A case of Pierre Robin sequence

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Objective

The Pierre Robin sequence (PRS) is a triad of retrognathia or micrognathia, glossoptosis, and U-shaped cleft palate. Its prevalence is 1: 8500 newborns. It represents an antenatal diagnostic challenge, which becomes a therapeutic challenge at birth due to the multiplicity of presentations when associating with other complex syndromes. We report the clinical case of a Pierre Robin sequence in a 20-week pregnant women.

Methods

We present a case of a 29-year-old primigravid at her 20th week of gestation who comes for the 20 week screening pregnancy ultrasound and showed retrognathia in the facial profile and a central cleft without any other malformation. After this finding, the patient was asked to carry out a karyotype performed through the analysis of amniotic fluid because we were not able to perform the fetal DNA test, and the result was normal (46 XX).

Results

A new ultrasound was performed at 29 weeks where a polyhydramnios was observed, so TORCH test and oral glucose tolerance test (OGTT) were performed, both being negative. Given the suspicion of a sequence of Pierre Robin and polyhydramnios the patient is bring up to the reference center for Fetal Medicine, where the probable diagnosis is confirmed. At 33 weeks, the patient begins with uterine dynamics and it was decided to initiate VO tocolytic therapy. At 34 weeks, the patient presented premature rupture of membranes without uterine contractions, having a eutocic delivery where a live female baby was born of 2100 grams, APGAR 6/7, who presented / displays retrognatia, glossoptosis and palatal cleft that required orotracheal intubation. Currently, the newborn is being followed up by the maxillofacial surgery unit.

Conclusion

Given the finding of retrognathia and cleft palate we should suspect a Pierre Robin sequence. In these cases, the pregnant woman must have a follow-up in a center that has an Neonatal Intensive Care Units because it is possible these newborns may need orotracheal intubation at birth and possible complications related to facial malformations may occur.