

Objective

Bruck syndrome is a type of skeletal dysplasia which causes significant bone fragility (with common fractures), short long bones and joint contractures. The syndrome is an autosomal recessive form of osteogenesis imperfecta (OI), associated with collagen folding and cross-linking defects in the FKBP10 and/or PLOD2 genes¹. Consequently, the clinical features and management overlaps with that of Osteogenesis Imperfecta (OI). ’

This disorder is extremely rare and the reported incidence rate is less than one in a million and to the best of our knowledge, less than 40 cases are reported in the existing medical literature.² The prognosis is generally unfavourable with some cases being lethal due to respiratory failure (small rib cage, broken ribs etc).

Case presentation

We present a case of Bruck Syndrome, a 28 years old woman, G5P3 (with 3 previous term normal vaginal deliveries), was referred to the Feto-maternal unit after the 12 weeks ultrasound showed short limbs and micrognathia.

The ultrasound performed by the Maternal Fetal Medicine specialist one day later confirmed the findings (abnormal bones and joints and micrognathia, single umbilical artery), **suggestive of skeletal dysplasia**, and genetic testing and pregnancy management options, including termination, were discussed. However, the patient was undecided and requested genetic counselling before any further investigations. After multidisciplinary counselling, the patient requested an amniocentesis that was performed at 16 weeks. The microarray identified a **homozygous partial deletion of the PLOD 2 gene** within chromosome 3q22.1q24, compatible with **Bruck Syndrome Type 2**.

The patient decided to **continue with the pregnancy**. Serial follow up scans identified multiple fractures, severe angulation deformities of the long bones and multiple joint contractures. The patient was counselled extensively by the Neonatologist regarding the poor prognosis. A management plan for perinatal and post natal care was completed including transfer to a hospital with paediatric surgical facilities.

The patient underwent an emergency caesarean section at 35 weeks (labour, breech position) . At birth the baby had pansystolic murmur on examination and bilateral humerus and femur fractures with obvious lower limb deformity bilaterally. the baby passed away 8 weeks later due to increasing ventilatory requirements and cardiac failure.

