Irregular lateral borders in mild unilateral ventriculomegaly as a sign of periventricular nodular heterotopia

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Case

A 32 years old primigravida was referred to our center at 18 weeks. The female fetus showed an isolated mild unilateral ventriculomegaly of 12 mm in the atrium. Amniocentesis and TORCH-Test showed inconspicuous results.

MRI

In 26 weeks MRI was carried out with an 1,5 Tesla system. The MRI confirmed the ventriculomegaly and described nodules of variable size protruding into the ventricular lumen (figure 1) and additionally a square shape of the frontal horns of the lateral ventricles. These changes are typical for a periventricular nodular heterotopia (PNH).

Ultrasound

Ultrasound images in 27 weeks showed the hyperechoic irregularities of the lateral borders of the lateral ventricles (figure 2) which corresponded, to the nodules in the MRI and the square shaped frontal horns described in the MRI (figure 3).

Consequences

The majority of patients with PNH suffer from epilepsy, seizures and/or developmental delay.

Mutations in the FLNA gene are associated with the most common phenotype of PNH, but in our case it was negative without proof of mutations.

Additional MRI of the mother gave no indication of PNH.

Conclusion

In routine prenatal ultrasound PNH is underdiagnosed. Irregular ventricular border with nodules protruding into the ventricular lumen and square shaped frontal horns can point to PNH. MRI can confirm the ultrasound suspicion.