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A rare case of hemimegalencephaly diagnosed prenatally

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ABSTRACT

Hemimegalencephaly (HME), or unilateral megalencephaly, is a rare congenital brain malformation defined as overgrowth of one cerebral hemisphere or part of it resulting from abnormal cortical development and neuronal migration. However, cortical developmental abnormalities are rarely diagnosed prenatally. This is the reason for our study, in which we describe and compare ultrasound and MRI findings in a fetus with HME.

Key words: fetus; hemimegalencephaly; central nervous system; ultrasound

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Hemimegalencephaly (HME), or unilateral megalencephaly, is a rare congenital brain malformation defined as overgrowth of one cerebral hemisphere or part of it resulting from abnormal cortical development and neuronal migration. In addition to overgrowth, abnormal cortical development can manifest as polymicrogyria, agyria, pachygyria, lissencephaly, or grey matter heterotopia [1, 2]. The risk of chromosomal anomalies with HME is low but the risk of non-chromosomal abnormalities is relatively high. HME may be either isolated or associated with some neurocutaneous syndromes such as Proteus syndrome, epidermal nervus syndrome, tuberous sclerosis [1, 2]. Clinically, HME is associated with severe psychomotor delay and intractable epilepsy. The prognosis is poor, and the only possible treatment is a hemispherectomy to control seizures [3]. Evaluation of HME may be made by ultrasound or magnetic resonance imaging (MRI) [1–5]. However, cortical developmental abnormalities are rarely diagnosed prenatally. This is the reason for our study, in which we describe and compare ultrasound and MRI findings in a fetus with HME.

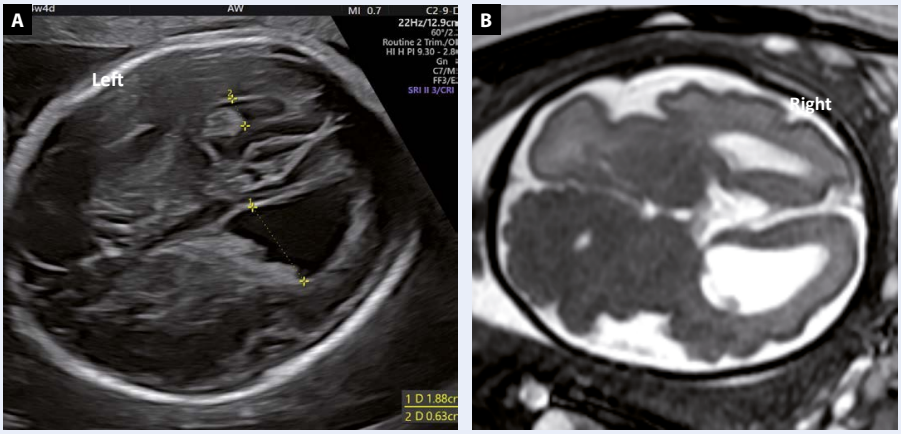

Figure 1. Hemimegalencephaly. Transventricular plane showing enlarged right cerebral hemisphere and the unilateral right side ventriculomegaly with midline shifted to the left. (**A**) 2D transabdominal ultrasound scan at 24 gestational weeks; (**B**) foetal MRI at 29 gestational weeks, FIESTA, axial plane

A 45-year-old pregnant woman at 22 weeks of pregnancy was referred to our department due to ventriculomegaly at anomaly scan. It was her fifth pregnancy, with no family history of central nervous system anomalies, and with a negative TORCH test result. A first trimester screening for aneuploidy was not performed. Ultrasound examination showed normal growth and normal anatomy except for the brain. Neuro-

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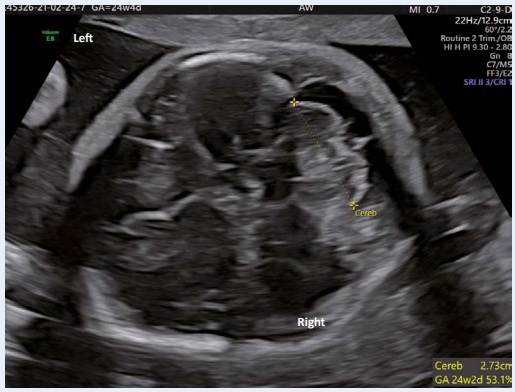

