

CASE REPORT

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Sacral cutaneous appendage associated with lipoma of the filum terminale and anchored medulla: a case of prenatal diagnosis

Apéndice cutáneo sacro asociado a lipoma del filum terminal y médula anclada: un caso de diagnóstico prenatal

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ABSTRACT

Closed spinal dysraphisms have a still unknown prevalence and involve a wide variety of forms. Lipoma of the filum terminale is considered within spinal lipomas and is usually associated with tethered medulla. Lumbosacral cutaneous stigmata are not always indicative of closed spinal dysraphism. We report a case of prenatal diagnosis of sacral cutaneous appendage with suspected tethered cord, confirmed at birth as lipoma of the filum terminale with tethered cord.

Key words: Tail, human, Skin appendage, Cauda equina syndrome, Spina bífida, Filum terminale, lipoma, Neural tube defects

RESUMEN

Los disrafismos espinales cerrados tienen una prevalencia aún desconocida e involucran una gran variedad de formas. El lipoma del filum terminal es considerado dentro de los lipomas espinales y suele asociarse a médula anclada. Los estigmas cutáneos lumbosacros no siempre son indicadores de disrafismo espinal cerrado. Reportamos un caso de diagnóstico prenatal de apéndice cutáneo sacro con sospecha de médula anclada, confirmado al nacer como lipoma del filum terminal con médula anclada.

Palabras clave. Apéndice cutáneo, Cauda equina, Síndrome de cauda equina, Espina bífida, Filamento terminal, lipoma, Defectos del tubo neural

INTRODUCTION

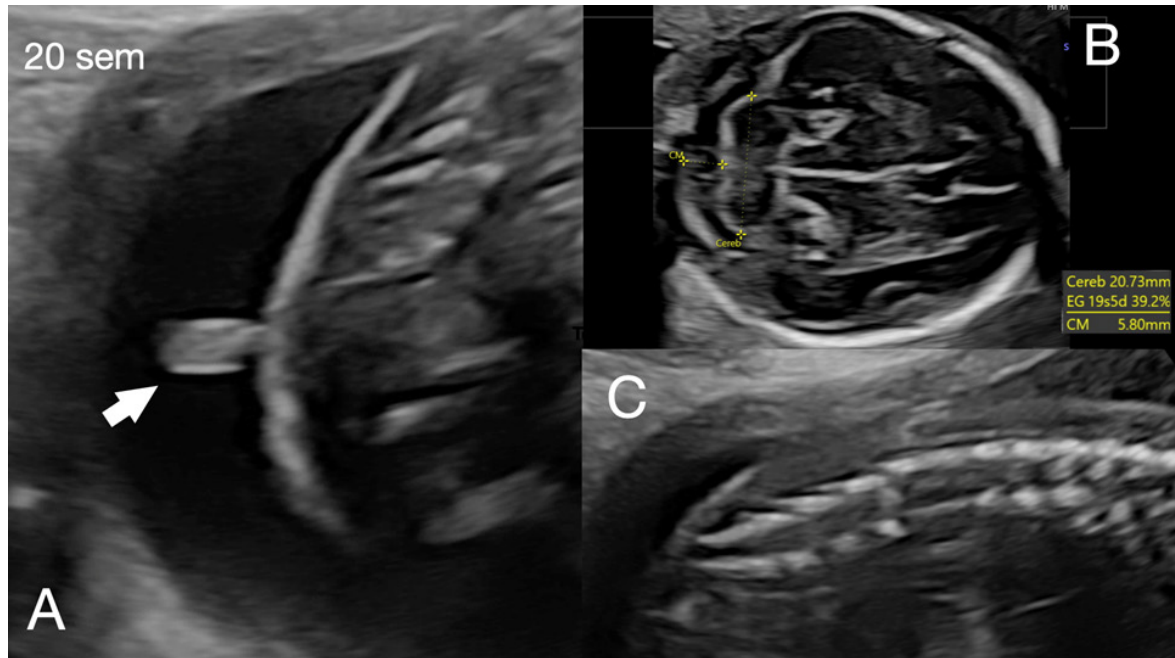
Closed spinal dysraphisms (CSD) have a still unknown prevalence and involve a great variety of forms. The most widely accepted clinicoradiological classification groups them according to the presence or absence of subcutaneous mass⁽¹⁾. The embryological origin of CSD is in the closure of the distal neural tube or secondary neurulation, and this also explains its association with the various forms of cutaneous stigmata⁽²⁾. The importance of the diagnosis of CSD lies in the early postnatal search for spinal cord alterations, intraspinal tumors or tethered cord that affect the prognosis of life. Prenatal suspicion is based on looking for skin-covered lumbosacral tumors, cutaneous stigmata or alterations in the shape or position of the fetal conus medullaris⁽³⁾. In the present report we will discuss a case of prenatal diagnosis of sacral cutaneous appendage (human tail) with suspected tethered medulla, in which lipoma of the filum terminale (FTL) with tethered medulla was postnatally confirmed.

