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. Courtens has established 4 diagnostic criteria to affirm the acrocallous syndrome and to eliminate the amalgam with other polymalformative syndrome.

1. Introduction and importance

Acrocallous syndrome is a rare polymalformative 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. Extrac

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 Courten 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.
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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CRediT authorship contribution statement

Lamrissi Amine: Corresponding author writing the paper and operating surgeon
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Jalal Mohamed: study concept
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Declaration of competing interest

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

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