

EP05.12 Non-rhizomelic chondrodysplasia punctata and Binder phenotype secondary to severe maternal vitamin K deficiency

O. Nieto¹, M. Recio², R. Revello¹, A. Garicano¹, M. Cañamares¹, J. A. Rodríguez León¹, S. Marín¹, P. Arias¹

1. Prenatal Diagnosis, Obstetrics and Gynaecology · 2. Radiology — Hospital Universitario Quirónsalud Madrid, Madrid, Spain

Introduction

Chondrodysplasia punctata (CDP) is a heterogeneous group of disorders characterised by punctiform calcifications in cartilage and periarticular soft tissues. Binder phenotype (maxillonasal dysplasia) is one of its facial expressions. We report a case of non-rhizomelic CDP with Binder phenotype caused by severe maternal vitamin K deficiency.

Prenatal findings

A 42-year-old primigravida. Routine ultrasound at 21 weeks of gestation revealed a fetus with a significantly flat profile, short nose and depressed nasal bridge, consistent with Binder phenotype. Stippled calcifications were identified in the tarsal and sacral regions.

Genetic work-up

Differential diagnosis included chromosomal anomalies and metabolic peroxisomal disorders. Amniocentesis yielded normal results for karyotype, array-CGH and clinical exome sequencing, excluding primary genetic causes.

Maternal work-up

With primary genetic causes excluded, maternal screening for teratogens and metabolic deficiencies was conducted, revealing severe maternal vitamin K deficiency.

Outcome

Intrauterine fetal death occurred at 22 weeks. Induction of labour was complicated by maternal hyperthermia and severe postpartum haemorrhage requiring vitamin K administration, curettage, Bakri balloon placement and blood transfusion. Postmortem CT confirmed maxillonasal dysplasia and skeletal calcifications.

Conclusion

Maternal vitamin K status should be investigated in fetuses with stippled epiphyses and midfacial hypoplasia when genetic testing is inconclusive: maternal malabsorption can mimic genetic forms of chondrodysplasia punctata.

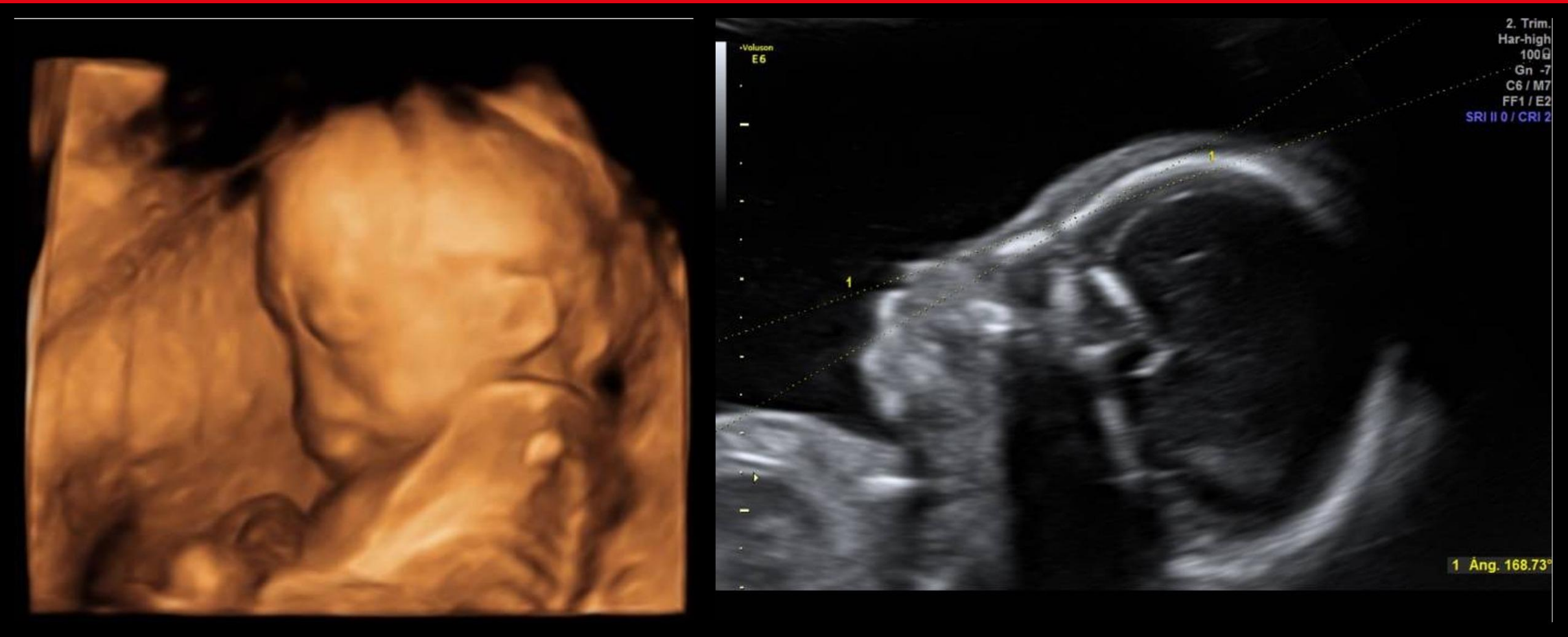


Figure 1. Fetal profile at 21 weeks: 3D and 2D mid-sagittal views showing the Binder phenotype.

