

A rare case of neonatal dwarfism – Question

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The answers can be found in the following article:

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Case summary

I. was born from a 34-year-old woman at the 35th week of gestational age, from an elective cesarean section due to premature rupture of membranes and suspected fetal malformation with polyhydramnios.

During a follow-up prenatal ultrasound at the 28th week of gestational age, a marked reduction in the length of the long bones, which also appeared curved, was detected. The lower thoracic diameter was normal for gestational age, while the upper chest was abnormally narrow and flat on sagittal scans. These elements lead to a first suspicion of Jeune Syndrome (asphyxiating thoracic dystrophy).

At birth, the baby showed a picture of asphyxia with 4 on Apgar score, therefore an endotracheal intubation was immediately performed, and the baby was quickly transported to the Neonatal Intensive Care Unit with an Ambu® bag.

On admission, the general conditions of the baby were compromised, because of a pronounced respiratory effort, with intercostals and diaphragmatic retractions, tachypnea, decreased breath sounds and widespread rales, so the patient was subjected to mechanical ventilation and oxygen therapy.

The anthropometric data were: weight 2,100 g (-0.98 SDs), length 34 cm (-4.48 SDs), head circumference 34.5 cm (+1.49 SDs).

On physical examination (**Fig. 1** and **Fig. 2**), it was highlighted a large head with prominent frontal bones, open normotensive bregmatic fontanelle, bell-shaped chest, narrow in the upper half and flared at the base, and prominent abdomen; the limbs were shortened, curved and stocky, thighs were adducted and externally rotated, arms extended, hands and feet were small and stocky. Cutaneous and subcutaneous tissues were well represented, with evident overabundant skin folds of the limbs and at the base of the neck, which moreover was shorter than normal.

A baby-gram radiography with the whole body of the newborn was performed (**Fig. 3**).



Figure 1. The limbs were shortened, curved and stocky, hands were small and stocky.



Figure 2. It was highlighted a large head with prominent frontal bones, open normotensive bregmatic fontanelle, bell-shaped chest, narrow in the upper half and flared at the base, and prominent abdomen. The limbs were shortened, curved and stocky, thighs were adducted and externally rotated, arms extended, hands and feet were small and stocky. Cutaneous and subcutaneous tissue were well represented, with evident overabundant skin folds of the limbs and at the base of the neck, which moreover was shorter than normal.



Figure 3. Baby-gram radiography with the whole body of the newborn.

Questions

1. What do you see in the baby-gram X-ray?
2. What is your diagnosis?

Declaration of interest

The Authors declare that there is no conflict of interest.