted in a predictable pattern of torque deformation sequence, which may account for many of the observed anomalies of the limb body wall complex. This includes: atresia, right heart; agenesis, right lung, constriction, umbilical cord; retroflexion and right lateral rotation, axial skeleton, with secondary contortion of the appendicular skeleton. The shortened right arm was frozen in a right angle contracture with amputated fingers. Talipes equinovarus of the left foot and cleft right foot with absent digits was seen. Radiographic examination of the body showed severe angulating scoliosis, hyperextension of the neck and a short, square pelvis. Pathologic examination was critical in defining these anomalies and permitted reinterpretation of the ultrasonographic findings. This case unequivocally illustrates the association of amniotic band syndrome and limb body wall complex.

Reference: Gunther, WM, Anderson, VM, Drut, RM. Torque deformation sequence associated with short umbilical cord and abdominal wall defect. *Blastogenesis: normal and abnormal*. ed. John M. Opitz. Birth Defects Original Article Series, 1993, 29: pp317-333.