

## A Rare Malformation of the Limbs

Raquel Oliveira, Pedro Marinho, Suzana Figueiredo, Sérgio Mendanha

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Newborn female, first born of non-consanguineous parents with a family history of ectro / syndactyly of the hands and feet present in the paternal lineage (father, grandfather and multiple relatives in second and third degree), without an established diagnosis. Her mother had a history of corrected cleft palate and bilaterally impaired auditory acuity. The pregnancy was uneventful and prenatal ultrasounds were described as normal. Vaginal delivery occurred at a gestational age of 41 weeks, with an Apgar score of 9/10. Weight and length were appropriate for gestational age. At birth, she presented with an absent first finger, clinodactyly of the second finger, and syndactyly of third and fourth fingers of the left hand. The right hand presented with syndactyly of the third and fourth fingers, an absent middle and distal phalanx of the second finger and absent distal phalanx of the first finger (Fig. 1). Both feet showed obvious median clefts, like a “lobster claw” (Fig. 2). The malformations were not diagnosed in the prenatal evaluation. Other malformations were excluded after a multidisciplinary evaluation.

The child is currently 4 years old, always presented an adequate development, with the progressive acquisition of motor and behavioral skills and with no sensorineural hearing loss. She has no limitations in her daily living, showing no need for corrective surgery, until now.



**Figure 1.** Left and right hand showing ectrodactyly malformation and the respective X-ray showing bone malformations.



**Figure 2.** Median clefts of feet, like a “lobster claw”, and the respective X-ray showing the bone malformations.

Split hand / foot malformation is a congenital limb malformation affecting predominantly the central rays of the hands and/or feet. At least seven split hand / foot malformation types have been distinguished in the literature. The majority undergoes autosomal dominant mode of inheritance with reduced penetrance but autosomal recessive, as well as X-linked inheritance have also been reported. It can occur as isolated malformations or as part of various complex syndromes.<sup>1-3</sup>

This case report aims to raise the awareness for the importance of first trimester ultrasound in prenatal diagnosis of malformations.

**Keywords:** Child; Infant, Newborn; Limb Deformities, Congenital

### WHAT THIS REPORT ADDS

- Raises the awareness for the importance of first trimester ultrasound in prenatal diagnosis of malformations.
- Split hand / foot malformation is a rare congenital limb malformation and its detection in prenatal ultrasounds is important for the psychological preparation of the parents.
- The diagnosis is clinical, and split hand / foot malformation is a heterogeneous abnormality, ranging from a relatively mild defect to a severe form or part of a multiple congenital anomaly syndrome. Consequently, interindividual and intrafamilial variability of the split hand / foot malformation is very high. Follow-up with multidisciplinary evaluation should be provided.
- The genetic study is of practical importance for the entire family. It allows the screening of at-risk individuals and it is important for a correct genetic counselling and for prenatal and pre-implantation diagnosis.

Pediatrics Department, Unidade Local de Saúde do Alto Minho, Viana do Castelo, Portugal

#### Corresponding Author

Raquel Oliveira

[raqueloliveira07@gmail.com](mailto:raqueloliveira07@gmail.com)

Department of Pediatrics, Unidade Local de Saúde do Alto Minho, Estrada da Santa Luzia, 4901-858 Viana do Castelo, Portugal

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### Confidentiality of data

The authors declare that they have followed the protocols of their work centre on the publication of patient data.

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