

Case Series on Prenatally detected rare cephaloceles with postnatal follow up (Anterior, Interparietal and Parietal)

Tulika Joshi¹, Khurshid Alam²(Consultants Fetal Medicine) (Tulip's Fetal care Ranchi¹) (Fetomat Wellness Kolkata²)



INTRODUCTION

Cephalocele is the protrusion of cerebral and/or meningeal structures through a midline cranial bone defect of variable size. The quoted incidence in various populations ranges from 1:2000 to 1:5000 live births with no gender predominance. Most of the cephaloceles are occipital in origin but ~15% are located in fronto-ethmoid region and rest in parietal region. We are presenting a case series of prenatally detected rare encephalocele of anterior interparietal and parietal region with 3D rendered images of the defect and postnatal and post-surgical follow up of one case.

Methods

Our study is a retrospective observational study of prenatally detected rare encephaloceles. All the cases with frontal, frontoethmoidal, parietal and interparietal encephalocele were included. Follow up calls were done to the patients regarding outcome of the cases.

Results

Case 1 - primigravida seen at 14w5days of gestation. A midline defect was seen in fronto-ethmoid region with herniation of meninges and brain matter presenting as cystic mass at glabella measuring 11.2 x 8.8mm with solid areas. Both orbits were widely separate suggestive of hypertelorism. Prenatal/ postnatal testing was refused by the patient and pregnancy was terminated. (figure 1)



