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Blunt trauma to the gravid abdomen as a cause of cerebral palsy and epilepsy. R.M. Roberts, Genetics and Prenatal Diagnostic Center, Chattanooga, TN

The cause of the majority of cases of cerebral palsy (CP) and epilepsy remains speculative. The physician and pregnant mother generally regard the fetus as relatively well-protected from physical harm *in utero*. Postnatal shaken-baby syndrome and cranial blows sufficient to cause loss of consciousness are well-established etiologies of both; intracranial forces sufficient to produce a similar effect can occur *in utero*. This presentation is intended to demonstrate how relatively trivial pressure from ultrasound-guided external palpation can cause remarkable compression and movement of fetal parts. A pregnant mother could place her fetus at risk by lifting a tantruming child, receiving a kick which by chance may be transmitted to the fetal cranium just below the maternal skin surface. She would not recall such an event when signs of CP and/or epilepsy first appear. A pregnant mother with a 1 to 2-year old could be at highest risk of damage. During the third trimester, many fetal heads are maintained in vertex position, and are protected from direct percussive effects by the maternal pubis (easily demonstrated with ultrasound)--which may explain the higher incidence of CP and epilepsy in breech presentations after excluding significant hypoxia. It is also possible that the higher rate of CP in multiple gestations, after excluding effects of prematurity, could be in part due to intrauterine blunt trauma from sudden complete extension of the lower extremity, a common recurrent fetal reflex, directed at a cranium --those who are breech/vertex in the 2nd and 3rd trimester might both be at increased risk, with vertex/vertex in the 3rd trimester at lowest risk (many twins remain fixed in relative presentation in the last 2 months of pregnancy). The most attractive aspect of this theory is that *prevention will be possible*, through patient and physician awareness of the potential for harm. This hypothesis may be tested, since if valid, firstborn and siblings 4 years or more apart should have significantly lower rates than those born 1 to 2 years apart; a significantly obese abdomen would be protective. Currently it is not standard practice for physicians to warn pregnant women with regard to the possibility of blunt trauma as a potential cause of significant damage to the fetus. Such education about potential danger appears to have little risk to the patient, and the potential of benefit may warrant immediate implementation as a common-sense precaution.

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On hiccuping and yawning: why we do it. R.M. Roberts, Genetics and Prenatal Diagnostic Center, 7302 Jarnigan Road, Chattanooga, TN

Common fetal reflexes include hiccuping and yawning. In the second and third trimesters, these reflexes are easily seen, and can be documented in any fetus if ultrasonographic examination is prolonged. If observed in the sagittal plane, the diaphragm may be seen moving caudad with the yawn; a full yawn sequence is characterized by a number of small mouth openings, each with concomitant mild neck extension, just prior to the full yawn, with maximal and prolonged jaw extension and extension of the neck, followed flexion at the end of the reflex, as occurs in children and adults. Similarly, hiccuping is easily seen in the second and third trimester, the sternum suddenly retracting concomitant with caudad diaphragmatic movement, and is commonly observed in an extended ultrasound. I have determined that both of these reflexes occur in the first trimester--documenting hiccuping at 10 weeks, and yawning sequence at 11 weeks, captured on videotape. The fact that these reflexes are present and active in the first trimester likely is not by chance. From an evolutionary biological perspective, such early expression of a reflex suggests that it has a selective advantage. Since all mammals yawn, as do reptiles and birds, the reason must be pan-species. The reflexes are disinhibited in the decerebrate adult. These reflexes may be necessary for the normal development of the airway passages during organogenesis in the first trimester, helping to prevent bronchial and tracheal webs. Bronchial webs may lead to focal adenomatous malformation of the lung. Should a tracheal web occur, the evolutionary fitness for that individual is 0, since the organism would die at birth. Both reflexes would serve to dislodge potential webs from formation through hydraulic pressure variation. Any anatomic anomaly which would interfere with normal flow prenatally (then liquid flow) would predispose towards tracheal webs and neonatal death--as in a neonate who died of asphyxia last year, whom I found at autopsy affected with Treacher Collins syndrome and a tracheal web. Hence, the reason why we yawn and hiccup is simply because we had to do it in the first trimester to assure patent airway development, and postnatlly the reflexes just keep going.