Figure 1 Short fracture of radius and ulna

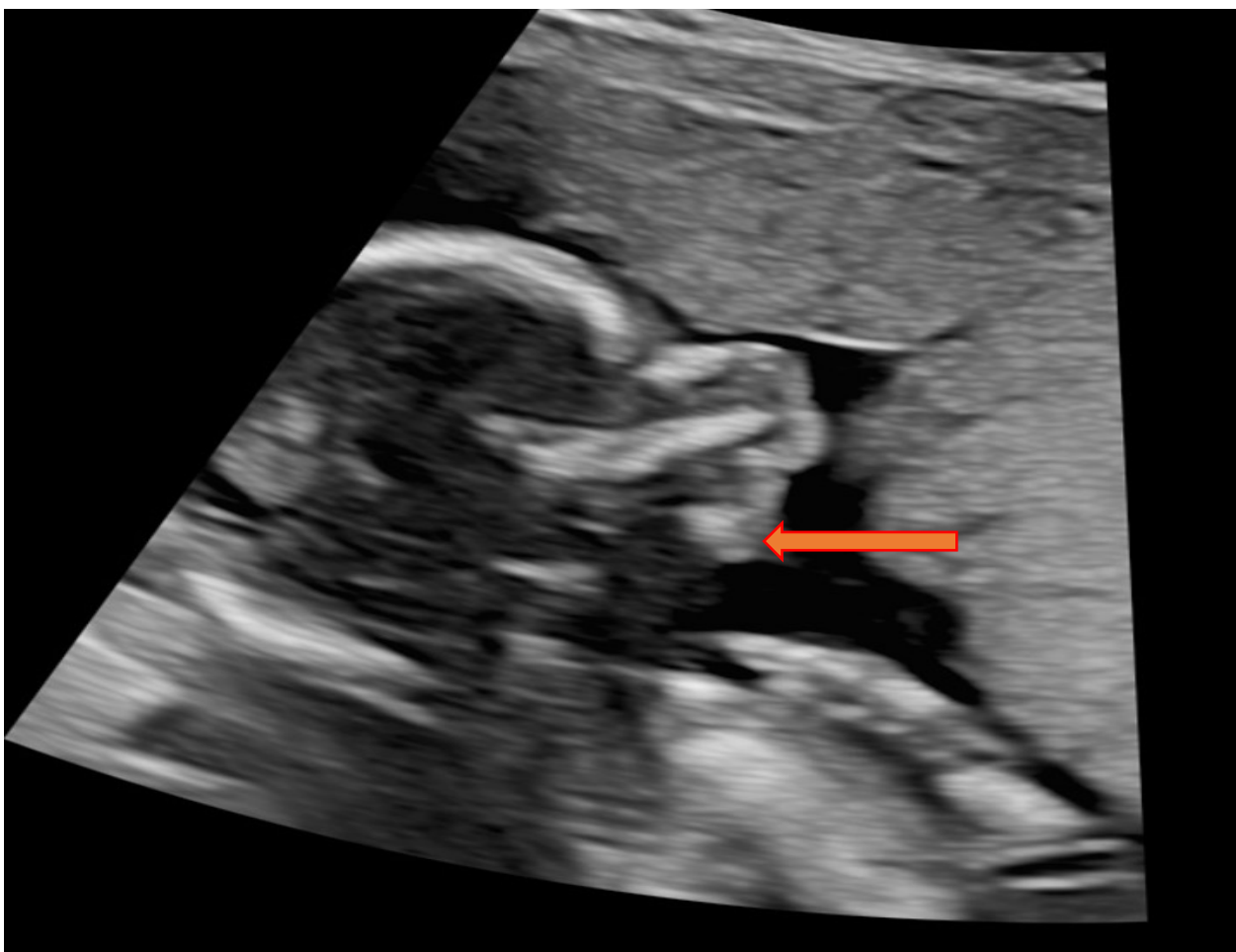


Figure 2: Features of micrognathia

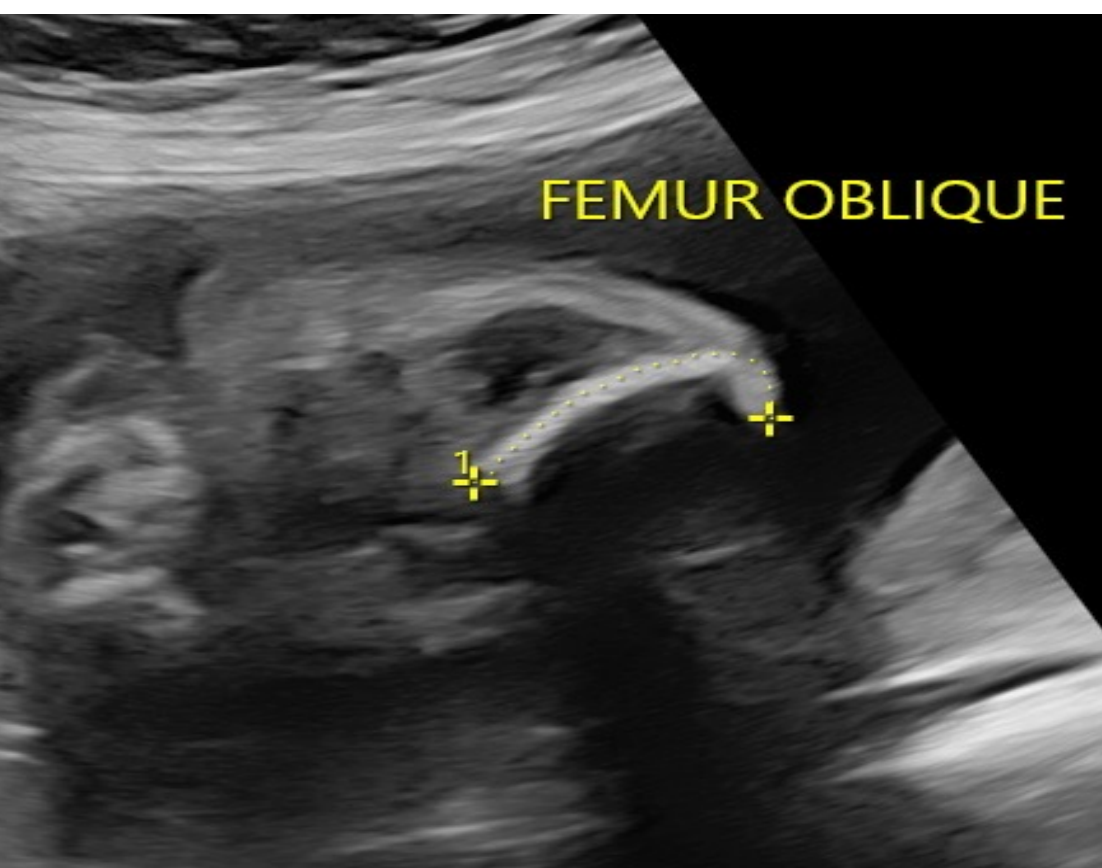


Figure 3. An oblique femur



Figure 4. Cardiomegaly with large right atrium

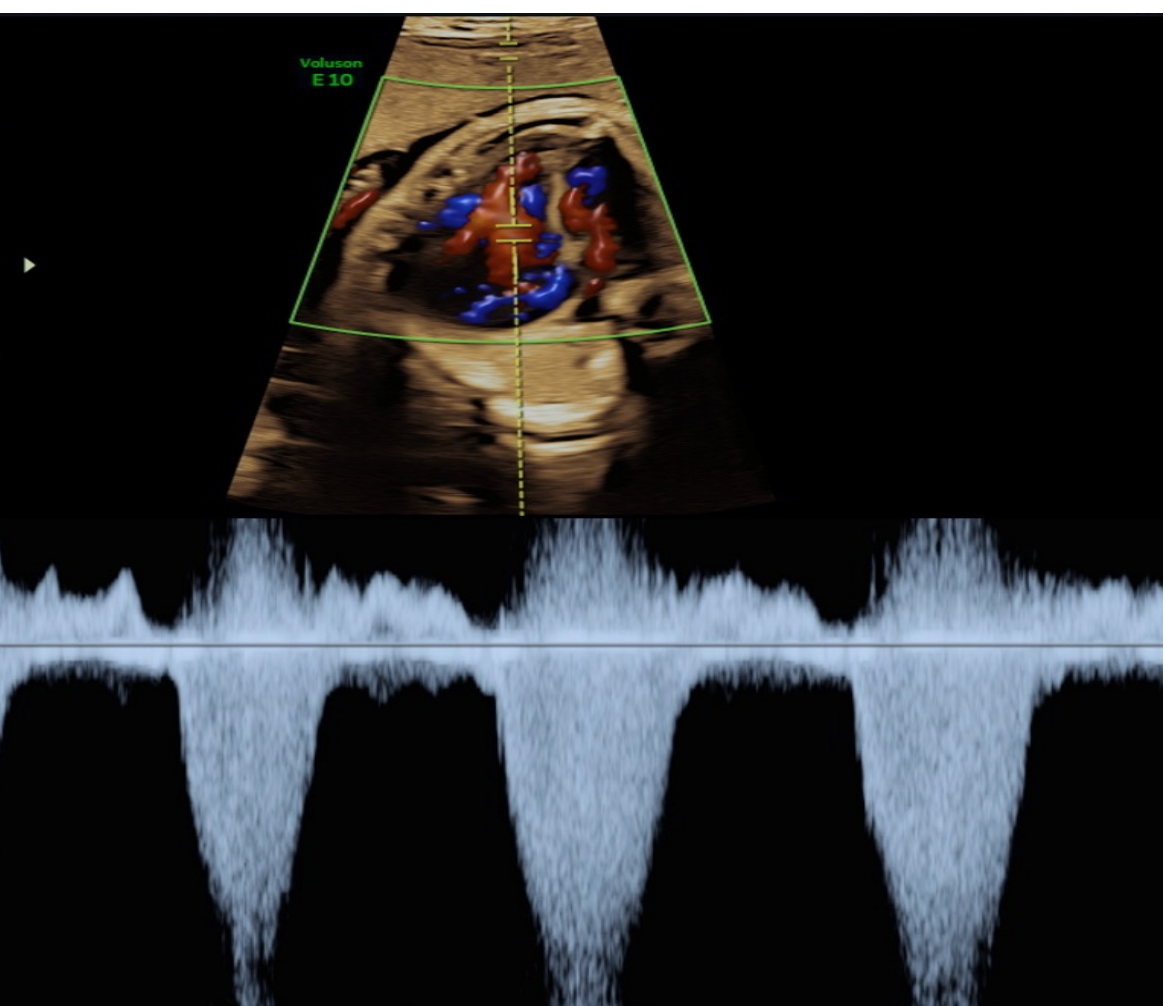


Figure 5. Tricuspid regurgitation

Discussion

This case illustrates the natural history of a rare type of skeletal dysplasia syndrome.

The perinatal management maybe challenging as a lot of the medical professionals may not be familiar with these rare conditions and special requirements. The parental genetic testing showed that the parents were asymptomatic heterozygous carriers of the conditions, so the risk recurrence was estimated as 25%. Options for testing pre or during future pregnancies were discussed

Conclusion

The diagnosis of Bruck syndrome is based on ultrasound findings combined with genetic testing. This emphasises the importance of doing an early fetal anatomy survey to allow the most adequate perinatal and postnatal care, including offering the option of termination of pregnancy in countries where it’s allowed. Until date there is no available prevention or treatment for the condition. Treatments are usually directed towards alleviating the symptoms: orthopaedic therapy, fracture management and bisphosphonate drugs are often used.

We hope that our case report can be useful to add information about the natural history of this rare condition.

References

- Steven Mumm : Bruck Syndrome 2 Variant Lacking Congenital Contractures And Involving A Novel Compound Heterozygous *PLOD2* Mutation Bone Volume 130 January 2020, 115047
- Parvin S, *et al. BMJ Case Rep* 2021;**14**:e246786. doi:10.1136/bcr-2021-246786