EP08.23

Abnormalities of the fetal corpus callosum: is it time to develop a standard approach?

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Objectives: The wide range of fetal corpus callosal (CC) anomalies from complete agenesis to hypoplasia, especially when isolated, pose challenges for both the patient and fetal medicine specialist. This is due to a lack of standard prenatal definition and nomenclature which also hampers follow-up of cohorts that could provide future data needed upon which to base counselling. Our goal was to review the published definitions of, and to propose a standardised nomenclature for, prenatal CC pathologies.

Methods: This review was registered with PROSPERO. Keywords, fetal and corpus callosum, in a PUBMED search yielded 981 results. Excluding single case reports and non-English/French studies, 102 abstracts were reviewed, and 38 studies form our review.

Results: 16 of 38 focus on CC biometry. 22 discuss pathologies such as complete or partial agenesis (also called dysgenesis), thick, thin, short or abnormally shaped CC. Most focus on sonography, without or with MRI. 2 focus on MRI. 4 studies focus exclusively on partial agenesis and 2 on a thick CC. None focus on only thin or short but otherwise complete CC. While there is agreement on what constitutes complete agenesis of the CC, there is heterogeneity of definition of other CC pathologies and there is overlap between partial agenesis and hypoplasia of the CC. Also, a hypoplastic CC is variably defined as a thin (6 of 7 studies) or short CC (1 study). A thick or abnormally shaped CC are other rarely reported entities (3 studies).

Conclusions: Our proposed standard definitions can be used together with a choice of an appropriate biometric chart. These can be expanded as increased information, particularly from advanced fetal MRI increases our understanding of different callosal pathologies. Such nomenclature could be useful and improve future data collection and analysis, ultimately improving our ability to counsel patients.

Supporting information can be found in the online version of this abstract

EP08.24

Malformations of cortical development: from prenatal diagnosis to postnatal outcome

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Objectives: Malformations of cortical development are rare congenital brain disorders and no curative therapy exists. We report a case series (retrospective and prospective) to better understand their physiopathology.

Methods: We collected 20 cases from our database since 20 years. Inclusion criteria were availability of fetal imaging, genetic analysis and neurologic outcome. The syndromic and congenital infection were excluded.

Results: Prenatal diagnosis was done at an average gestational age (GA) of 23 weeks by ultrasound and fetal magnetic resonance imaging. Half of cases had genetic diagnosis: clinical exome found three mutations. Nine pregnancy were interrupted at an average GA

of 29 weeks and time to termination of pregnancy was of 6 weeks as a mean. One is an ongoing pregnancy. Neurologic development at two years of the ten children has been evaluated.

Conclusions: This study implements prenatal imaging and genetic data on these pathologies to adapt pregnancy follow-up and postnatal work-up.

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Gestational age at diagnosis (weeks)	18-36 (23 as average)	
Genetic diagnosis	11/20	
Prenatal imaging diagnosis	15/20	
Fetal magnetic resonance imaging	11/15	
Number of termination of pregnancy	9/20	
Alive children (age as a range)	10 (1-20 years)	
Gestational age at termination of	22-37 (29 as average)	
pregnancy (weeks)		
Time from diagnosis to termination of	1-13 (6 as average)	
pregnancy (weeks)		

EP08.25

Fetal severe exoftalmos associated to mycophenolate mofetil embryopathy

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Mycophenolate mofetil is a prodrug that prevents the proliferation of lymphocytes and the creation of adhesion molecules in response to an antigenic or mitogenic stimulus. This drug is classified within the FDA category D. The most frequent fetal malformations were microtia, micrognathia, microphthalmia, hypertelorism, cleft lip, cleft palate, agenesis of corpus callosum and esophageal atresia.

A 31-year-old pregnant woman, with personal medical history of Overlap syndrome, with component of systemic sclerosis without scleroderma, with microstomia and alteration of intestinal motility, arthritis, myositis and progressive pulmonary involvement with thrombophilia due to protein-S deficiency. The patient was under treatment with mycophenolic acid (500mg/8h), prednisone 5mg/24h, rituximab IV 2g/6m, enoxaparin 40mg/24h, AAS 150mg/24h. She was referred at week 18+1 to the Prenatal Diagnosis Unit. Agenesis of the corpus callosum, hypertelorism with severe exophthalmos, broad facial defect without objectifying palate, left congenital diaphragmatic hernia with displacement of the mediastinum and heart to the right and umbilical artery was observed. Amniocentesis is performed with the result of a normal karyotype, XY.

The parents opted for TOP. A fetal autopsy is performed, which reports cleft lip, low implantation ears, hypertelorism, exophthalmos, short neck, hypoplastic left lung, left diaphragmatic hernia.

