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A unique case of prenatal diagnosis of vascular Ehlers-Danlos syndrome

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Introduction

Ehlers-Danlos Syndrome (EDS) is a group of hereditary connective tissue disorders caused by genetic mutations affecting collagen synthesis and structure. Diagnosis typically occurs in childhood when characteristic signs and symptoms emerge. Prenatal diagnosis is usually performed in families with a known history of EDS, identifying the specific mutation either in the preimplantation phase or through invasive testing. Prenatal diagnosis of EDS via ultrasound remains limited due to the absence of universally recognized markers. Consequently, in cases without a known family history or identified mutation, prenatal diagnosis can be challenging.

Our case

We present a case where ultrasound at 16+2 weeks revealed **trigonocephaly** and **micrognathia**. Amniocentesis showed a normal male karyotype and Array-CGH. At 19+3 weeks, the fetus experienced a late miscarriage. Histological examination of the fetus, limited by autolysis, did not detect evident malformations.

However, placental histology revealed a slightly reduced-weight placenta with findings suggestive of **maternal and fetal malperfusion** and superficial implantation. Given the normal karyotype and Array-CGH results, exome sequencing was conducted, identifying a new **de novo heterozygous variant in COL3A1**, associated with vascular EDS (vEDS), never previously reported in the literature. Parental testing confirmed the absence of this variant.

Conclusion

This case represents a rare instance of **prenatal diagnosis of vEDS without a family history**. The suspicion of a genetic syndrome arose from an incidental ultrasound finding of a facial anomaly—previously associated with vEDS in adulthood but never described prenatally. Additionally, the possible link between **placental implantation, feto-maternal malperfusion**, and altered type III collagen in vEDS fetuses offers a novel perspective.



Figure 1. Fetal micrognathia.

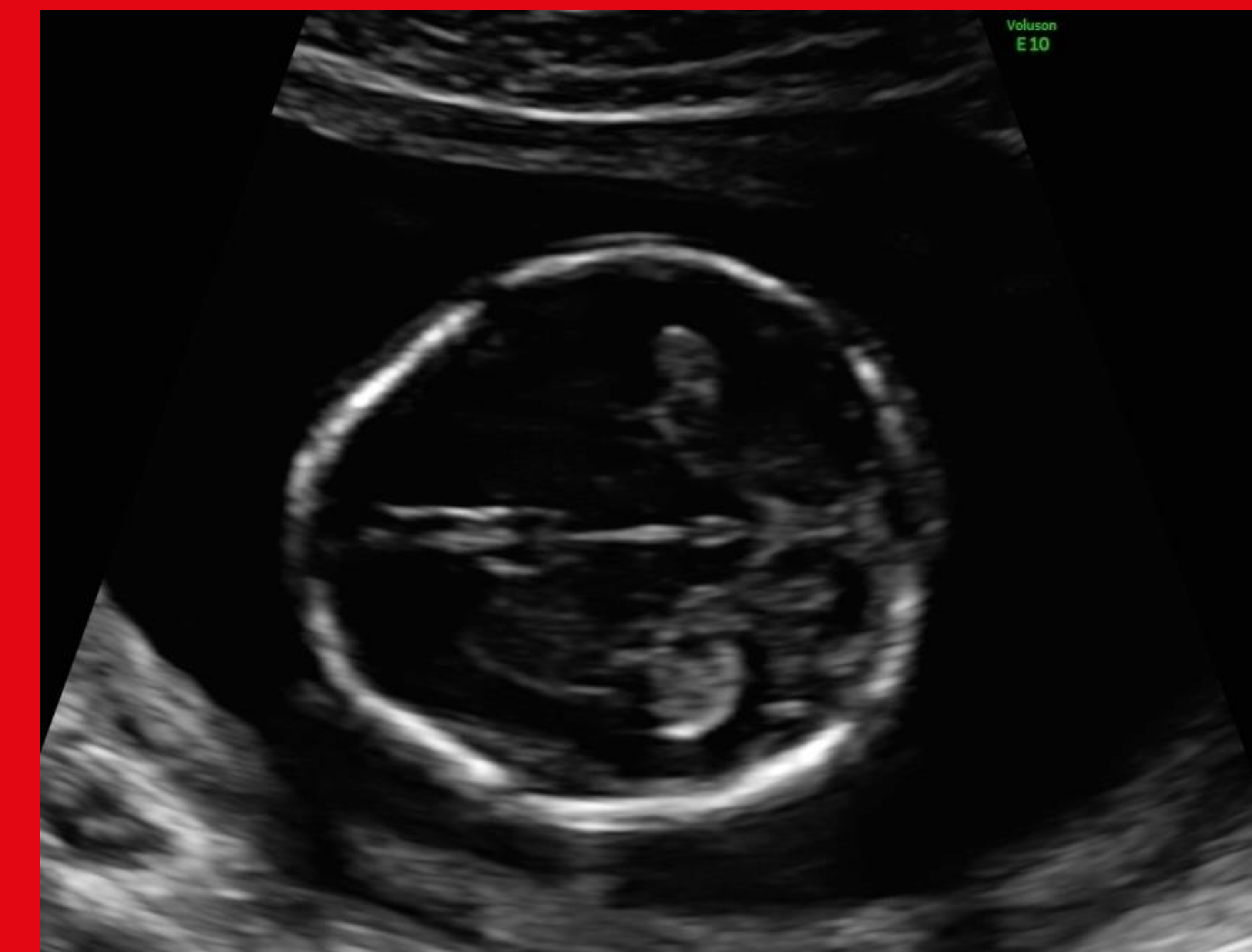


Figure 2. Fetal trigonocephaly.