



EP06.26 A rare presentation of megalencephaly - capillary malformation - polymicrogyria syndrome (MCAP) with hydrops and rhombencephalosynapsis (RES).



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Introduction

Megalencephaly - capillary malformation - polymicrogyria syndrome (MCAP) is a rare disorder within the PIK3CA-related overgrowth spectrum caused by somatic activating mutations in the PIK3CA gene. Most cases are diagnosed postnatally, as the fetal phenotype may be subtle or evolve only later in pregnancy and also because the underlying mutation occurs in mosaic form, which can limit prenatal detection.

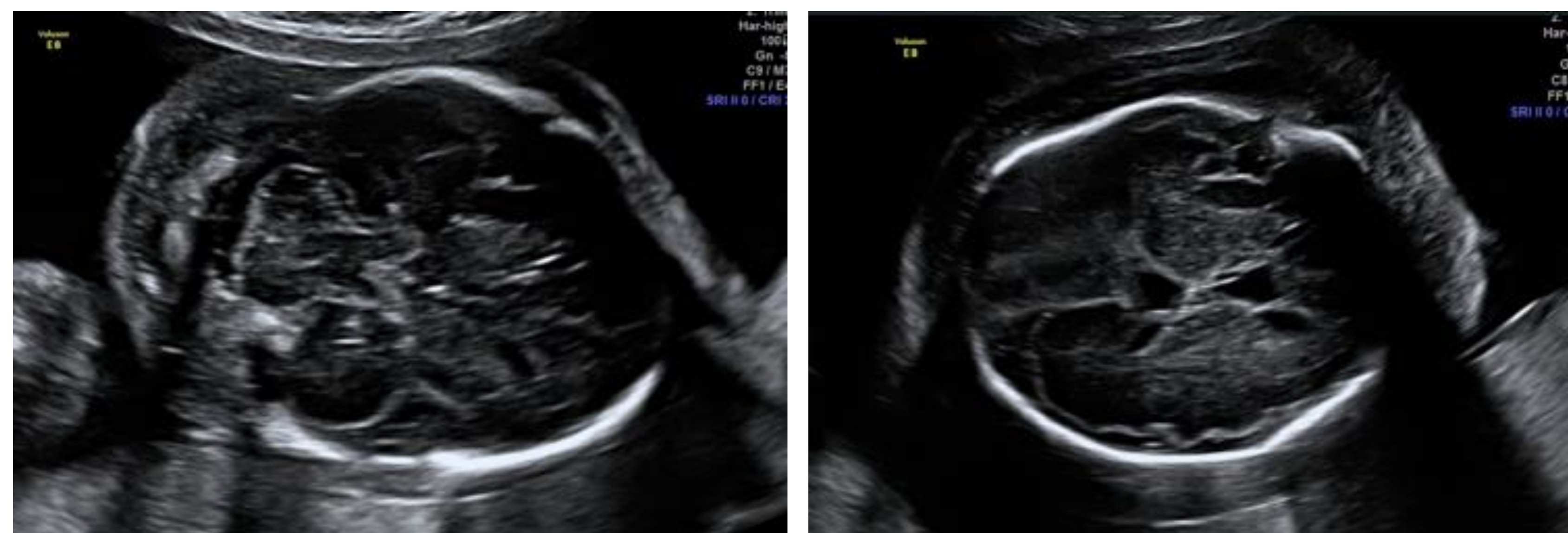


Figure. Prenatal axial ultrasound images of the fetal brain demonstrating cerebellar hypoplasia and abnormal cortical gyration, raising suspicion for RES.

Case Presentation

In this case report, a 29-year-old primigravida was referred at 22 weeks of gestation due to aberrant right subclavian artery, right aortic arch, and right-sided pleural effusion. We also noted fetal overgrowth (head and abdominal circumference on the 100th percentile), frontal bossing and cerebellum hypoplasia with suspicion of rhombencephalosynapsis (RES). Amniocentesis was performed and follow-up in one week showed progression to overt hydrops fetalis with severe bilateral pleural effusions, ascites, nuchal edema and abnormal gyration was suspected. Fetal MRI at 24 weeks confirmed partial RES and polymicrogyria of the frontal lobes. While chromosomal microarray was normal, Whole Exome Sequencing identified a pathogenic somatic mosaicism in the PIK3CA gene: c.241G>A p.(Glu81Lys). Notably, despite the severity of the hydrops, fetal Doppler studies remained normal up to 27 weeks when parents elected for termination of pregnancy. Postmortem examination was requested to correlate prenatal imaging with pathological findings.

Conclusion

This is a rare manifestation of MCAP in the second trimester with hydrops and RES. Posterior fossa abnormalities in MCAP typically involve cerebellar overgrowth and tonsillar ectopia with crowding of the posterior fossa. In this case, the presence of RES represents an atypical phenotype of the PIK3CA p.Glu81Lys variant and when identified prenatally in association with progressive megalencephaly, cortical malformations and unexplained hydrops fetalis, it should raise suspicion for a PIK3CA-related overgrowth disorder and prompt genomic testing.



36TH ISUOG WORLD CONGRESS
ON ULTRASOUND IN OBSTETRICS & GYNECOLOGY
LONDON, UK 4-7 SEPTEMBER 2026

