Constriction band syndrome in a healthy full-term newborn

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Congenital limb deficiency disorders (CLDDs) comprise a collection of anomalies that are characterized by skeletal aplasia or hypoplasia. The incidence of CLDD is approximately 1 in 1700 live births[1] and the pathogenesis varies from genetic and vascular causes, to environmental exposures, to iatrogenic causes. As CLDDs have such a broad range of origins, family physicians should have an approach to their diagnosis and subsequent management that optimizes patient care.

The case presented below describes a baby who was born at a low-risk obstetrics hospital staffed by family physicians. After delivery, a velamentous cord insertion was noted and the subsequent newborn examination findings revealed a CLDD in the left upper extremity.

We conducted a comprehensive literature search spanning January 1970 to July 2016 in PubMed and Google Scholar using the MeSH terms (*velamentous* or *marginal*) *cord insertion* and (*constriction* or *amniotic*) *band syndrome*. This search did not produce any studies associating velamentous cord insertions with constriction band syndrome (CBS), which prompted us to put forth this case study.

Case

The patient is an infant born to a 39-year-old woman (gravida 3, para 2, aborta 1). Prenatally, the mother was followed by the family physician obstetric team and was assessed in the second trimester by the maternal-fetal medicine team owing to advanced maternal age. A detailed anatomic ultrasound at 20 weeks noted an anterior, non–low-lying placenta with a centrally inserted, 3-vessel umbilical cord. The pregnancy progressed uneventfully and the mother presented at 39 weeks and 4 days' gestation in well-established labour. Membranes ruptured spontaneously for clear amniotic fluid. The baby was in an occiput posterior position and had a spontaneous vaginal delivery. There were no abnormalities on fetal monitoring throughout labour and delivery. The baby's Apgar scores were 8 at 1 minute and 9 at 5 minutes, with a birth weight of 3549 g. The placenta was delivered spontaneously 12 minutes after delivery. There was a velamentous, eccentric cord insertion 2.5 cm from the closest edge of the placental disk (**Figure 1**). The initial newborn examination findings revealed an intrauterine amputation of the intermediate and distal phalanges of digits 2 to 4 of the left hand, with a thin incurved left fifth finger (**Figure 2**). In addition, syndactyly of the second and third toes of the left foot without any sign of hemihypertrophy was identified. No other abnormalities were noted in the newborn examination findings but the newborn was referred to a pediatric plastic surgeon for assessment to optimize function of the left hand.

Discussion

As we have already highlighted, CLDDs can arise through many different causes, including vascular causes, intrauterine amputations, and teratogen exposure, while still other suspected genetic CLDDs have yet to be elucidated.[2] What many congenital limb deficiencies do have in common is that the mechanism of injury tends to occur during weeks 4 to 8 of gestation, which is a critical period of limb morphogenesis.

The international standard nomenclature for congenital limb deficiencies can help guide the initial assessment of CLDD by dividing limb

Editor's key points

- Congenital limb deficiency disorders (CLDDs) are a broad group of conditions that are characterized by skeletal aplasia or hypoplasia. They can arise through vascular causes, intrauterine amputations, teratogen exposure, and suspected genetic factors.
- Assessing and treating any newborn with a CLDD requires a thorough physical examination looking for other anomalies, a detailed history of the mother's pregnancy, genetic counseling, and potentially surgical assessment for optimization of limb function.
- When a transverse CLDD is identified, the placenta should be examined for pathology, as most cases of this type of limb deficiency are caused by amniotic banding within the amniotic sac.

Points de repère du rédacteur

- La maladie des brides amniotiques touchant les membres (MBAM) regroupe un vaste ensemble d'anomalies congénitales caractérisées par une aplasie ou une hypoplasie squelettique. Ces anomalies peuvent être attribuables à des causes vasculaires, à des amputations intra-utérines, à une exposition tératogène ou à des facteurs génétiques soupçonnés.
- L'évaluation et le traitement d'un nouveau-né atteint d'une MBAM exigent un examen physique rigoureux pour dépister d'autres anomalies, une anamnèse détaillée de la grossesse de la mère, un counseling génétique et, potentiellement, une évaluation en chirurgie pour optimiser le fonctionnement du membre.
- Lorsqu'une MBAM transversale est constatée, il faut un examen pathologique du placenta, parce que la plupart des cas de ce genre d'anomalies aux membres sont causés par la formation de bandes amniotiques dans l'amnios.

