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A case of microlissencephaly

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Objective

Microlissencephaly describes a heterogenous group of a rare cortical malformations characterized by lissencephaly in combination with severe congenital microcephaly. We aimed to present a case of fetal microlissencephaly that is first presented at 23th gestational week with cerebellar hypoplasia.

Methods

A 33-yearold multigravid woman was diagnosed with cerebellar hypoplasia during routine screening at 23 weeks gestation. Amniocentesis was revealed a pathological deletion in 2q13 (110103001_110225743). Ultrasound findings suggestive of cortical malformations were prominent subarachnoid spaces, the presence of shallow sylvian fissure, delayed appearance of landmark sulcation, and a small head circumference at 25th week. Follow-up scans demonstrated a persistent microcephaly, cerebellar hypoplasia and a lack of the brain sulcation, subsequently confirmed by Magnetic Resonance Imaging (MRI). The couple opt for termination of pregnancy and termination was done at 28 th week.

Results

Microlissencephaly is a cortical malformation characterized by overfolding and abnormal lamination of the cerebral cortex due to a neuronal migration alteration. Manifestations include epilepsy, speech disturbance and motor and cognitive disability. The severity of microlissencephaly is highly dependent on the location and size of the affected area. Causes include acquired prenatal insults and inherited and de novo genetic variants. While microlissencephaly is usually an isolated finding it can be seen in association with other brain malformations including gray matter heterotopia and ventriculomegaly, others types of neuronal migration alterations, as well as abnormalities of the white matter, corpus callosum, brainstem and cerebellum.

Conclusion

Although the diagnosis of microlissencephaly is typically made by MRI, ultrasound is a useful tool for an early screening of this brain disruption assessing the morphology of the fissures and sulci according to gestational age.