

EP 05.62. Megacystis–microcolon–intestinal hypoperistalsis syndrome type 5 with ACTG2 gene mutation: a case report with prenatal diagnosis and postnatal care

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Introduction

Megacystis-microcolon-intestinal hypoperistalsis syndrome is a rare entity with female predominance and autosomal recessive inheritance pattern, mutations in the ACTG2 gene, leading to altered actin filaments and smooth muscle function, have been linked to this syndrome. Prenatal diagnosis remains a challenge and only 26% of cases can be detected prenatally. Survival is poor, with renal failure and sepsis as the leading complications. We present a case report of prenatal diagnosis of megacystis-microcolon-intestinal hypoperistalsis syndrome, genetic confirmation and postnatal interdisciplinary care.

Case report

A 33-year-old woman was referred at 23 4/7 weeks' gestation for maternal-fetal medicine evaluation. A megacystitis of 5.6x5.4 cm was observed, as well as bilateral hydronephrosis with renal pelvis of 10 mm, in the absence of obstruction and fetus with female genitalia. At 32 0/7 weeks a fetal MRI was performed, finding small bowel loops and colon displaced by the distended bladder with an approximate volume of 378 cc, as well as

bilateral hydronephrosis. At 37 3/7 weeks, a female newborn of 3,620 grams was delivered by cesarean section. During postnatal approach, the bilateral urinary tract dilatation associated with megacystis and nephromegaly, as well as decreased caliber of the colon in all segments and loss of the usual haustral pattern was confirmed. The exome genetic study reported a pathogenic variant in heterozygosity in the ACTG2 gene, confirming the diagnosis of autosomal dominant megacystic bladder-microcolon-hypoperistalsis type 5. A multidisciplinary follow-up was performed at a tertiary pediatric hospital where she received multiple surgical interventions such as vesicostomy, however, with repeated urinary tract infections, at 5 months she presented septic shock and death.

Conclusion

This case report displays the spectrum of complications associated with this pathology, even with prenatal diagnosis and postnatal interdisciplinary care, we observe the poor prognosis.

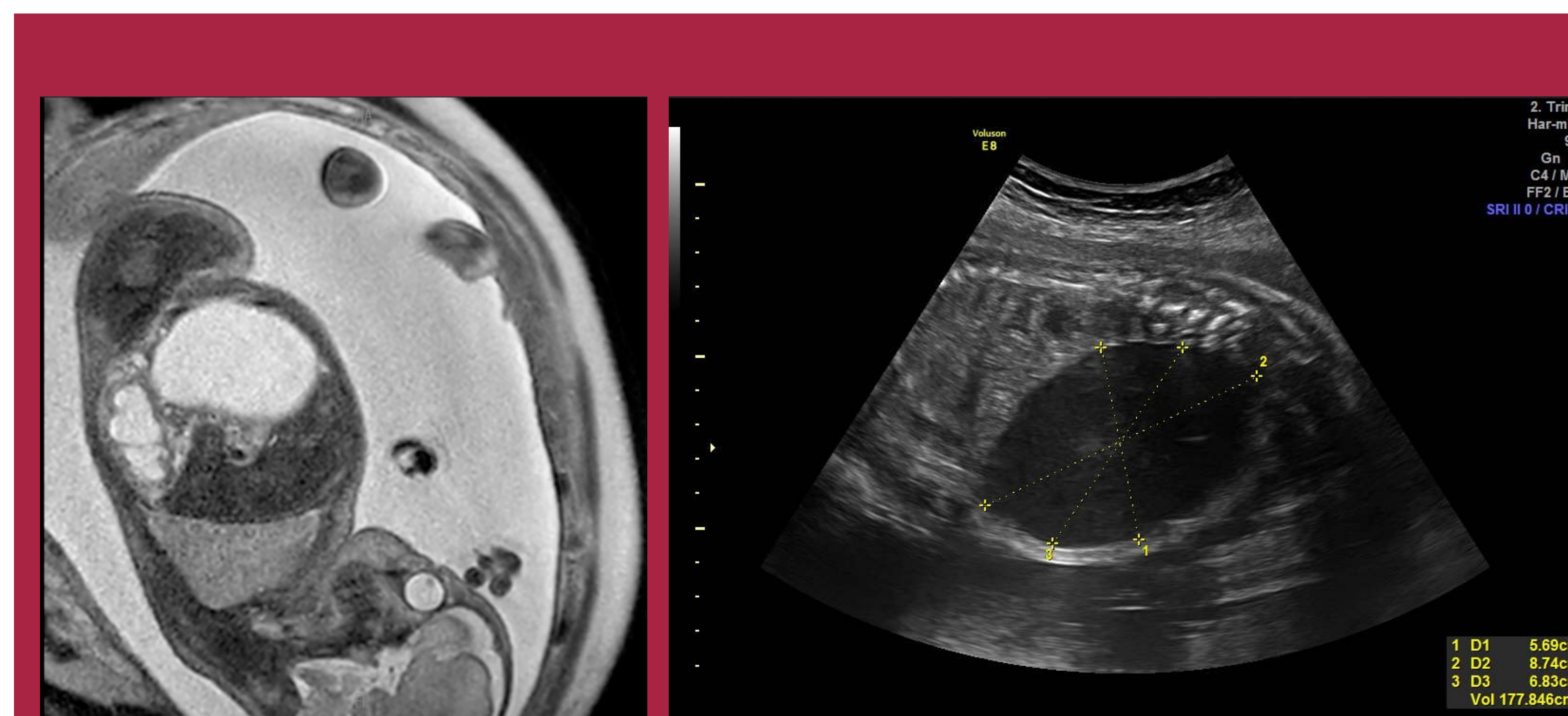


Figure 1. Sagittal view of evidence of megacystitis and microcolon (left MRI image and right ultrasound image)



Figure 2. Axial view of megacystitis and bilateral hydronephrosis (left MRI image and right ultrasound image)