

Poster number 0be0107

PRENATAL SIGNS OF CARTILAGE HAIR HYPOPLASIA OVERLAPPING PHENOTYPE WITH OMENN SYNDROME

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Introduction

Cartilage Hair Hypoplasia (CHH) is a rare autosomal recessive metaphyseal dysplasia, characterized by short stature, hypotrichosis, and extra-skeletal manifestations. Neonatal erythroderma (NE) is a skin inflammation associated with Omenn Syndrome (OS), inflammatory condition with severe combined immunodeficiency (SCID).

Case Presentation

We report a case of a 30-year-old Asian mother, gravida 4 para 2, consanguineous couple, who was referred to our hospital at 19+5 weeks gestation due to a shortened femur. Micromelia was diagnosed, thoracic diameter and cranial circumference were normal. Skeletal dysplasia was suspected, invasive tests were declined. At 21+6 weeks, lower limb rhizomelia was diagnosed, and tibia and fibula measurements fell below the 5th percentile by 26+6 weeks.

Subsequent scans up to 33+1 weeks showed dilated cardiomyopathy, nuchal edema, and abnormal septum pellucidum. Multidisciplinary counseling was provided. At 34+4 weeks, a cesarean delivery was performed due to preterm rupture of membranes, with Apgar scores of 1, 3, and 4 at 1°, 5°, and 10° minutes, respectively. At birth, the infant's auxological data were: weight 2130 g (38th percentile; -0.30 SD), length 40 cm (< 1st percentile; -2.59 SD), and head circumference 33 cm (89th percentile; 1.28 SD). The proband presented with rhizomelic limb shortening and severe erythroderma. Hair and eyebrow loss appeared in the first week. X-ray confirmed rhizomelia and dumbbell-shaped femurs. Genetic tests revealed a homozygous NR_003051(RMRP):c.C35A pathogenic variant in the RMRP gene, confirming Omenn Syndrome in a patient with CHH. Parents were heterozygous for the mutation.

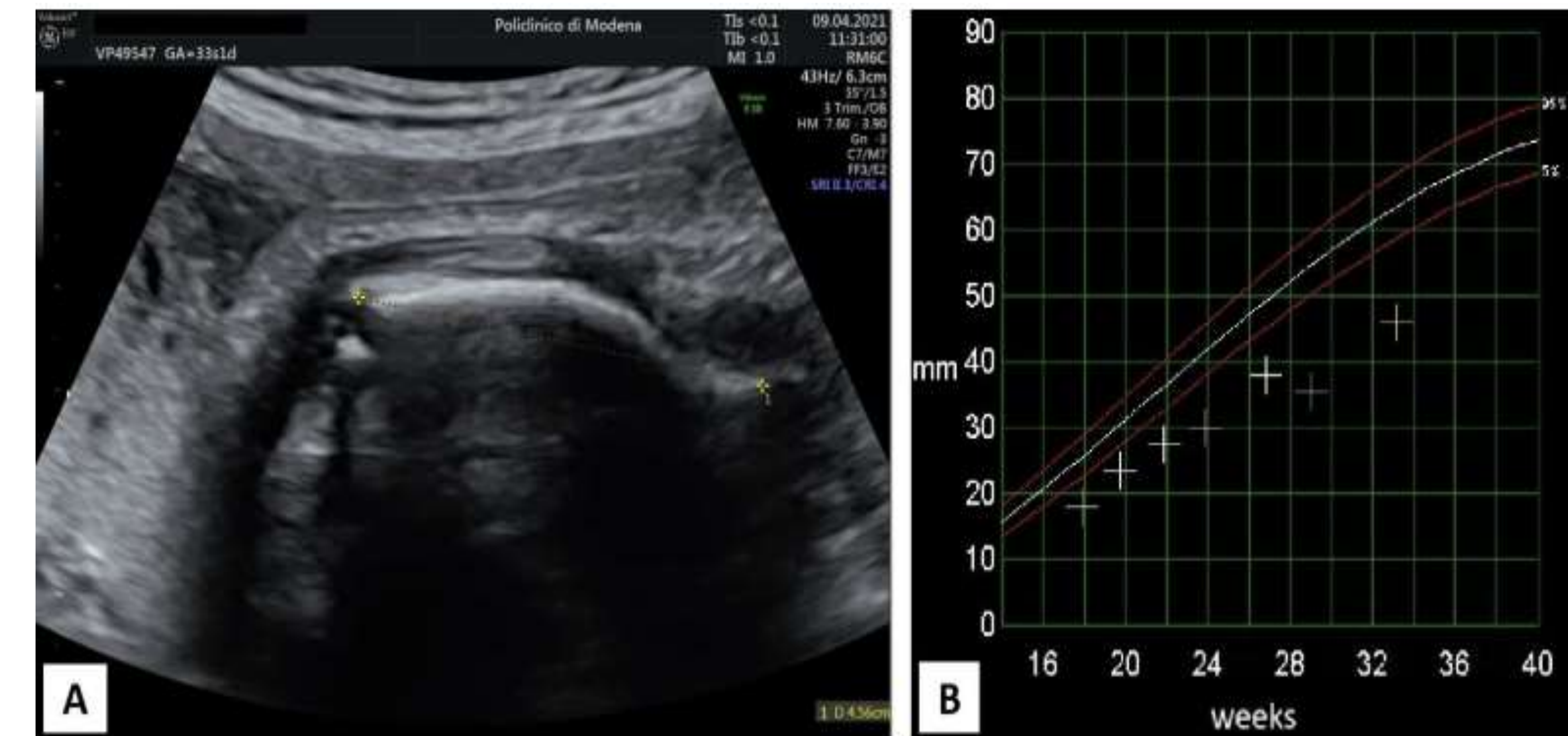


Figure 1. A. Shortened and bowed femur at 33+1 weeks B. Femur growth curve from 19+5 to 33+1 weeks

Conclusion

This case highlights the overlap between CHH and OS, emphasizing the need for RMRP gene analysis in fetuses with skeletal dysplasia and newborns with SCID-like features.