Figure 1

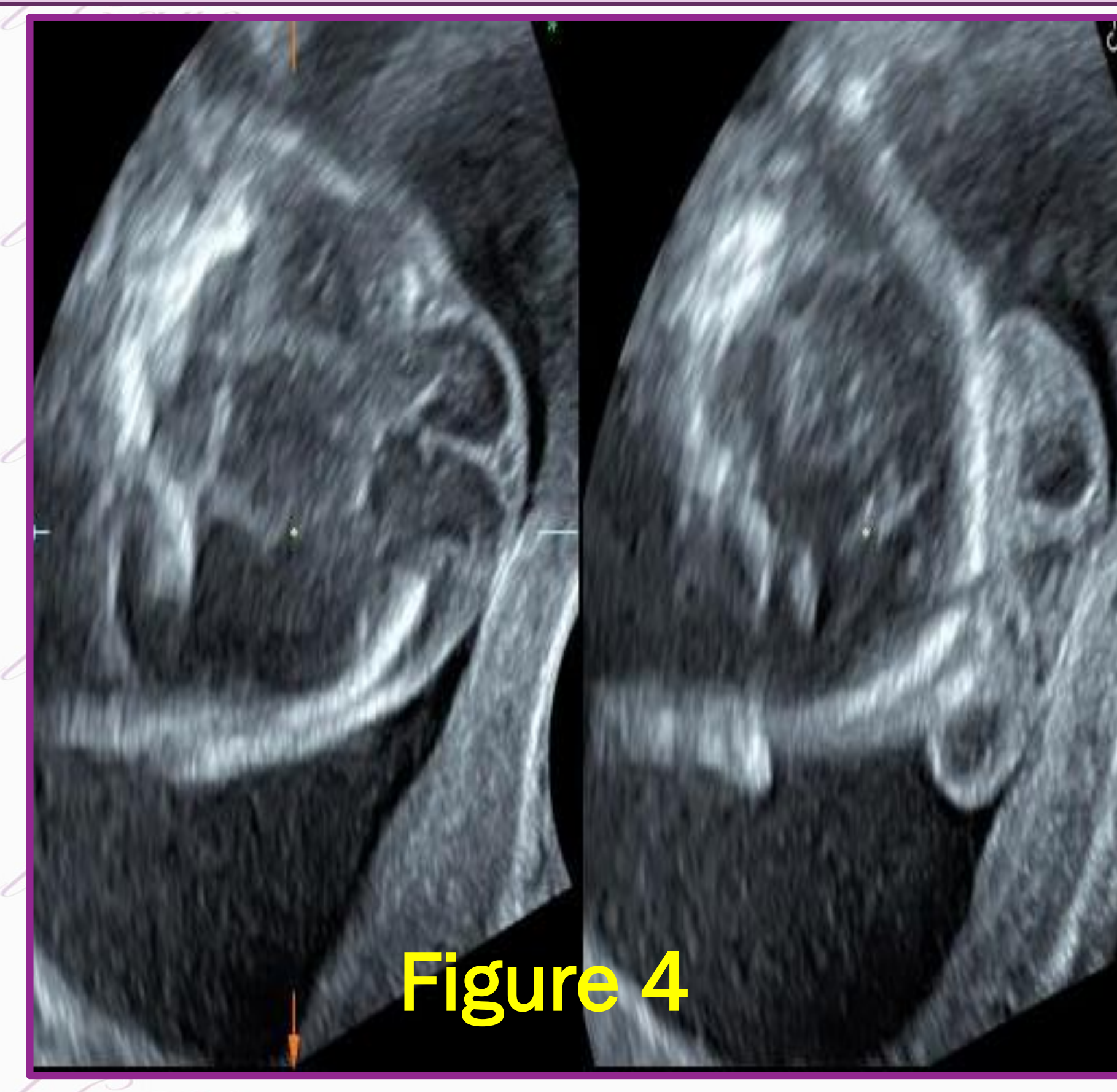


Figure 4

Case 2 -G1P0 DCDA twin gestation seen at 25W5D gestation. The twin B showed Anterior Ethmoidocele with hypertelorism. Preterm caesarean delivery was done at 35 weeks gestation but corrective surgery was deferred till 3 months of gestational age due to financial reasons. The encephalocele increased in size during the interval. Post surgical repair the baby developed CSF rhinorrhoea and died due to sepsis. (Figure 2 a&b prenatal , c postnatal 2 months, d post operative)

