

# A True Human Tail in a Neonate

## Case report and literature review

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### ذيل بشري حقيقي في طفل حديث الولادة تسجيل حالة واستعراض للأدبيات العلمية

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**ABSTRACT:** A true human tail is a benign vestigial caudal cutaneous structure composed of adipose, connective tissue, muscle, vessels, nerves and mechanoreceptors. A true human tail can be distinguished from a pseudotail as the latter is commonly associated with underlying spinal dysraphism, which requires specialised management. True human tails are very rare, with fewer than 40 cases reported to date. We report a healthy one-day-old male newborn who was referred to the Bharath Hospital, Kottayam, Kerala, India, in 2014 with a cutaneous appendage arising from the lumbosacral region. Magnetic resonance imaging of the spine ruled out spinal dysraphism. The appendage was removed by simple surgical excision. Clinicians should emphasise use of 'true tail' and 'pseudotail' as specific disparate terms as the clinical, radiological and histological findings of these conditions differ significantly, along with management strategies and outcomes.

**Keywords:** Tail; Newborn; Magnetic Resonance Imaging; Spinal Dysraphism; Case Report; India.

**المخلص:** يعد الذيل البشري الحقيقي بنية أثارية زنبية جلدية حميدة، تتكون من أنسجة ضامة وأنسجة دهنية، وعسل وأوعية دموية وأعصاب ومستقبلات ميكانيكية. ويمكن التفريق بين الذيل البشري الحقيقي وغير الحقيقي بأن الذيل غير الحقيقي يرتبط عادة بوجود خلل الرفاء الشوكي المستبطن، الذي يتطلب معالجة متخصصة. ويعد وجود ذنب بشري حقيقي أمراً نادراً، حيث أن الحالات المسجلة عن وجوده إلى الآن أقل من 40 حالة. ونسجل هنا حالة طفل صحيح حديث الولادة تم تحويله إلى مستشفى باهرات بكوتيام في كلبيرا بالهند في عام 2014م بسبب وجود زائدة جلدية ناشئة من المنطقة القطنية العجزية. وتم استبعاد وجود خلل الرفاء الشوكي عن طريق تصوير العمود الفقري بالرنين المغناطيسي. وتمت إزالة تلك الزائدة باستئصال جراحي بسيط. ينبغي على الممارسين التفريق بين "الذيل الحقيقي" و"الذيل غير الحقيقي"، واعتبارهما مصطلحين مختلفين، وحالتين منفصلتين، إذ أن لهما خواصا سريرية وأشاعية وهيستولوجية مختلفة، ويتطلبان استراتيجيات علاجية مختلفة، ويفضيان إلى نتائج متباينة.

**الكلمات المفتاحية:** ذيل؛ حديث الولادة؛ التصوير بالرنين المغناطيسي؛ خلل الرفاء الشوكي؛ تقرير حالة؛ الهند.

**A** HUMAN TAIL IS A BENIGN CONGENITAL anomaly resulting in a vestigial lumbosacral dorsal cutaneous appendage which can be categorised as either a 'true tail' or a 'pseudotail' by clinical, radiological and histopathological examination.<sup>1,2</sup> Unlike a true tail, a pseudotail indicates possible underlying spinal dysraphism and accurate distinction between these two entities is vital as both management and outcomes can vary.<sup>1-3</sup> True human tails are very rare, with fewer than 40 cases reported to date.<sup>4</sup> This case report describes an otherwise healthy neonate who was born with a true human tail.

## Case Report

A one-day-old male neonate was referred to the Bharath Hospital, Kottayam, Kerala, India, in 2014 with a cutaneous appendage arising from the lumbosacral region which had been present at birth. He was the second child born to non-consanguineous

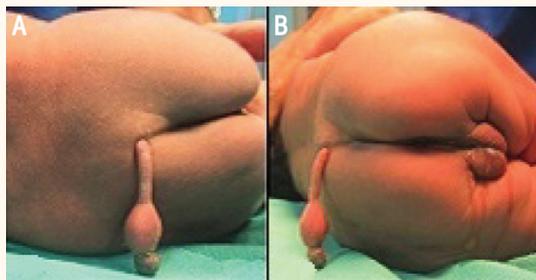
parents and his sibling was clinically normal. The neonate had been born by vaginal delivery and the mother had had no history of illness, radiation exposure or drug intake other than vitamin supplementation during the antenatal period. In addition, there was no history of congenital anomalies in any of the family members. Apart from the appendage, the male newborn was otherwise healthy.