CASE REPORT

During the second trimester ultrasound of a 20 weeks of gestation pregnant woman, an elongated cutaneous appendage was found at the midline of the fetal sacrum with no evidence of connection between the tumor and the spinal canal (Figure 1). In the evaluation of the fetal



FIGURE 1. A: CUTANEOUS APPENDIX (TAIL) IS VISUALIZED AT SACRAL LEVEL IN A 20-WEEK GESTATION FETUS. B: TRANSVERSE SECTION, SHOWING NORMAL POSTERIOR FOSSA. C: SAGITTAL VIEW OF THE SPINE AT THE LEVEL OF THE SACRUM SHOWING BONY INTEGRITY.



head and brain no alterations were found, the spine was normal in all its segments (Figure 1). In the 24th week of gestation a conus medullaris at the level of the body of the fourth lumbar vertebra (L4) and an elongated and pedunculated cutaneous appendix with characteristics similar to fatty tissue covered with skin near S5 were visualized (figure 2). In the following controls the conus medullaris was observed up to L3 in the 28th week, with suspicion of closed spinal dysraphism and anchored cord. In the newborn

(NB) an elongated cutaneous appendage (tail) in the midline of the sacral region with a thin base and globular towards the end and intergluteal groove with deformation and asymmetry in its upper portion was found (Figure 3). No other malformations were detected in the newborn. Magnetic resonance imaging located the conus medullaris at the level of the inferior plate of L3. In the filum terminale a hyperintense area was observed in the T1 sequence, 1.5 mm thick by 14 mm long, which corresponded to FTL. At the

FIGURE 2. A: SAGITTAL SECTION OF THE SPINE AT THE LEVEL OF THE SACRUM SHOWING THE CONUS MEDULLARIS AND ITS POSITION WITH RESPECT TO THE VERTEBRAL BODIES. B: CUTANEOUS APPENDAGE (TAIL) VISIBLE AT THE LEVEL OF THE LAST SACRAL VERTEBRAE.