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An unusual case of Smith-Lemli-Opitz syndrome and first trimester ultrasonographic exclusion in a subsequent pregnancy. R.M. Roberts[1] and R.I. Kelley[2], [1]Genetics and Prenatal Diagnostic Center, Chattanooga, TN, and [2]Kennedy Krieger Institute, Baltimore, MD

The index case was referred at 25 weeks to another center because of severe oligohydramnios. Post amnioinfusion, female external genitalia, dolichocephaly, renal agenesis (inferred from absent bladder filling), and severe IUGR led the perinatologist to suggest elective termination. Postmortem, dysmorphologic evaluation revealed dolichocephaly, asymmetric IUGR, a significant nuchal cyst, downward-slanting palpebral fissures, missing 5th ulnar ray of one hand with ulnar deviation of metacarpals, toes "cupped" dorsally, 1st and 5th (and 6th of one foot) most dorsal, female external genitalia, clitoris mildly prominent. Internal examination revealed no cerebral gyrae, bilateral unilobular lungs, membranous VSD, discoid adrenals, renal agenesis, phenotypic "ovaries" unusual in placement--high and attached to lateral pelvic wall, uterus and fallopian tubes absent. Karyotype revealed 46,XY. In spite of the rare and possibly unique occurrence of oligodactyly (absent 5th finger), a clinical diagnosis of SLO was made, and the family counseled. They elected to have detailed ultrasonography in the next pregnancy. At 10 weeks gestation both kidneys were visualized, nuchal cyst was not present, and normal growth documented. At 11w3d, 20 digits were counted, and ultrasound at 13 weeks revealed a normal number of phalanges in both hands and feet. Gas chromatography from a formalin-fixed specimen from their affected fetus revealed 7-dehydrocholesterol /Cholesterol ratio percent of 443, a markedly increased ratio of 7-dehydrocholesterol to cholesterol, consistent with the diagnosis of SLO. Normal appearing male genitalia were seen at 16 weeks; triple screen drawn then revealed a uE3 level of 0.64 ng/ml, 0.85 MOM. In the Baltimore experience, no affected fetus has had a uE3 higher than 0.65 MOM, and the more severe cases had significantly lower MOM values. The patient subsequently delivered a healthy male infant. The early ultrasound evaluation greatly alleviated the patient's level of anxiety.

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Brain and cranial disruption sequence from amniotic bands: a result of traction from fetal and embryonic swallowing of attached bands. R.M. Roberts[1] and W. Blackburn[2], [1]Genetics and Prenatal Diagnostic Center, Chattanooga, TN, and [2]Greenwood Genetics Center, Greenwood, S.C.

Ultrasonographic evaluation of a 16 week fetus referred for elevated MSAFP revealed multiple amniotic bands; the right digits were tethered to a ball of umbilical cord, in turn attached by a long band to the fetal surface of the amnion. Severe hydrocephaly was present, remarkable this early in gestation. The appearance of the choroid plexes was highly unusual, as if compressed focally, with a "butterfly" appearance in the coronal plane. A disruptive cleft extended from the right orbit through the right lateral maxilla and upper lip. Because of the poor prognosis, the patient elected to terminate the pregnancy. Chromosomal analysis revealed a normal karyotype. 3 dimensional computed tomography of the fetus skull revealed irregular synostosis of the right lambdoidal and right coronal sutures, with abnormal widening of the contralateral sutures. The fetal skin over the cranium was remarkable for the presence of three skin tags. The terminal phalanges of the right hand were disrupted by the band, which attached to the cord and amnion. Fetal surgery has demonstrated how the skin can regenerate without scarring. The cranial skin showed no evidence of scarring, but what most likely happened is that the fetus swallowed the bands attached at the 3 points on the cranium marked by the skin tags, and traction caused the bands to slice down through the cranium and brain structures, disrupting the choroid, brain anatomy, and causing the asymmetric craniostenosis and facial cleft. The skin over the cranium then healed over the defect without scarring. Also presented is a neonate with focal disruption of the toes, most likely caused by a band originally attached to the lateral surface of that foot's heel, leaving only a large skin tag as evidence of the original attachment. Other fetuses are shown with an amniotic band swallowed. The caveat: whenever amniotic bands are seen, meticulous evaluation of brain structures is warranted, and asymmetry of the choroid could be an ominous sign of more severe disruption.