Figure 1. Velamentous, eccentric cord insertion 2.5 cm from the closest edge of the placental disk

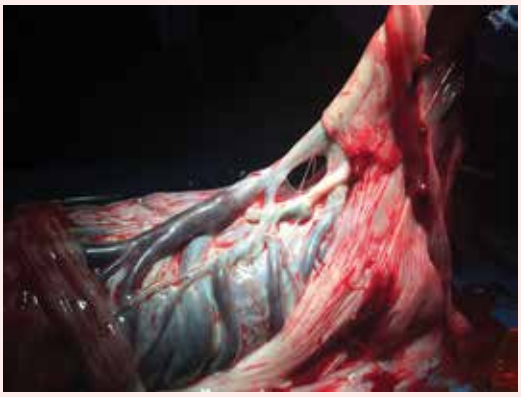
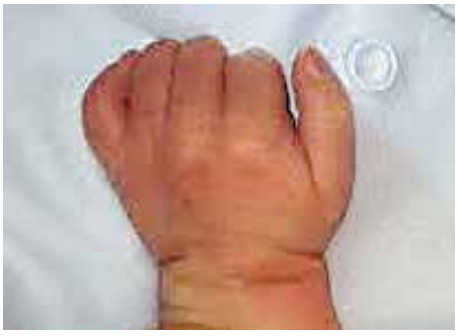


Figure 2. Intrauterine amputation of the intermediate and distal phalanges of digits 2 to 4 of the left hand, with a thin incurved left fifth finger



deficiencies into 2 types: longitudinal and transverse.[3] In longitudinal deficiencies, there is a reduction or absence within the long axis of the limb and these deficiencies are often part of a more complex syndrome. Transverse limb deficiencies are defined as having a developmentally normal limb until a specific transverse plane is reached, with absent or dysmorphic limb features distal to that plane.[3] They are often unilateral and are less likely to be part of a syndrome. However, care must be taken on the physical examination, as a genetic condition known as Adams-Oliver syndrome also presents with asymmetric transverse limb deficiencies in addition to other characteristic features such as aplasia cutis congenita of the scalp, cardiovascular malformations, and brain anomalies.[4] More commonly, transverse limb deficiencies result from amniotic banding within the amniotic sac, which occurs when the amnion layer of the placenta ruptures and separates from the chorion, producing fibrous strands that entangle and compromise blood flow in affected extremities.[2]

When a congenital limb deficiency is identified, a thorough physical examination should be performed to assess for additional anomalies that could be part of a broader syndrome. A detailed history of the pregnancy should also be obtained, focusing on potential teratogen exposures and whether chorionic villus sampling occurred, as this can be associated with transverse limb deficiencies.[5] In addition, a pedigree should be obtained including history of pregnancy loss, other family members with congenital anomalies, and consanguinity.[2] In terms of management, all newborns with a CLDD and their family members should be offered genetic counseling. In addition, the newborn might benefit from assessment by an orthopedic or plastic surgeon to optimize function of the affected limb, but this can vary based on clinical presentation.

As many CLDDs occur in the first trimester, the greatest challenge for prenatal diagnosis and treatment is timely detection. However, potential curative treatments are currently evolving with the use of in utero fetoscopic surgery. For instance, a review by Javadian et al identified 14 cases where amniotic bands were relieved by in utero fetoscopic surgery, with preserved limb function in 50% of the cases.[6] While promising, this study highlighted the need for earlier CBS diagnosis, which can only come by better understanding the basic pathophysiologic processes of CBS.[6]

Conclusion

Congenital limb deficiency disorders are a collection of disorders that have a range of causes, the most common being transverse limb deformities caused by amniotic banding within the amniotic sac. When a congenital limb deficiency is identified, the family physician should perform a thorough physical examination looking for other anomalies and obtain a detailed pregnancy history that includes potential teratogenic exposures and a multigenerational pedigree. Management of CLDDs will vary depending on the type but can include genetic counseling and surgical referral for optimization of limb function.

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Competing interests
None declared

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