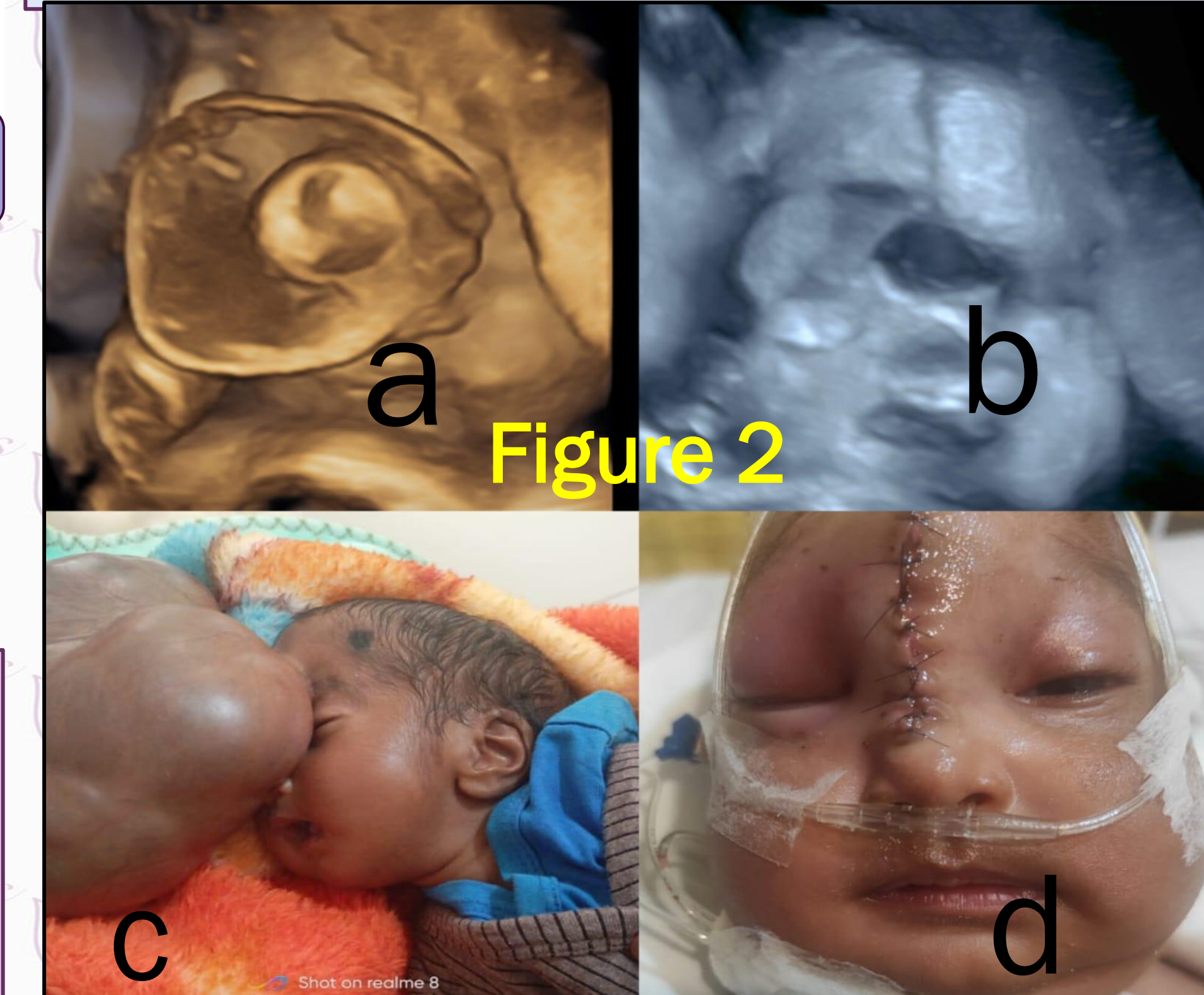


Figure 2

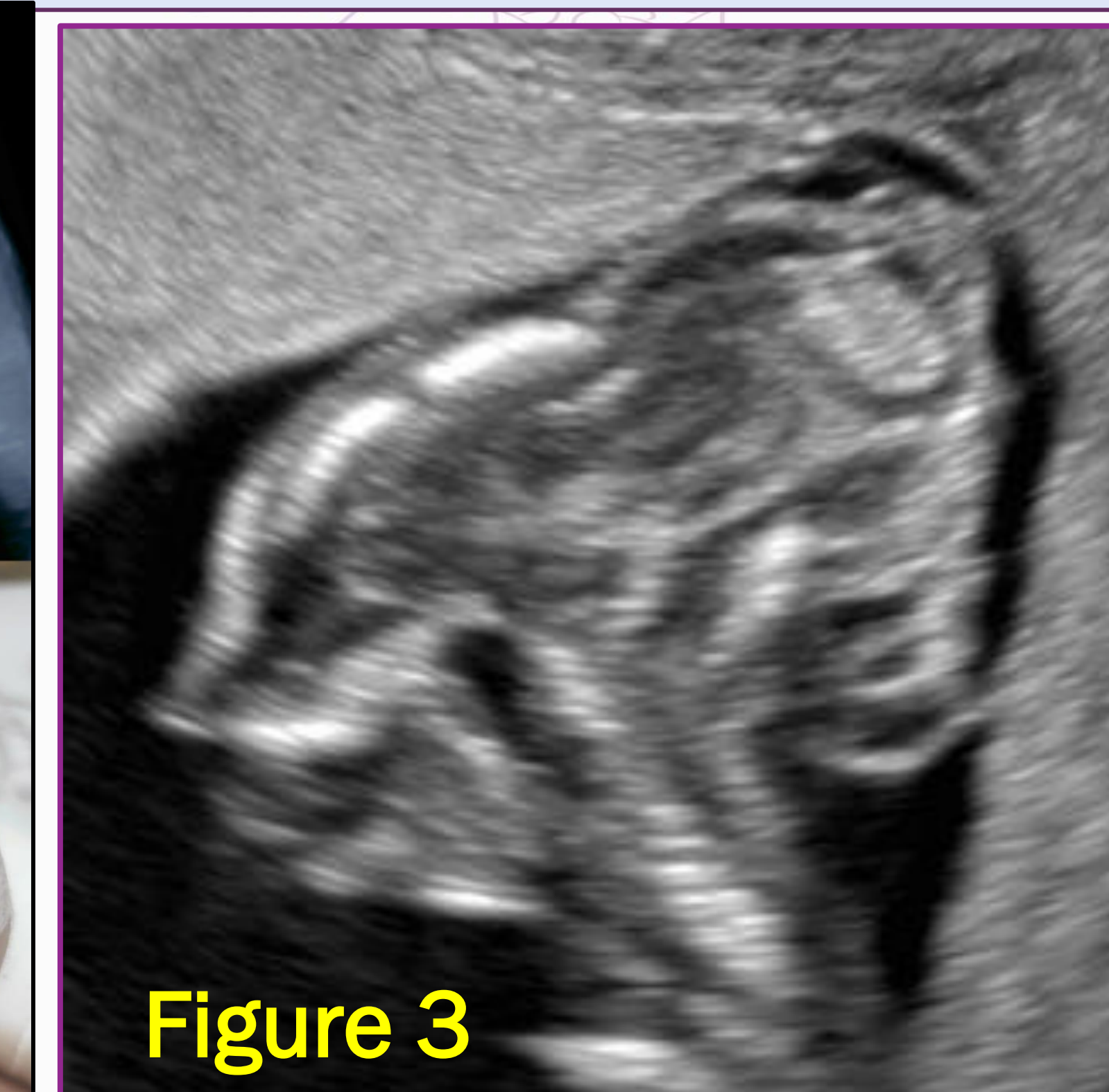


Figure 3

Case 3 -A large parieto occipital defect was seen in a 14 weeks fetus of primigravida with herniation of brain matter and meninges through widely separated parietal bones. The termination of pregnancy was done in this case and postnatal karyotype was reported normal. Figure 3

