



Pregnancy complicated by Spina Bifida and Edwards Syndrome: A Case Report and Review of the Literature

Rochelle Johns, MD, Maria Rojas, MD, Israel Benjamin, Ogochukwu Oseji, MD, Torri Anderson, Charelle Allen, Andrea Redmond Wilson, Jana Yancey, RDMS Andrej Bogojevic, MD, Lorraine Toner, MD, Kecia Gaither, MD, MPH, FACOG

Case Presentation

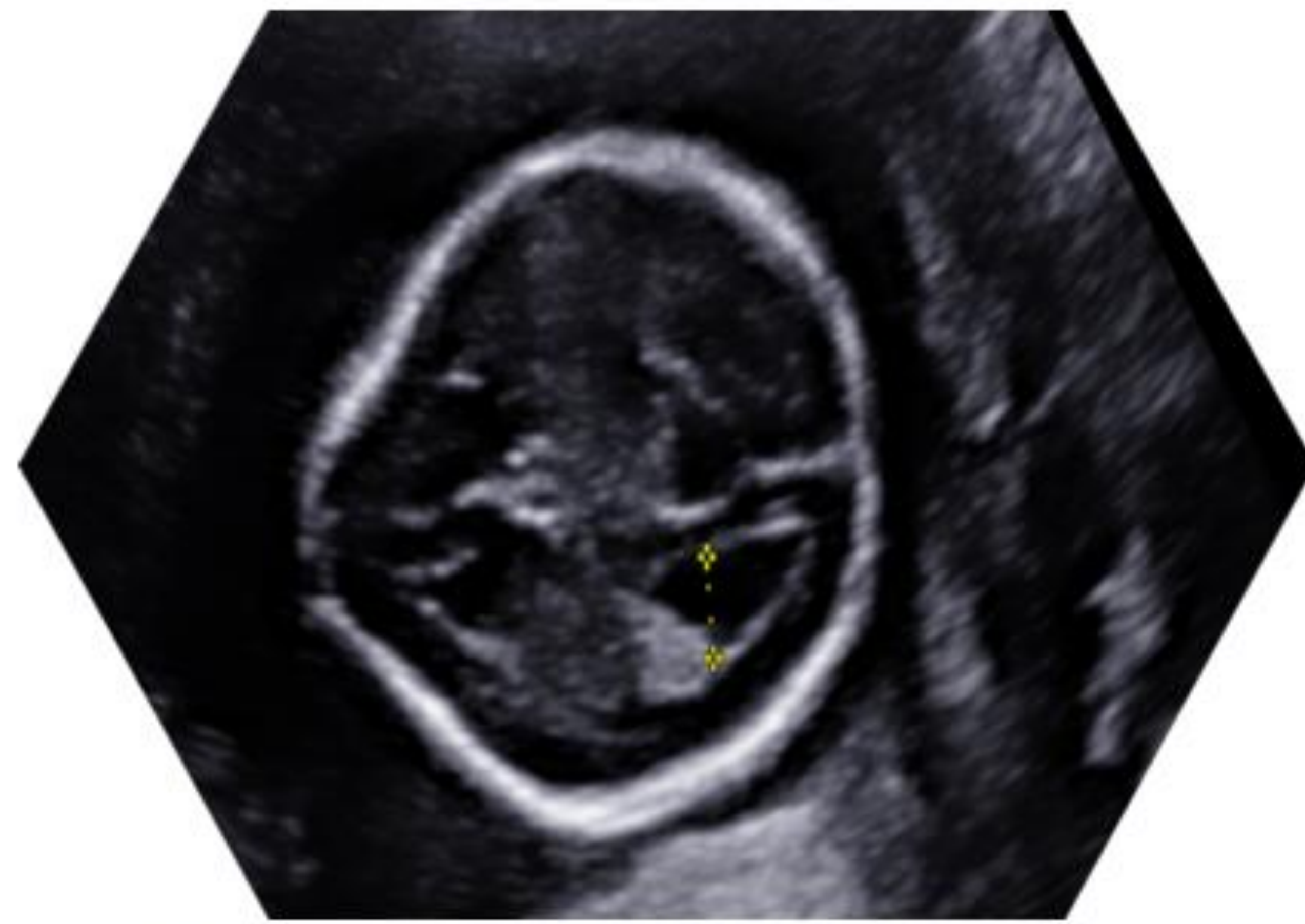


Figure 5

Ventricles appear enlarged with "dangling choroids" intracranial anatomy appears abnormal

- ▶ 37-year-old G4P2012 presented at 11wks gestation. Her prenatal history was uncomplicated; no genetic conditions or NTD's were noted. Genetic evaluation with Maternal Serum Quad screen noted an increased risk for trisomy 18, open neural tube/ventral wall defects. The patient was referred to Genetics and elected to have Non-invasive Prenatal Testing (NIPT). NIPT also suggested an increased risk for trisomy 18

- ▶ Early anatomical sonogram at 17w3d revealed a multitude of anomalies: banana sign, lemon sign, enlarged ventricles with dangling choroids, myelomeningocele and talipes of the left foot, with suspicion of a cardiac anomaly.
- ▶ Patient underwent genetic consultation and elected to have an amniocentesis at 19 weeks gestation, for definitive confirmation of the suspected aneuploidy. Fetal echocardiogram was also recommended.



Figure 1

Anatomy scan at 18w6d. "Lemon sign".