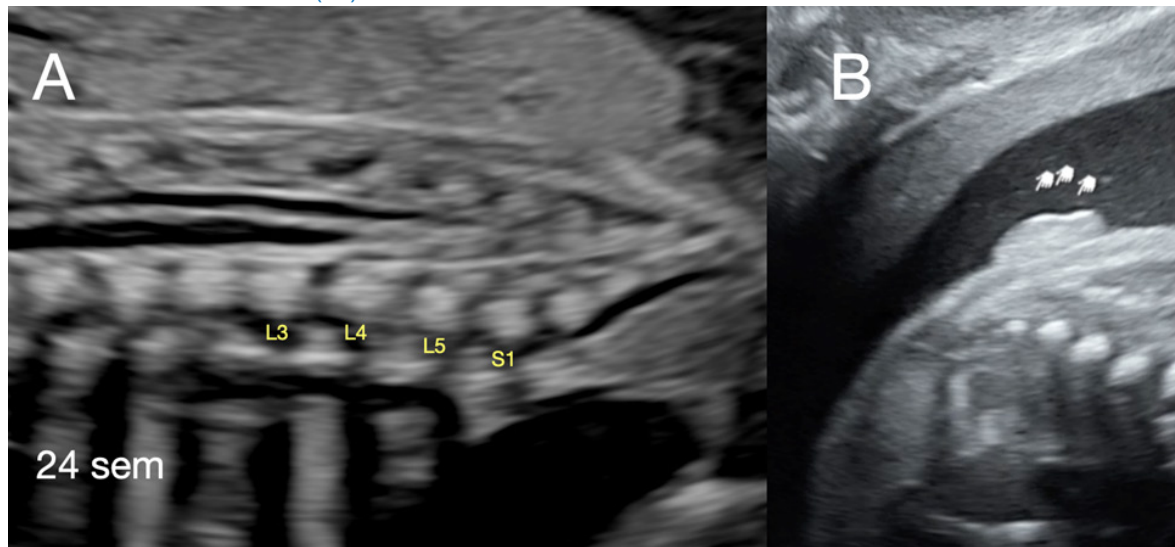
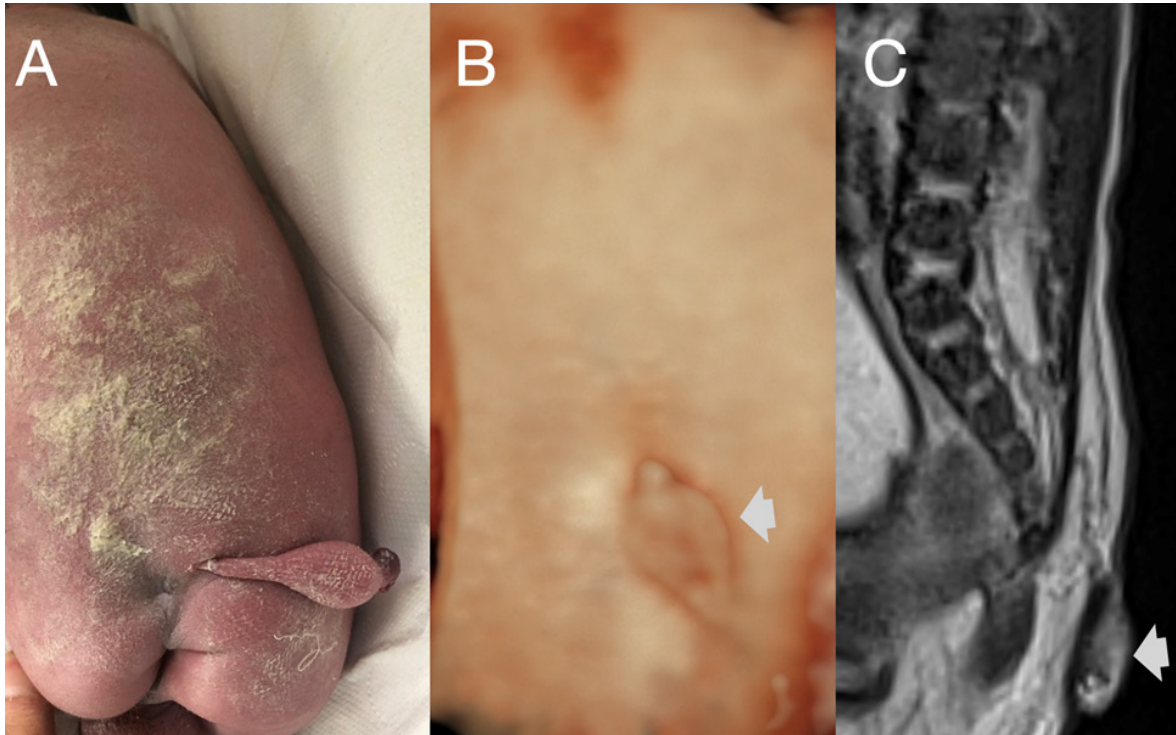




FIGURE 3. A: IMAGE OF THE CUTANEOUS APPENDAGE AND THE ASYMMETRY OF THE INTERGLUTEAL LINE IN THE NEWBORN. B: 3D RECONSTRUCTION IMAGE OF THE CUTANEOUS APPENDAGE IN THE FETUS. C: CUTANEOUS APPENDAGE VISIBLE ON MRI, SHOWING NO CONNECTION WITH THE MEDULLARY CANAL.



level of the sacrum, a cutaneous appendage of fatty tissue 20 mm long and 16 mm thick was seen, which did not show continuity with the medullary canal (Figure 4). The NB was evaluated by neurosurgery; the specialists considered that for the moment he did not require surgery to release the conus medullaris and proposed a follow-up in 6 months.