Case 4 -G1P0 with prenatal ultrasound at 16 weeks showing a paracrani al mass measuring 4.3x3.9 cm in relation to parietal bone in midline extending posteriorly. Posterior Fossa appeared obliterated. The pregnancy was terminated and genetic studies were declined. Figure 4

Case 5 - G3P1L1A1 nonconsanguineous marriage referred for routine mid trimester anomaly scan at 20w4d. Her first pregnancy was terminated at 6 months for brain abnormalities. On examinations fetus has **bilateral enlarged echogenic kidneys** (Fig 5), **Bilateral polydactyly** (fig8), **microcephaly**, **retrognathia** (figure 7) and **oligohydramnios**. The **ruptured occipital encephalocele** (figure 8) was visualised only after termination of the pregnancy. The clinical exome sequencing was sent to confirm the diagnosis of Meckel Gruber syndrome.

Results

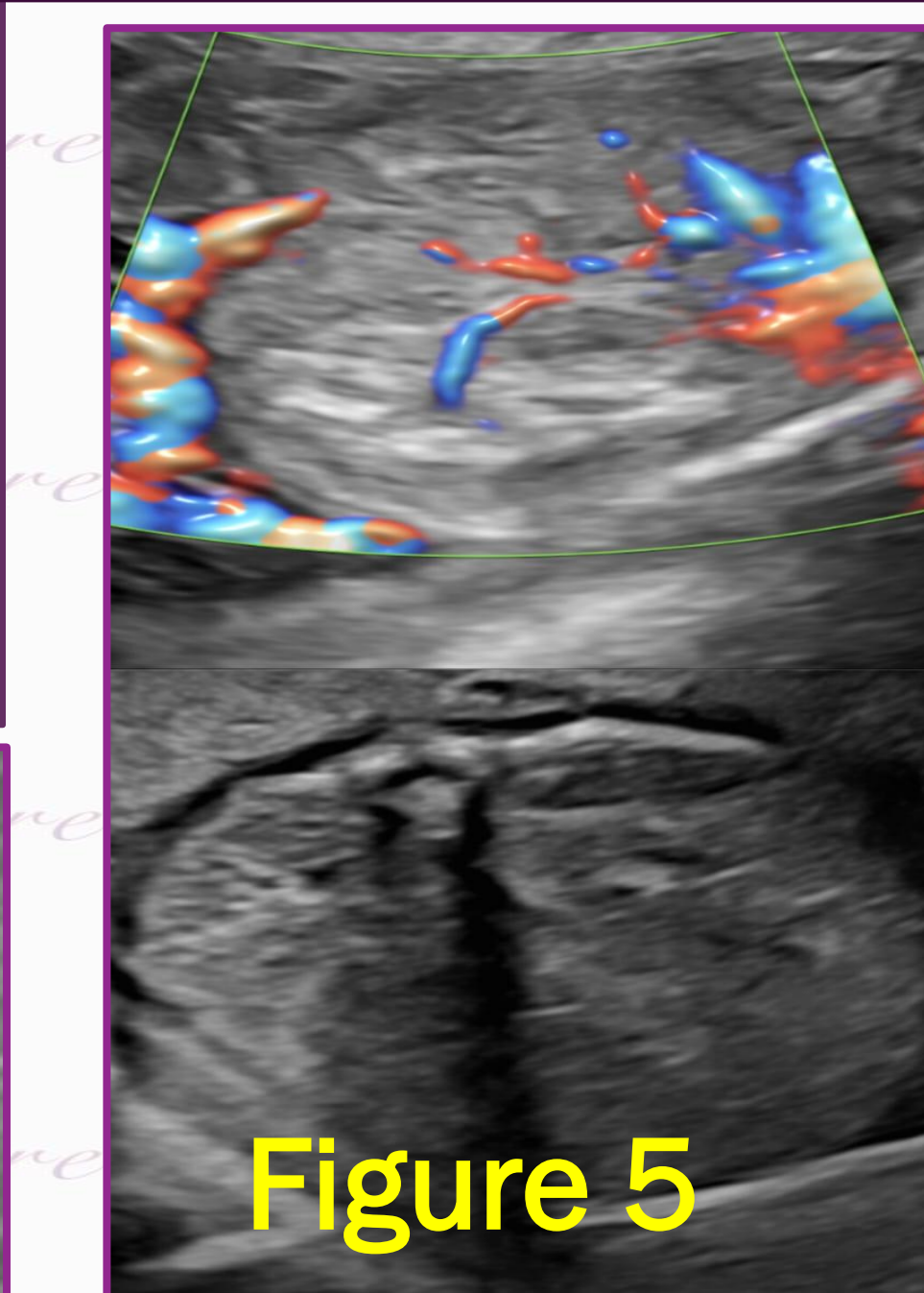


Figure 5

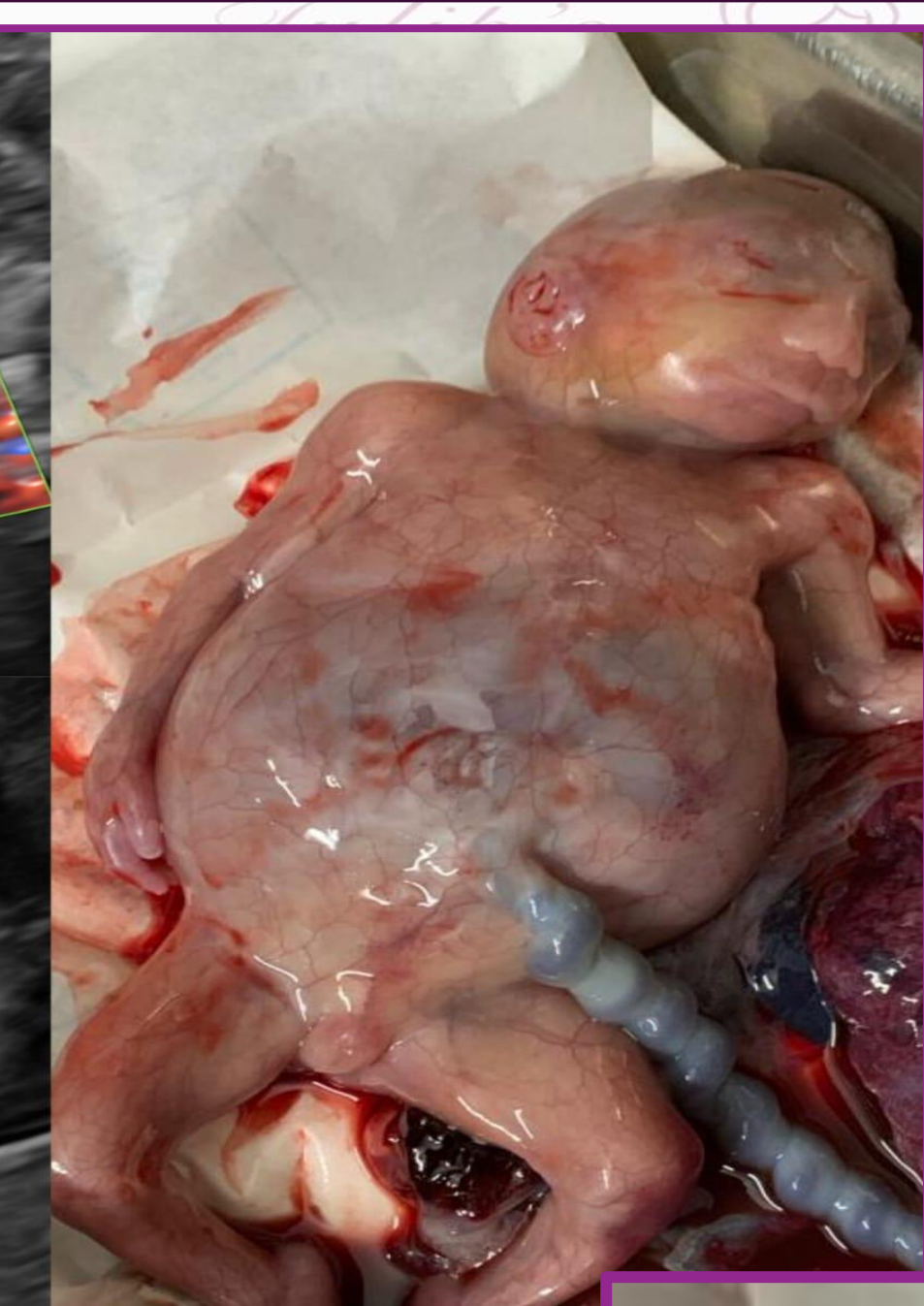


Figure 7

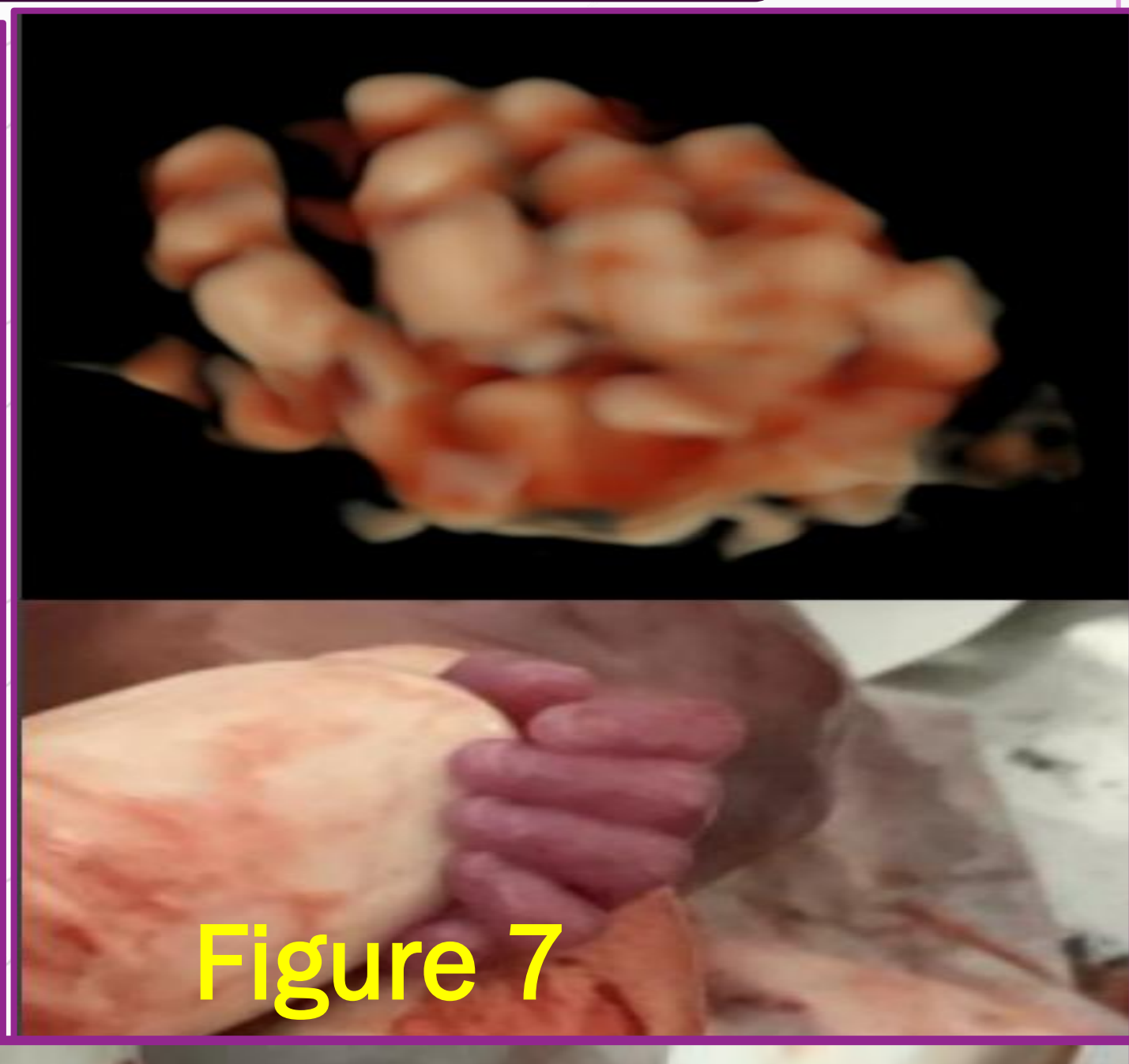


Figure 8



Figure 6

Conclusion

High index of suspicion is crucial for prenatal diagnosis of Rare Encephaloceles. Transvaginal access and 3D visualisation of the defect helps in pinpointing the diagnosis and characterisation of the defect. Since post-surgical fair outcomes are reported, a proper identification of the defect location, the neural contents involved, and also the possible association with other malformations or genetic associations should be ruled out prenatally.