Figure 2. Hemimegalencephaly with normal anatomy of the cerebellum (2D ultrasound at 24 weeks of gestation)

sonography revealed unilateral right ventriculomegaly, the midline shifted to the left with right hemisphere overgrowth (Fig. 1A), and a head circumference in the 98th percentile. Transcerebellar section showed normal anatomy of the posterior fossa (Fig. 2). In the coronal plane, both the genu of the corpus callosum, and an abnormal and excessive gyration of the right hemisphere were visible (Fig. 3A). Hemimegalencephaly with gyral abnormality was suspected and the patient was referred for an MRI, by which, at 29 weeks of gestation our diagnosis was confirmed. Fetal MRI showed hemimegalencephaly of the right cerebral hemisphere with polymicrogyria and heterotopia (Fig. 1B and Fig. 3B). During pregnancy the head circumference and biparietal diameter remained greater than the 90th percentile. The mother underwent an elective cesarean section at 40 weeks' gestation and gave birth to a male infant weighing 3710 g, Ap 10 with birth head circumference of 38 cm (> 99th percentile). Array-based comparative genomic hybridisation (aCGH) performed after birth from peripheral blood was normal and genetic counselling did not show any other signs of neurocutaneous syndromes. Postnatally the child was provided with neurological and rehabilitation care. No seizures were observed during the neonatal period.

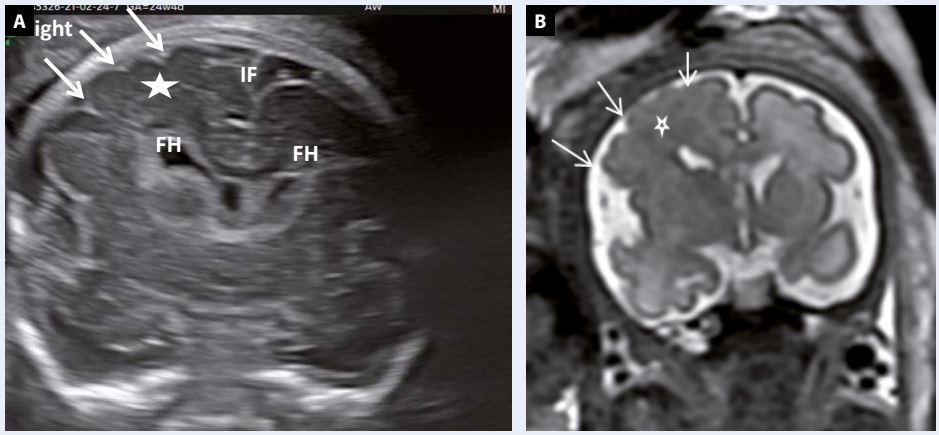

Figure 3. Hemimegalencephaly. Coronal view of the fetal brain showing an owergrowth of the right hemisphere and abnormally developed sulci and gyri (arrows): (**A**) transabdominal 2D ultrasound view at 24 weeks; (**B**) fetal MRI at 29 gestational weeks; SSFSE/T2, coronal plane, apart from abnormally developed sulci and gyri (arrows) hypointensity of the right cerebral white matter (asterisk) as a sign of migration abnormality is visible; IF — interhemispheric fissure; FH — frontal horn

In conclusion, HME is a difficult but possible diagnosis to make using antenatal ultrasound. However, fetal MRI should be considered in cases where unilateral ventriculomegaly is diagnosed at ultrasound. In utero diagnosis of HME allowed a multidisciplinary approach for providing optimal prenatal and postnatal patient counselling and treatment.

Conflict of interest

All authors declare no conflict of interest.

REFERENCES

1. Flores-Sarnat L. Hemimegalencephaly: part 1. Genetic, clinical, and imaging aspects. J Child Neurol. 2002; 17(5): 373–84; discussion 384, doi: 10.1177/088307380201700512, indexed in Pubmed: 12150586.
2. Manoranjan B, Provias JP. Hemimegalencephaly: a fetal case with neuropathological confirmation and review of the literature. Acta Neuropathol. 2010; 120(1): 117–130, doi: 10.1007/s00401-010-0663-4, indexed in Pubmed: 20237789.
3. Alvarez RM, García-Díaz L, Márquez J, et al. Hemimegalencephaly: prenatal diagnosis and outcome. Fetal Diagn Ther. 2011; 30(3): 234–238, doi: 10.1159/000329937, indexed in Pubmed: 21921574.
4. Romero XC, Molina FS, Pastor E, et al. Hemimegalencephaly: 2D, 3D Ultrasound and MRI Correlation. Fetal Diagn Ther. 2011; 29(3): 257–260, doi: 10.1159/000322012, indexed in Pubmed: 21109730.
5. Senapati G, Levine D. Prenatal-postnatal correlations of brain abnormalities: how lesions and diagnoses change over time. J Pediatr Neuroradiol. 2012; 1(3): 171–184, doi: 10.3233/PNR-2012-027, indexed in Pubmed: 24078783.