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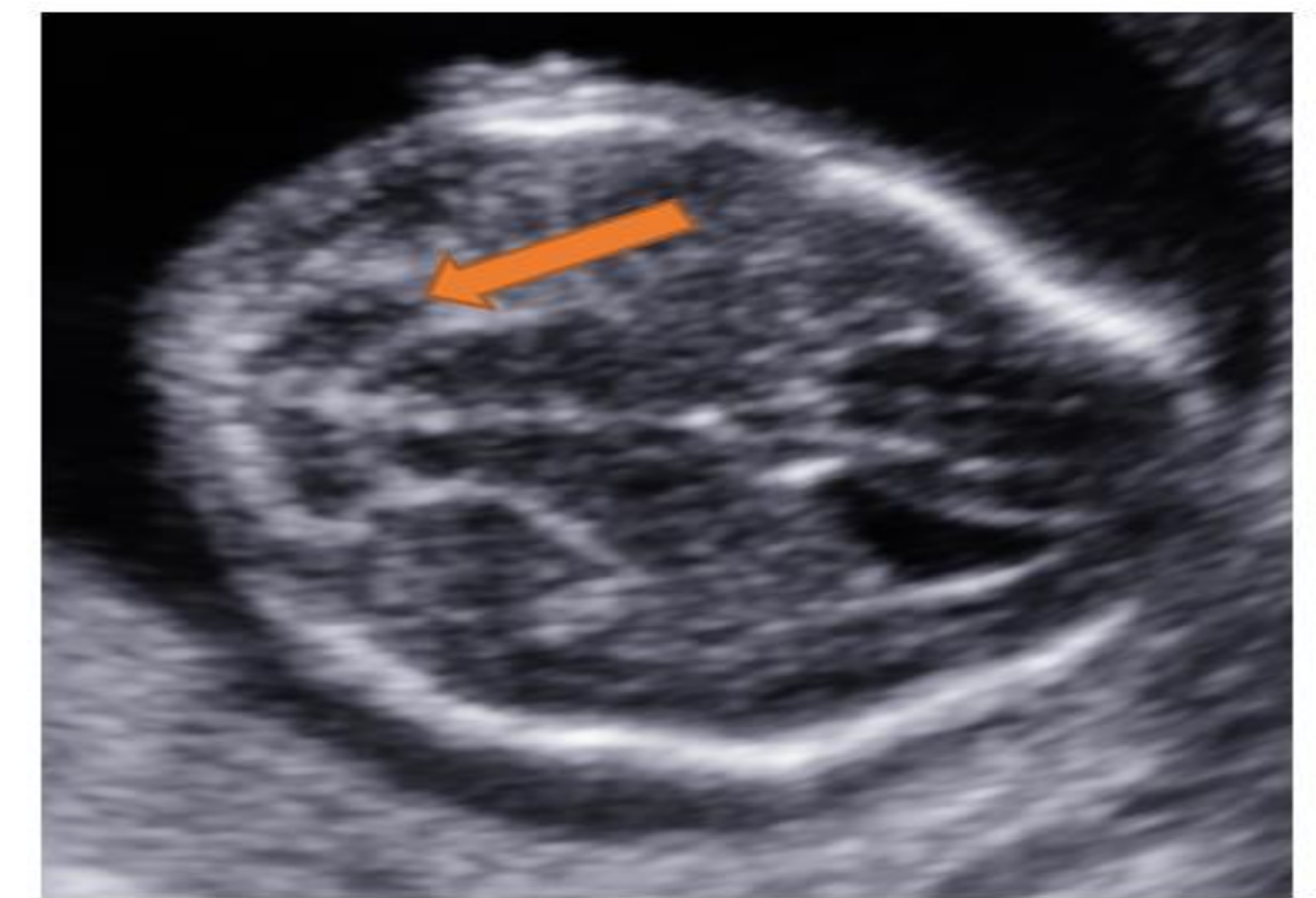


Figure 2

"Banana sign", (arrow) of the cerebellum.
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- ▶ Karyotypic evaluation revealed a male with mosaic Trisomy 18. An interdisciplinary team, consisting of Maternal Fetal Medicine Specialists, Neonatologists, and the Geneticist held multiple discussions with the patient, the result of which resulted in her electing termination. As of the writing of this report, the patient met with the Women's Options Clinic to plan for termination.

Conclusion

- The etiologies of NTD's are quite complex and multifactorial. Folate supplementation has proven to decrease the incidence of NTD's, however despite adequate supplementation these anomalies still occur. Trisomies 13 and 18 typically have a paucity of associated defects involving the CNS. Studies show the most common aneuploidy associated with myelomeningocele is trisomy 18. Technological advancements in sonography have made prenatal diagnosis of NTD's possible at earlier gestational ages. Once sonographic evaluation has been noted, full evaluation consisting of genetic consultation, amniocentesis for evaluation of aneuploidy, fetal echocardiography should be recommended. A multidisciplinary team, consisting of Neonatologists, MFM Specialists, Geneticists, and Pediatric Neurosurgeons should have a frank discourse with the parents to discuss options inclusive of termination, fetal surgery, postnatal surgery, and risk of recurrence in subsequent pregnancies.



Figure 4

Talipes of the left foot. Posted with patient permission.

References

1. Annu Rev Neurosci. 2014 ; 37: 221–242. doi:10.1146/annurev-neuro-062012-170354.
2. Birth Defects Res. 2019 November 15; 111(19): 1455–1467. doi:10.1002/bdr2.1380.
3. Childs Nerv Syst (2017) 33:1125–1141 DOI 10.1007/s00381-017-3442-x
4. Hindawi Publishing Corporation Scientifica. Volume 2017. Article ID 5364827, 29 pages <http://dx.doi.org/10.1155/2017/5364827>