Macrocephaly and dilated lateral brain ventricles at third trimester, differential diagnose

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Objective

Differential diagnosis for macrocephaly and dilated lateral brain ventricles at third trimester of pregnancy.

Clinical case

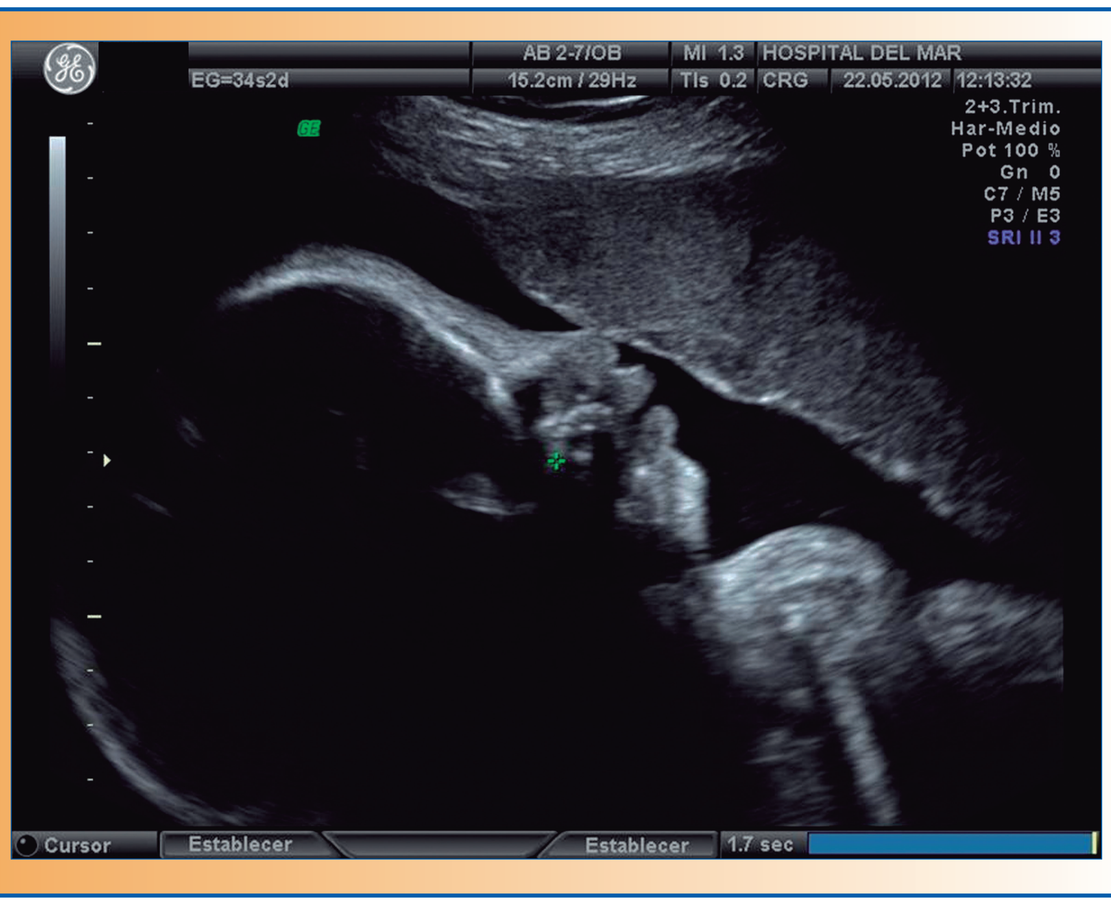
A 21-year-old primipara with no previous medical records, 34 weeks pregnant, presents at third trimester ultrasound. At the ultrasound we find 11 mm lateral ventricles with non-normal structure, enlarged third ventricle and cranial biometry over p98 for gestational age. Also findings show retrognatia and unusual fetal face line. No other significant findings.

