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Case report

Acro-callous syndrome: A case report

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ABSTRACT

A malformative syndrome of unknown prevalence, the diagnosis is based on morphological ultrasound and magnetic resonance imaging of the fetus. The transmission is genetic and autosomal recessive. Courtnes has established 4 diagnostic criteria to affirm the acrocal syndrome and to eliminate the amalgam with other polymalformative syndrome.

1. Introduction and importance

Acrocallous syndrome is a rare polymatformative syndrome defined by agenesis of the corpus callosum, distal limb anomalies, craniofacial anomalies of varying severity and intellectual deficit, although the absence of mental retardation has been reported in 3 cases worldwide [1]. Its prevalence is not well known, but the number of reported cases is increasing after the first description in 1979, by Schinzel and Schimid. The work has been reported with respect to the SCARE 2020 criteria [2].

2. Case presentation

We report the case of an acrocallous syndrome diagnosed on morphological ultrasound. This was a well-monitored pregnancy in a primiparous 31-year-old woman of Moroccan origin, with no notable pathological history and no history of drug use. There was consanguinity. Morphological ultrasound revealed a double malformation: complete agenesis of the corpus callosum (Fig. 1) associated with a bilateral cleft lip and palate (Fig. 2), the rest of the examination did not reveal any other malformation. The karyotype has shown a trisomy 13. The whole associated with an intrauterine growth retardation. Extraction was performed by cesarean section following the observation of a pathological pattern during labor. The clinical examination of the newborn has objectified the presence of a bilateral cleft lip and palate (Fig. 3).

3. Clinical discussion

The acrocallous syndrome is a rare malformation, only about forty cases have been described worldwide and nowadays. It is described as an autosomal recessive mode of inheritance, consanguinity is found in the majority of reported cases [3]. The responsible gene is not yet known, however Pfeifer et al. have conducted studies incriminating chromosome 12p [4], recently studies involving mutations of the genes of kinesis KIF (15q26.1) and transcription factor Gli3 (7p14.1), these two genes are involved in the ciliary signaling pathway Sonic Hedgehog and their mutation would influence the early development of medial structures [5]. The recurrence rate is estimated at 25% in subsequent pregnancies, hence the interest of antenatal diagnosis. However, since acrocallous syndrome does not represent a lethal risk, medical termination of pregnancy is not recommended in our country [6]. There is a phenotypic variability of expression making the diagnosis difficult and posing the problem of differential diagnosis with several malformative syndromes namely: Greig syndrome, oro-facial-digital syndrome, Meckel Gruber syndrome and many other malformative entities. It is to underline the interest of the diagnostic criteria proposed by Courtens which make the diagnosis if at least 3 of the 4 following criteria are met and which are: absence of the corpus callosum, craniofacial dysmorphia, polydactilia and psychomotor retardation [7]. Our case met the first 3 diagnostic criteria. It should be noted that the absence of psychomotor delay does not exclude the diagnosis. Finally, the prognosis of the acrocallous syndrome depends essentially on the importance of hypotonia and the early appearance of convulsions, in addition to the severity of the malformations.

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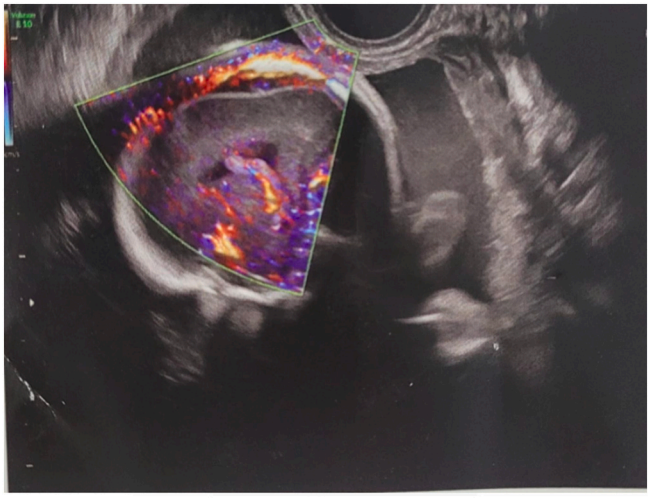

Fig. 1. Complete agenesis of the corpus callosum.

