AUTOSOMAL RECESSIVE OMODYSPLASIA: A RARE CAUSE OF DISPROPORTIONATE SHORT STATURE

Cristiane Kopacek (1), Luciana Amorim Beltrão (2), Victória Bernardes Guimarães (2), Julia Santana Trombetta (2), Karen Lizeth Puma Lliguin (2), Vinicius de Souza (2), Géssica Haubert (2), André Campos da Cunha (1), Paulo Ricardo Gazzola Zen (2), Rafael Fabiano Machado Rosa (1,2).

(1) Hospital Materno Infantil Presidente Vargas (HMIPV), Porto Alegre, RS, Brazil;
(2) Universidade Federal de Ciências da Saúde de Porto Alegre (UFCSPA), Porto Alegre, RS, Brazil.

BACKGROUND

Autosomal recessive omodysplasia is considered a rare skeletal dysplasia characterized by severe micromelia with shortening and distal tapering of the humeri and femora.

OBJECTIVE AND HYPOHESIS

To report the prenatal findings of a patient with autosomal recessive omodysplasia, a rare condition characterized by disproportionate short stature.

CASE REPORT

The pregnant woman presented 38 years and she was in her seventh pregnancy. She was initially assessed in fetal medicine at the hospital at 29 weeks and 6 days of gestation due to obstetric ultrasound showing short femur and humerus.

The pregnancy was uneventful. Her husband presented 40 years of age and was healthy and non-consanguineous. Family history was positive for a maternal uncle with nanism.

Fetal ultrasound performed at 29 weeks and 6 days of pregnancy showed reduced amniotic fluid and important rhizomelic shortening of the limbs. The humerus measured 1.8 cm and the femur 2.4 cm. The hands and feet, as well as the face and thorax seemed normal. Bone mineralization was also normal (Figure 1).

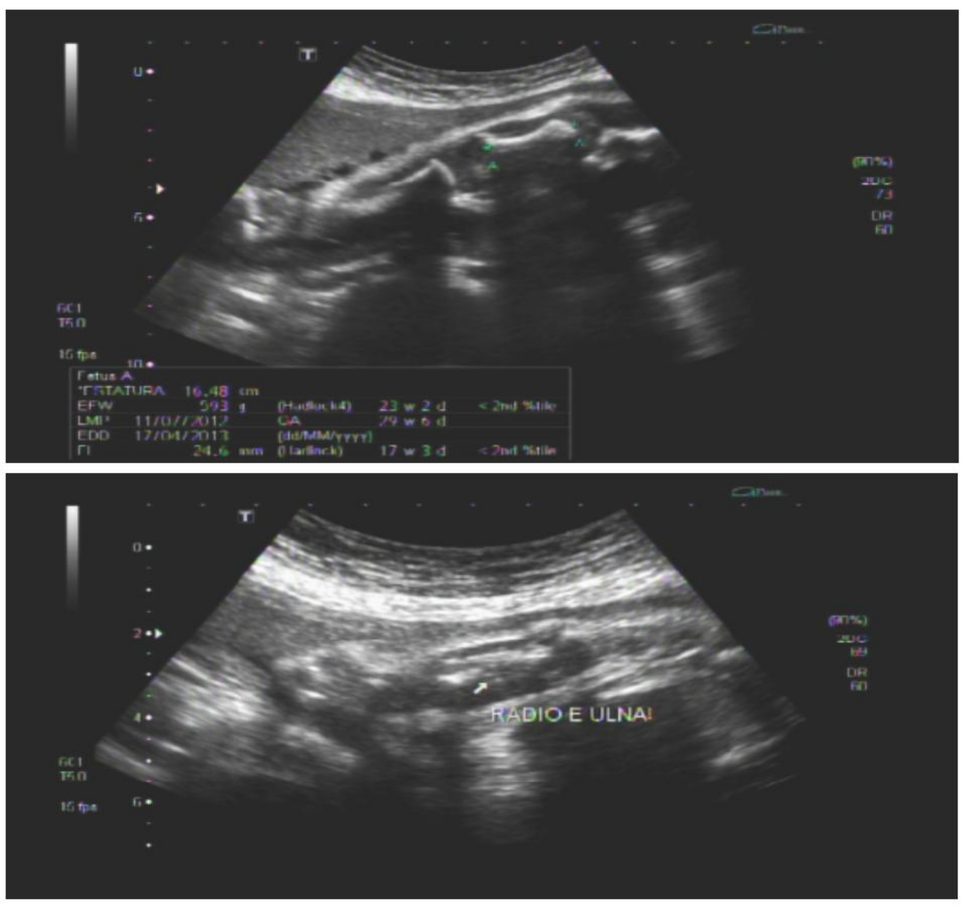

Figure 1. Fetal ultrasound at 29 weeks of pregnancy showing the shortening of the long bones.

