

Lobar hemimegalencephaly, diagnosis through prenatal neurosonography and correlation with exome

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INTRODUCTION:

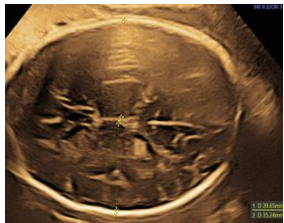
Hemimegalencephaly (HMG) is a severe and extremely rare brain malformation caused by the overgrowth of an area of the brain. There are no reported cases of prenatal ultrasound diagnosis of lobar hemimegalencephaly.

SUMMARY:

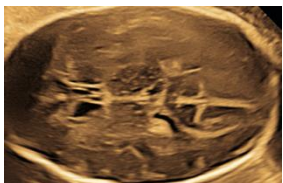
This study describes the neurosonographic findings that suggested lobar hemimegalencephaly, as well as its correlation through magnetic resonance imaging (MRI) and exome results.

CASE REPORT:

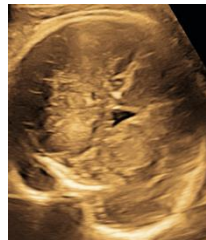
A 30-year-old patient was found to have a slightly asymmetric anterior complex, deviated from the midline, during routine screening at 21 weeks of gestation. Amniocentesis was indicated with normal results in QF-PCR, karyotype, and array. At the neurosonographic control at 30 weeks, the head circumference was at the 99th percentile (Z score = +2.6 SD) with an abnormal anterior complex in axial view, due to CSP asymmetry. In coronal sections, the right anterior horn was dysmorphic, and the left was collapsed due to increased volume in the perisylvian area. The alteration was confined to an area of the left hemisphere (lobar). Regarding cortical development, an abnormal Sylvian fissure and indentations suggestive of polymicrogyria were observed. The rest of the intra- and extracranial structures were normal. In order of fNSG findings we indicated exome by suspecting a MTOR anomaly. We also indicated MRI. The patient opted for termination. Necropsy confirms findings.



Biometry (30w)
CC at the 99th p (Z = +2.6 SD)
Lobar asymmetry

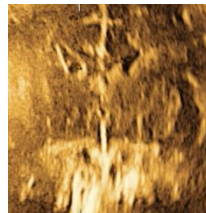


Axial view (30w)
Abnormal anterior complex
due to CSP and lobar asymmetry.



Coronal sections (30w)

right anterior horn dysmorphic,
left collapsed,
increased volume in the
perisylvian area,
abnormal Sylvian fissure
and indentation
(suggestive of polymicrogyria).



EXOME:

alteration MTOR pathway exon 10 PIK3R2 gene

MRI (32w)

malformation of the left cortical development,
with polymicrogyria in the frontal lobe and
perisylvian region, possible fusion of the left
caudate and lenticular nucleus,
alteration of the anterior limb of the left internal
capsule.