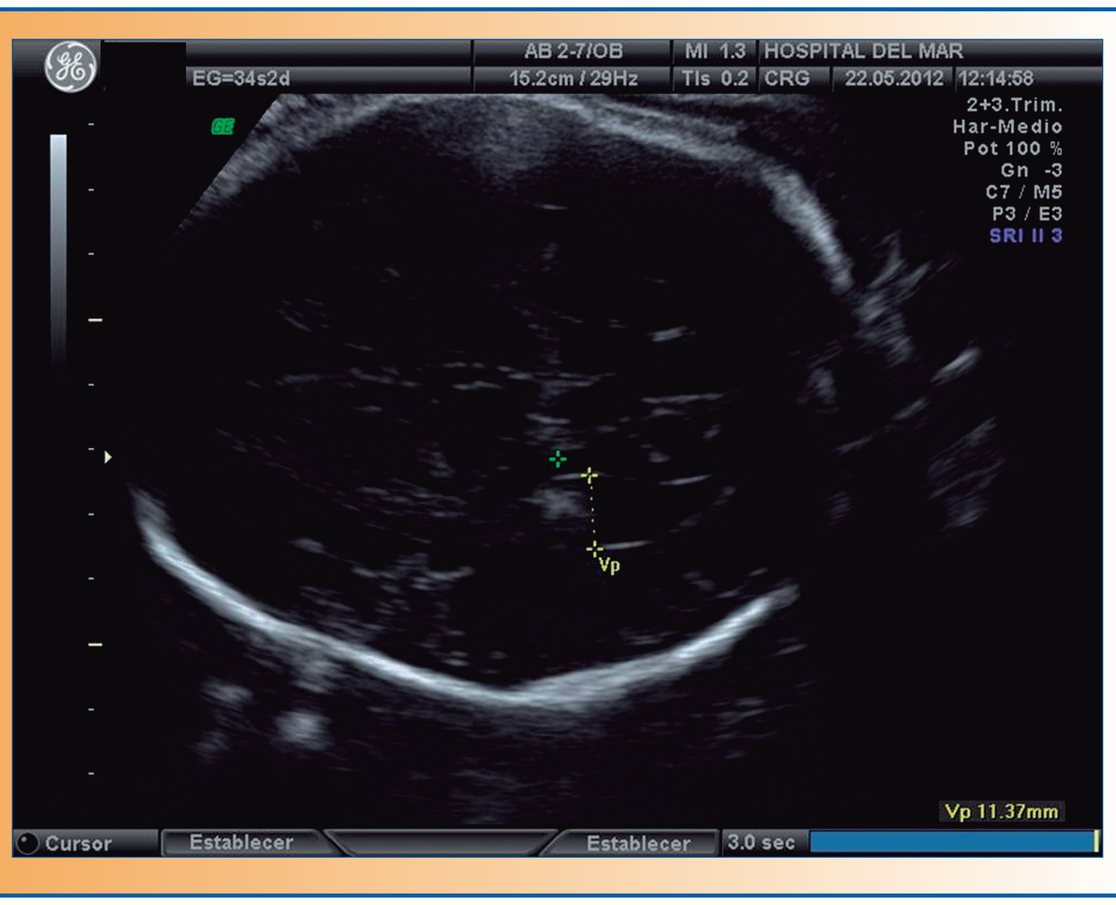
Maternal serological findings result all negative and amniocentesis shows karyotype 46XX (add(1)(q?42)), with unknown origin chromosome 1 material. Both parents karyotype are normal. Fetal MRI confirms ultrasound findings but adds no more information. As we couldn't get a better diagnose ante-partum it is decided to perform more tests after birth.

At 39.2 weeks patient gives birth a 3000 gr female baby after vaginal delivery, APGAR score is normal and cord pH are over 7.20.

Neonatal exploration shows macrocephaly, low implantation ears, generalized muscular low tone, apnea and increased brain lateral ventricles (18 mm) in ultrasound. Neonatal karyotype confirms a partial chromosome 1 trisomy.

Final diagnose shows mental retardation, seizures and abnormal neurological development as main clinical features.





Conclusion

Finding a brain lateral ventricles enlargement at third trimester forces us to make differential diagnose, initially, of infectious illness. Once those are excluded, main diagnoses are genetic defects and purely morphological defects.

After a non-conclusive genetic finding during fetal life, parents must be studied to exclude family variants that aren't pathologic. If parental karyotype is normal, further genetic studies and genetics specialist counsel must be provided.

Fetal defects at third trimester ultrasound are increasing. Case differential diagnose can help to establish severity of the findings. Unfortunately, most of the times a conclusive diagnose results impossible to obtain antepartum and final diagnose is achieved after birth.

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