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Objectives: To assess the role of fetal magnetic resonance imaging (MRI) in detecting associated anomalies in fetuses presenting with isolated severe ventriculomegaly (VM) undergoing multiplanar ultrasound (US) evaluation of fetal brain.

Methods: Multicentre, retrospective, cohort study involving eight referral fetal medicine centres in Italy, United Kingdom and Spain. Inclusion criteria were fetuses affected by isolated severe (>15mm) VM on US, defined as VM with normal karyotype and no other additional CNS and extra-CNS anomalies on US, undergoing detailed assessment of fetal brain via a multiplanar approach as suggested by ISUOG guidelines on fetal neurosonogram. The primary outcome of the study was to report the rate of additional CNS anomalies detected exclusively at prenatal MRI and missed at US.

Results: 43 fetuses with a prenatal diagnosis of isolated severe VM on US were included in the analysis. Additional structural anomalies were detected at prenatal MRI and missed at US in 23.3% (95% CI 13.1-37.8) of cases. When considering the type of anomalies, midline (mostly hypoplasia of the corpus callosum) and cortical anomalies were both detected only on MRI in 40.0% of fetuses, while supratentorial intracranial hemorrhage was detected on MRI in 20.0% of fetuses, and polymicrogyria and periventricular heterotopia in 10.0%. The results of the logistic regression analysis showed that bilateral rather than unilateral severe VM (OR: 4.3, 95% CI 3.7-5.2, $p < 0.001$) and the degree of ventricular dilatation (OR: 1.5, 95% CI 1.2-1.5, $p < 0.001$ each mm of dilatation) were indipendently associated with the probability of detecting associated anomalies only at MRI.

Conclusions: Although the rate of associated fetal anomalies missed at neurosonography and detected only at fetal MRI in fetuses with isolated severe VM is lower than that previously reported, the findings from this study support the practice of MRI assessment in every fetus with a prenatal diagnosis of severe VM.

VP25.24

Fetal ultrasound, MR imaging and MR autopsy in Cri du chat syndrome: two case reports and review of the literature

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Although rare, Cri du chat (CdCS or 5p-) syndrome (OMIM #123450) is one of the most common contiguous gene deletion disorders. It is caused by chromosome 5p deletions (5p-), encompassing the critical regions 5p15.3-p15.2, and it is characterised by a high-pitched cat-like cry, distinct craniofacial dysmorphisms, microcephaly, abnormal dermatoglyphics, ophthalmologic anomalies, hypotonia, growth delay and severe psychomotor and mental retardation. Although the syndrome is well characterised in children, only a few cases of prenatal diagnosis of 5p- syndrome have been reported and knowledge is very limited as regards the prenatal imaging markers of CdCS.

We describe the ultrasonographic and magnetic resonance imaging (MRI) findings in two cases of prenatally diagnosed CdCS at 21 weeks GA: both fetuses showed mild nuchal edema, borderline ventriculomegaly, pons hypoplasia and a reduction in cerebellar diameters. We also revised all reported 5p- cases detected prenatally with CNS anomalies, finding that in 43.5% of cases, the CNS anomaly was apparently isolated. Taken altogether, cerebellar hypoplasia was described in about 65% of cases and ventriculomegaly in about 35%. Along with the reduction in cerebellar diameters, our cases presented a marked reduction in pons bulging, detected at dedicated neurosonographic evaluation and confirmed with fetal and autoptic MRI. These overall data indicate that the most frequent prenatal morphological feature of CdCS is cerebellar hypoplasia, often associated with hypoplasia of the pons.

In conclusion, it would appear that cerebellar hypoplasia associated with pontine hypoplasia and enlargement of the ventricular system are significant diagnostic indications in utero for Cri du chat syndrome. The present report also points to the usefulness of fetal MRI for the detection and accurate description of cerebral anomalies such as brainstem hypoplasia, which can be missed during ultrasonography.

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Ultrasound and morphological features of fetal anencephaly

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Anencephaly is defined as total or partial absence of the calvarium, with absence of the brain. Anencephaly has an incidence of 1 to 5 in every 1,000 births, and the mortality rate is 100% during intrauterine life or within hours or days after birth. The etiology of anencephaly remains unclear, but various maternal-related environmental and genetic risk factors have been reported, which include diabetes, obesity, exposure to different drugs or toxins, genetic polymorphisms and mutations, as well as positive family history for neural tube defects. One of the most important nutritional factors in the development of anencephaly is folate deficiency. Etiopathogenesis of anencephaly includes several mutations in various other genes, such as: PDGFRA, CELSR1, VANGL1 and VANGL2, the last two being involved in the process of neurulation. Screening tests include maternal serum AFP level and ultrasound examination. During the first trimester ultrasound screening, anencephaly is now detected in all cases, but in order to decrease the complication rate of pregnancy termination, the diagnosis should be established as soon as possible, during the pregnancy confirmation ultrasound. We conclude that given that anencephaly is a severe malformation of the central nervous system, morphological characterisation could improve the screening by ultrasound that is mandatory in the early first trimester in order to plan the best, safe and early management. For this aim, we describe the whole spectrum of ultrasound and morphologic features of anencephaly.