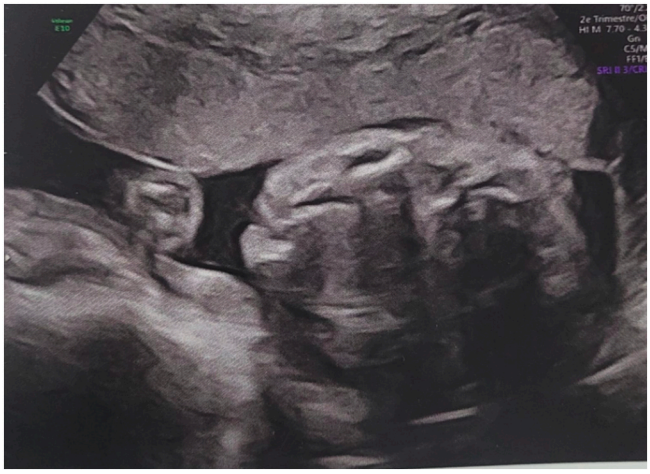

Fig. 2. Morphological US showing a bilateral cleft lip and palate.

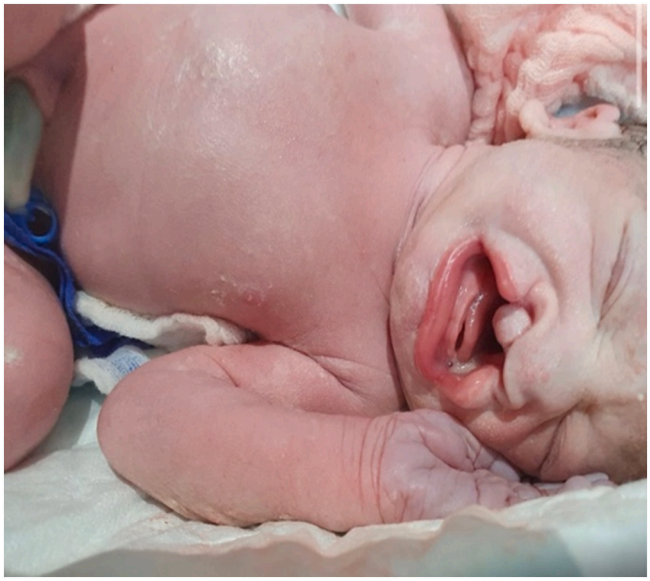

Fig. 3. Fetus with a bilateral cleft lip and palate.

4. Conclusion

Prenatal diagnosis is based on morphological ultrasound from 20 weeks of amenorrhea and also on magnetic resonance imaging of the fetus. Molecular analysis can be proposed if the mutation of the kinesis gene or the transcription factor has been identified in an affected sibling. Management is based on surgical correction of apparent malformations and monitoring of psychomotor development and the occurrence of seizures, as well as supportive and stimulating therapy within a specialized program.

Consent

Written informed consent for publication of their clinical details and/or clinical images was obtained from the patient.

Ethical approval

I declare on my honor that the ethical approval has been exempted by my establishment.

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Guarantor

Dr Lmrissi Amine.

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CRediT authorship contribution statement

Lamrissi Amine: Corresponding author writing the paper and operating surgeon
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Charkaoui Malak: writing the paper
Mourabbih MARIAM: writing the paper
Jalal Mohamed: study concept
Bouhya Said: correction of the paper and operating surgeon.

Declaration of competing interest

The authors declare having no conflicts of interest for this article.

References

[1] https://www.em-consulte.com/article/201836/syndrome-acro-calleux-sans-retard-mental-a-propos-.
[2] R.A. Agha, T. Franchi, C. Sohrabi, G. Mathew, For the SCARE group the SCARE 2020 guideline: updating consensus surgical CAse REport (SCARE) guidelines, Int. J. Surg. 84 (2020) 226–230.
[3] E. Bonatz, M. Descartes, J.R. Tamarapalli, Acrocallosal syndrome: a case report, J. Hand. Surg. [Am.] 22 (1997) 492–494.
[4] R.A. Pfeiffer, G. Legat, U. Trautmann, Acrocallosal syndrome in a child with de novo inverted tandem duplication of 12p11.2-p13.3, Ann. Genet. 35 (1992) 41–46.
[5] S. Bijarnia, A. Baijal, I.C. Verma, Genetic counseling in acrocallosal syndrome, Indian J. Pediatr. 70 (2003) 169–171.
[6] https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Expert=36&lng=FR.
[7] W. Courtens, E. Vamos, C. Christophe, et al., Acrocallosal syndrome in an algerian boy born to consanguineous parents: review of the literature and further delineation of the syndrome, Am. J. Med. Genet. 69 (1997) 17–22.