At this point, achondroplasia/hypochondroplasia emerged as diagnostic hypotheses. GTG-Banding karyotype performed through cordocentesis revealed a normal chromosomal constitution (46,XY). Fetal echocardiography was also normal. The ultrasound performed at 35 weeks and 6 days of gestation revealed femur measuring 3.4 cm. The estimated fetal weight was 1,441 grams.

The patient was born through vaginal delivery, at 37 weeks and 4 days of gestation, weighing 2,320 grams, measuring 40 cm, with head circumference of 33 cm and Apgar scores of 9 at first minute and 10 at fifth. He presented micromelia with important rhizomelic shortening of the upper and lower limbs, normal thorax and some dysmorphia: nevus flammeus at nose and glabella, small mouth, micrognathia, small ears with overfolded helix, bilateral single palmar crease and cryptochid testis. These clinical data added to the radiological features (that included radial head dislocations) were consistent with the diagnosis of autosomal recessive omodysplasia (Figures 2 and 3).

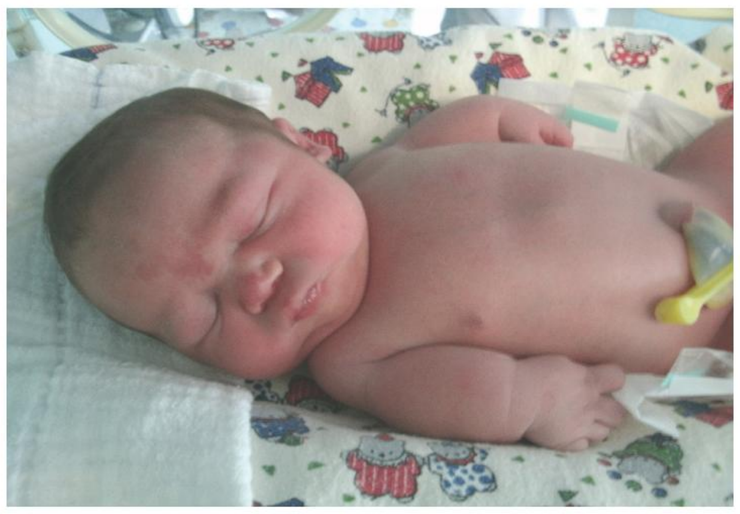

Figure 2. Postnatal appearance of the patient. Note the important rhizomelic shortening of the upper limbs.

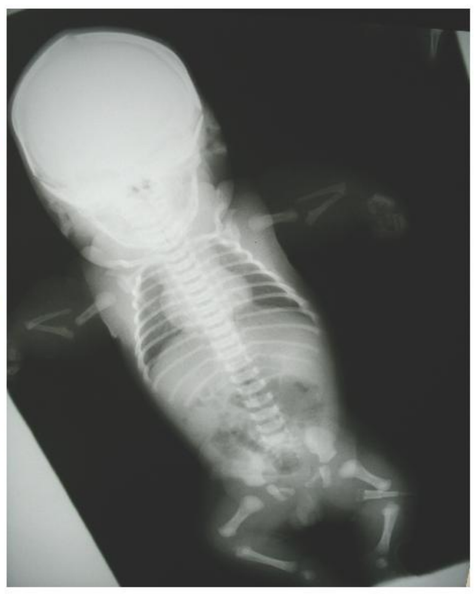

Figure 3. Postnatal radiography showing the important rhizomelic shortening of the limbs and the radial head dislocations.

CONCLUSIONS

There are few reports in the literature of prenatal features of patients with this genetic condition. In prenatal, autosomal recessive omodysplasia has also been confused with other syndromes, as diastrophic dysplasia or even hypo/achondroplasia, due to the similarity of the findings. The definition of the diagnosis has important implications over the genetic counseling and patient management.

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CRISTIANE KOPACEK
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