

A newborn internal and external malformations

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DESCRIPTION

We presented a case of a female newborn (NB), the fifth daughter of healthy non-consanguineous parents. It was an uneventful biamniotic bicorionic pregnancy except for a prenatal diagnosis of a right clubfoot of the first twin at the 16th week of gestation. A caesarean section was performed at 36-week gestation: the first twin was found to be involved by amniotic adhesions and the skin overlying the right knee was attached to the uterine wall. She also presented an amniotic band constricting the second and third fingers, a malformation of the right lower limb (shortness of the limb and a pterygium between foot dorsum and leg) and a caudal appendage (figures 1 and 2). She was admitted to the neonatal intensive care unit for surveillance.

A cranial and spinal MRI was performed and confirmed a *spina bifida occulta* (figure 3). Further investigation revealed no abdominal, renal or cardiac malformation. Chromosomal analysis was normal. The second twin showed no malformations.

Amniotic band syndrome has an incidence of 1:1200–1500 NB and can result in various body malformations due to constriction bands with interference in fetal development.¹ *Spinal bifida occulta* associated to caudal appendage is due to a failure in the secondary neurulation or segmentation process, which occurs in the first weeks of gestation.²

Presently, at 3 years of age she is unable to walk due to the right limb malformation and has



Figure 1 Caudal appendage.



Figure 2 Malformation of the right lower limb (shortness of the limb and a pterygium between foot dorsum and leg) and adhesion of the skin overlying the right knee to the uterine wall (already sectioned).



Figure 3 Spinal MRI confirming the presence of spina bifida occulta (S2–S3).

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diminished strength in lower limbs as well as no sphincter control (urethral and rectal).

Learning points

- ▶ Association between human tail and limb defects and amniotic bands have been described in the literature.³
- ▶ Although amniotic bands do not explain the caudal appendage, recurrence of this association suggests a possible common aetiology.
- ▶ Experienced prenatal diagnosis could have contributed to counselling and therapeutic orientation.

Contributors MIM and MF were responsible for the newborn while she was admitted in NICU. They were supervised by RB and JM did the follow-up after discharged.

Competing interests None.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

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