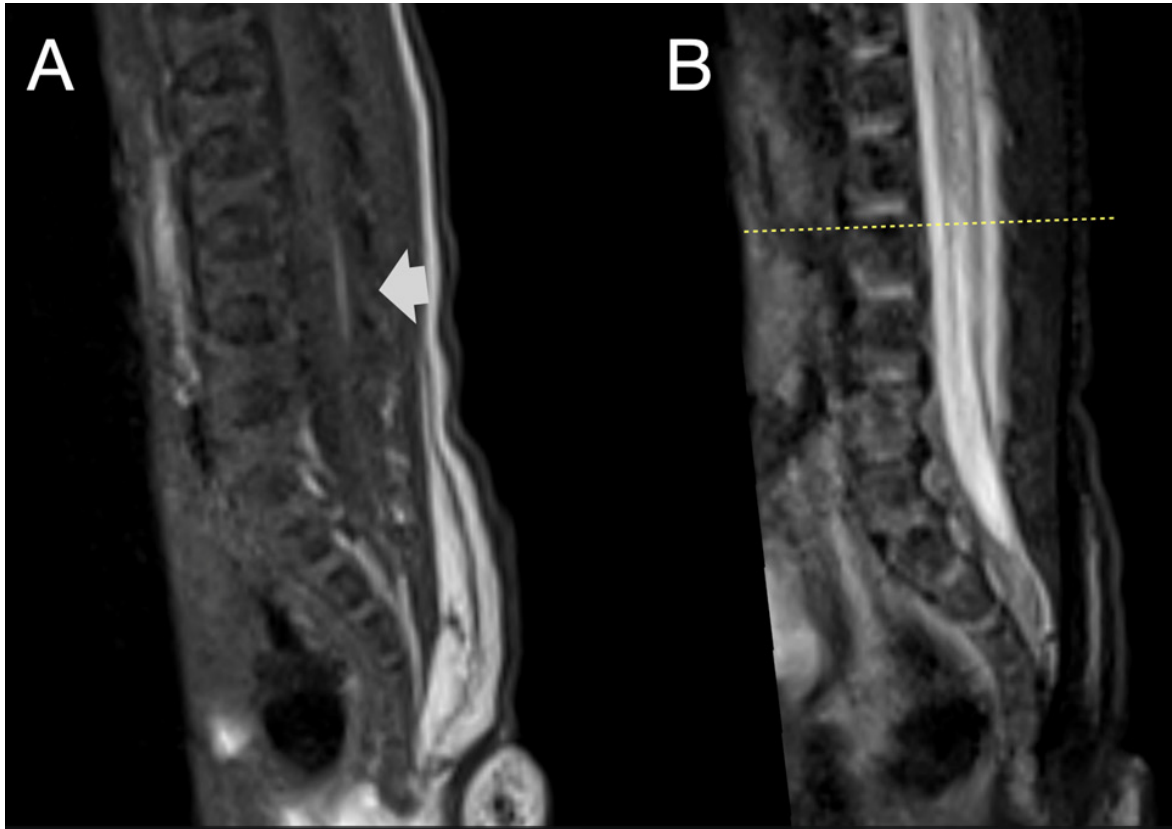
DISCUSSION

FTL is defined as the presence of fatty tissue in the filum terminale, which results in its thickening. The fatty infiltration of the filum terminale compromises its flexibility and produces the tethered medulla. This traction would generate alterations in microcirculation, electrophysiology, and nerve metabolism, which explains the neurological symptoms⁽²⁾. The prevalence of FTL is not well determined, but it is reported in some retrospective series to be found in 0.2% to 5% of the population⁽⁴⁻⁷⁾. FTL constitutes a type of CSD within spinal lipomas, such as lipomyelocele or lipomeningocele (84%), lipoma of the filum terminale (12%) and intradural lipoma (4%)^(8,9). The diagnosis of FTL is ideally made by MRI, observing at the level of the filum terminale a hyperintense area in T1 sequence that varies in its ex-

tension and thickness, as was evidenced in the case presented (figure 4)^(6,7). Spinal ultrasound of newborns can contribute to the suspicion if a thickness of the filum terminale greater than 1.6 to 2 mm or a hyperechogenic area is found⁽²⁾.