Supporting information can be found in the online version of this abstract

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Outcome of the first two confirmed cases of congenital Zika virus

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Case reports involving the first two cases of congenital Zika syndrome confirmed by RT-PCR. The cases were followed from the gestation through serial ultrasound (GE E8). Amniocentesis was performed at week 28 and magnetic resonance at week 29. Postpartum outcomes were also monitored. Case 1: A physical therapist, 33 years old, exanthema with seven weeks of pregnancy. Neuroimaging findings: microcephaly, lysencephaly, subcortical calcifications, cerebellar vermis and corpus callosum hypoplasia, increased interhemispheric space, and reduced brain volume. Zika virus was detected by RT-PCR in amniotic fluid. Newborn female, cephalic perimeter of 30.5 cm, Apgar at 1st and 5th, of 9 and 10. The child received therapy at the Professor Joaquim Amorim Neto Research Institute until the present. The gross motor function measure was applied with 16 (median of 116), 23 (median of 170) and 34 (median of 206) weeks of life, level II by GMFCS, without seizures, walking and attending to school. Case 2: A 23 year old presented rash with 18 weeks. Neuroimaging findings: severe and asymmetric ventriculomegaly, lysencephaly, absence of thalamus, severe cerebellar and cerebellar vermis hypoplasia, unilateral microphthalmia, cataract and arthrogryposis. Newborn male, cephalic perimeter of 36.5 cm and Apgar 3/3. He died at 10 hours of life. The presence of Zika virus had already been detected in the amniotic fluid by RT-PCR and the presence of viral material was confirmed after necropsy in brain tissue, meninges and kidney. Histopathological study evidenced an important reduction of the cerebral parenchyma, important hypoplasia and calcifications in Brain stem, with presence of lymphocytes and histiocytes in deep gray nuclei, histiocytes and brainstem.

Supporting information can be found in the online version of this abstract

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Spontaneous resolution of ventriculomegaly and tethered cord in a case of spina bifida

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A 17-week pregnant was referred for a routine scan which revealed bilateral mild ventriculomegaly, a lemon-shaped head but no Chiari malformation. Close examination of the spine revealed a small sacral meningomyelocele. The spinal cord was seen apparently attached to the lesion. No other obvious defects were detected, and in particular fetal leg movements were normal. The pregnancy was closely followed-up and by 20 weeks there was complete reversal of the ventriculomegaly and by 26 weeks the conus medullaris detached from the defect, in spite of remaining lower than expected. MRI confirmed that there was no further neurological tissue present in the sac, which was then named as a meningocele. It is unclear why this case presented ventriculomegaly in the early second trimester. It is possible that epithelisation of the sac could have prevented further leakage from the defect. Interestingly, intrauterine correction of meningomyelocele has demonstrated success in partially reversing hindbrain herniation but not ventriculomegaly. One could speculate that hindbrain herniation and ventriculomegaly could involve partially independent mechanisms or that intervention occurs in too late a stage to be able to reverse ventriculomegaly.

Supporting information can be found in the online version of this abstract

EP08.28 Abstract withdrawn

EP08.29

Neurodevelopmental outcome following prenatal diagnosis of a short corpus callosum

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Objectives: Data regarding the neurodevelopmental outcome of fetal short corpus callosum (CC) diagnosed according to standard reference charts is scarce. The purpose of this study was to assess whether the finding is related to neurodevelopmental delay, and to examine reclassification to normal fetal CC length using CC length/EFW ratio.

Methods: Historical prospective cohort study including pregnant women who were referred for fetal neurosonogram due to abnormal CC. Short CC was defined below the 5th percentile according to reference charts. Twenty cases were included in the study group and compared with a control group of 59 normal cases. The patients in the study group were divided into two groups according to CC length/EFW ratio. Children's neurodevelopment was assessed using the Vineland Adaptive Behaviour Scale (VABS).

Results: VABS scores were within normal range in 90% of the cases. There was no significant statistical difference between the study group and the control group. In addition, there was no statistically significant difference between fetuses reclassified as normal callosal length according to CC length/EFW ratio in comparison to the control group.

Conclusions: The neurodevelopmental outcome of fetuses with diagnosed short CC did not differ from the neurodevelopment of normal fetuses in the control group.

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Fetal brain MRI in polyhydramnios: is it justified?

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Objectives: Despite meticulous investigation of polyhydramnios cases, in many of these cases, congenital anomalies are detected only after birth. The aim of our study was to explore the contribution of fetal brain MRI to the detection of CNS anomalies in cases of polyhydramnios.

Methods: This was retrospective cohort study on fetuses referred for the investigation of polyhydramnios at a single tertiary centre. All fetuses underwent a detailed sonographic anatomical scan and a fetal brain MRI. Isolated and non-isolated polyhydramnios were differentiated according to associated anomalies. MRI findings were compared between the groups.

Results: A total of 46 fetuses were included in the study. Brain anomalies were detected in ultrasound in 12 (26%) cases while MRI detected brain anomalies in 23 (50%) cases. MRI detected more anomalies in fetuses with non-isolated compared to isolated polyhydramnios (62.9% and 31.6% respectively, p = 0.019).

Conclusions: Fetal brain MRI may contribute to the evaluation of fetuses with polyhydramnios. The clinical value and cost-effectiveness of MRI use in the routine work-up of polyhydramnios should be assessed in future studies.