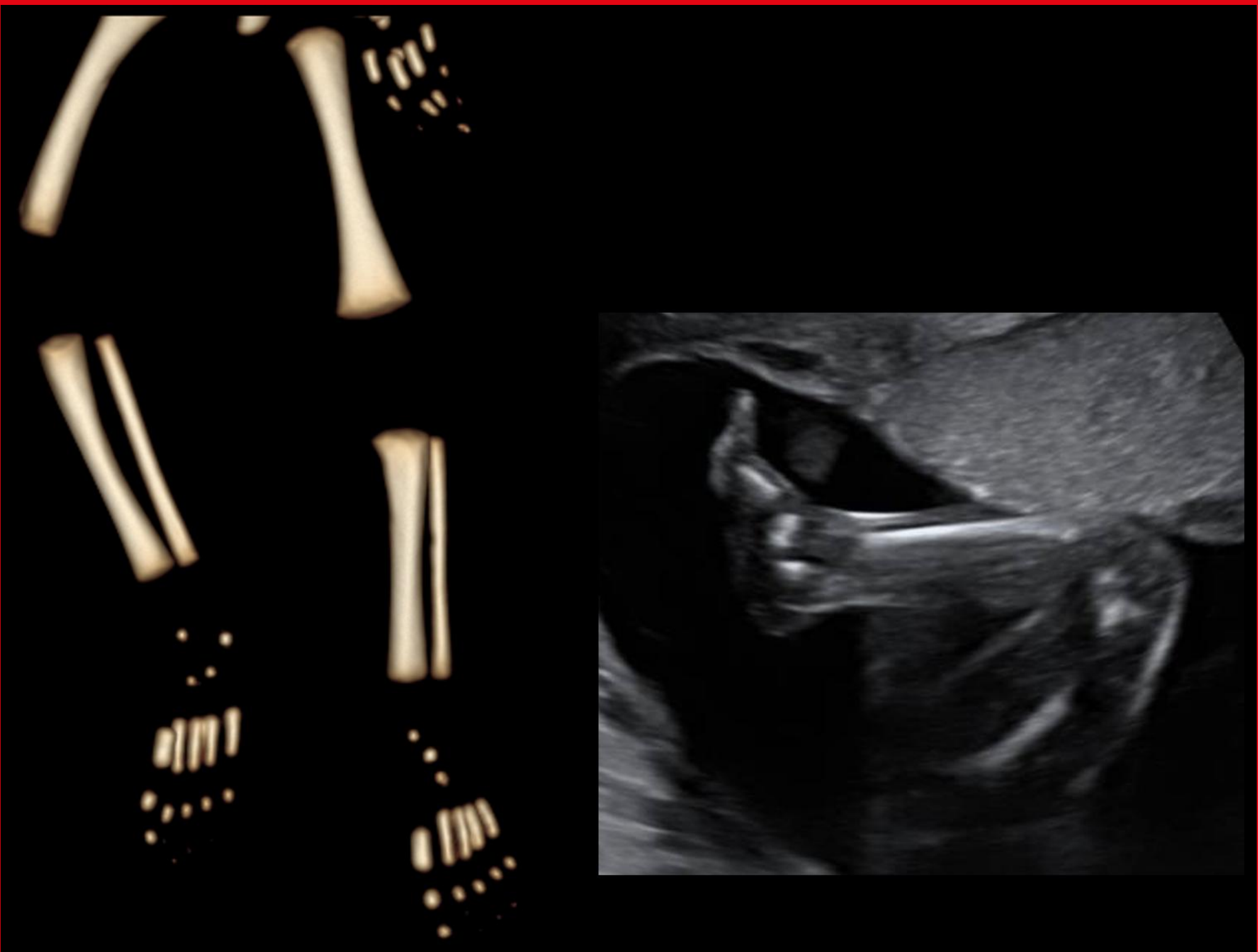


Figure 2. Postmortem CT (left) and prenatal ultrasound (right): stippled periarticular calcifications.



Figure 3. Postmortem CT 3D reconstruction: midfacial hypoplasia and stippled skeletal calcifications.