Lumbosacral cutaneous stigmata (dimple, dermal stigma, lipoma, subcutaneous mass, hypertrichosis, hemangioma, aplasia cutis or cutaneous appendage) may be present in 2-7% of newborns and their presence is not always indicative of CSD^(10,11). A systematic review found that only 3.1% of newborns with one cutaneous stigma had CSD; however, if more than one cutaneous stigma was present, the prevalence rose to 10%⁽¹⁰⁾. Usami, in a series of 174 cases of children with FTL found that 54% were associated with some cutaneous stigma, such as deviation of the intergluteal line 19%, sinus, dermal 15%, hemangioma 14%, cutaneous aplasia 5%, hypertrichosis 3% and sacral cutaneous appendage (tail) 1%. In the present case the cutaneous stigmata were two: cutaneous appendage or tail and asymmetry of the intergluteal line (Figure 3). In Oh's series of 337 newborns with sacral dimpling, LFT was found in 40 cases (12%), with none of them evidencing the presence of anchored medulla on MRI⁽¹²⁾.

FIGURE 4. A: MRI OF THE NEWBORN IN T1 SEQUENCE SHOWING AN ELONGATED HYPERINTENSE AREA AT THE FILUM TERMINALE CORRESPONDING TO LIPOMA (ARROW). B: POSITION OF THE CONUS MEDULLARIS IN RELATION TO THE L3 VERTEBRA.



The human tail or sacrocutaneous appendix can be classified by the presence (true tail) or absence (pseudotail) of bony or cartilaginous vertebral tissue, as proposed by Dao and Netsky in 1984⁽¹³⁾. The case presented would be classified as a pseudotail, being fatty tissue and skin (Figures 2 and 3). Tojima, in a new classification, also considers the terminology of true tail and pseudotail, but prioritizes the location, with the intention of correlating it with the prognosis. In 195 published postnatal cases he finds that the association with CSD (44 of 92 cases) was mainly with the presence of pseudocola in the midline of the sacrum⁽¹⁴⁾. Published cases of prenatal diagnosis of sacral appendage are scarce, single reports associated with other malformations. Dobson⁽¹⁵⁾ published the largest series of fetal diagnosis, with 7 cases found in 10 years, 6 with multiple malformations that ended in fetal or perinatal death and only one case reached term with low conus medullaris (L3 vertebra). He also reported that the presence of the sacral appendix may no longer be visible in subsequent controls, especially those of small size. We publish one of the few cases of sacral appendix associated with FTL with prenatal diagnosis.

The diagnosis of tethered cord is related to the level of the conus medullaris. In newborns the position is usually at L2, with ranges from the lower plate of L1 to the lower plate L2, depending on the series being reviewed^(9,16). For fetuses, the position of the conus medullaris is visualized in a gradual ascent from L4 at week 16, L3 at week 24 to the similar position of newborns in the third trimester^(17,18). Our case showed ascending conus medullaris, but in lower positions than usual, so CSD was suspected (Figure 2). Many methods have been developed to assess the position of the fetal conus medullaris, such as conus position, distances from the conus to the end of the sacrum or distance to the end of the intradural space⁽¹⁷⁻¹⁹⁾.

FTL is a mostly asymptomatic condition, as can be seen in several published series in children and adults⁽⁴⁻⁶⁾ and is not necessarily associated with tethered cord. Symptomatic cases are associated with urological problems (sphincter dysfunction, dysuria, or repeated urinary tract infection), low back pain and neuro-orthopedic symptoms of the lower limbs⁽¹⁻³⁾. Symptomatic cases of FTL are mainly related to the low posi-



tion of the conus medullaris as the main associated factor⁽⁵⁾. The need for FTL surgery is related to the associated symptomatology; the low level of the conus medullaris without symptoms is a point under discussion to be considered as a surgical indication since there are contradictory data^(4,5). Likewise, it is under discussion whether prophylactic FLT surgery should be offered for asymptomatic cases with conus medullaris in normal position. In general, the results of surgery are good for the resolution or stabilization of symptoms^(4,5).

In conclusion, the association of cutaneous stigmata and closed spinal dysraphism warrants an exhaustive search in the newborn. Prenatal screening for spinal cord abnormalities, cutaneous stigmata and sacral spine abnormalities should be routinely used, given the life-threatening impact of these malformations.

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