On examination, the neonate was observed to have a 6.5 cm-long club-shaped soft tissue appendage arising and hanging down from the mid-sacral region, which was completely covered by skin and had no spontaneous movement [Figure 1]. The mass of the appendage was fleshy but firm and was not translucent. During palpation of the mass, the neonate cried when the appendage was pressed and cried even more loudly when it was pricked with a pin. The anal muscle tone was normal and the neonate had active movement in both lower limbs. A detailed examination revealed no other associated anomalies.

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**Figure 1:** Clinical photographs of a male newborn showing a cutaneous appendage arising from the sacral region from the (A) frontal and (B) side views.

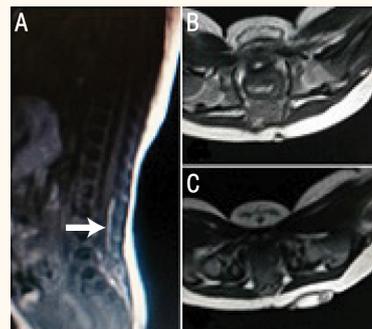
Magnetic resonance imaging (MRI) of the spine showed no evidence of spinal dysraphism, confirming the diagnosis of a true human tail [Figures 2 and 3]. The tail was excised by making an elliptical incision at the base. The wound was examined to rule out any connections between the tail and deeper structures which might not have been evident on the MRI scans. Microscopy of the excised tail showed that the tissue was lined by a squamous epithelium containing adipose tissue, muscle bundles, blood vessels and nerve bundles. The postoperative period was uneventful. At a one-month follow-up, the wound had healed well and a neurological examination was normal.

## Discussion

In developing human embryos, a tail bud starts to develop during the fourth gestational week and grows unevenly for the next two weeks, extending and curling behind the hindgut.<sup>2,5-9</sup> The proximal portion of the tail bud has 10–12 vertebrae, whereas the distal portion is composed of mesodermal elements without vertebrae. During the seventh and eighth gestational weeks, the proximal mesodermal elements retract into the soft tissue and the distal mesodermal



**Figure 2:** A–C: Sagittal T2-weighted magnetic resonance imaging (MRI) scans of a male newborn showing a pedunculated appendage (arrows) with fat intensity at the sacral region. D: Sagittal T2-weighted MRI scan showing the *conus* located at the level of the third lumbar vertebra (arrow).



**Figure 3:** A: Sagittal T1-weighted magnetic resonance imaging (MRI) scan of a male newborn showing the thickened *filum terminale* with fat intensity (arrow). Axial (B) T1- and (C) T2-weighted MRI scans showing the origin of the appendage in the midline at the sacral level.

elements project outwards temporarily, before disappearing completely via *apoptosis* by the end of the eighth gestational week.<sup>2,5-9</sup> According to Lu *et al.*, the presence of a human tail is an abnormality in embryonic development, rather than a regression in the evolutionary process.<sup>2</sup> In mice and other vertebrates, the genes that control the development of tails are the *Wnt family member 3A (WNT3A)* and *caudal type homeobox 1* genes; the downregulation of the *WNT3A* gene has been observed to induce *apoptosis* of the tail cells in mice.<sup>10-13</sup> Therefore, it is possible that mutations resulting in increased upregulation of the *WNT3A* gene may result in retention of the embryonic tail in human newborns. However, further genetic research on this topic is necessary.

The presence of cutaneous midline congenital lesions in the lumbosacral region—such as vascular *naevi*, tufts of hair, dermal sinuses, subcutaneous lipomas, deviated gluteal furrows or a human tail—may indicate the presence of occult spinal dysraphism.<sup>1</sup> A congenital cutaneous appendix arising from the lumbosacral region is referred to as a human tail.<sup>14</sup> On histopathological examination, human tails can be classified into either true tails or pseudotails.<sup>1,15</sup> A true human tail is a distal skin-covered boneless midline protrusion which is a remnant of the embryonic tail, composed of a core of striated muscle, adipose and connective tissue and containing blood vessels, nerve fibres, ganglion cells and mechanoreceptors, such as Pacinian corpuscles.<sup>4,15,16</sup> True tails are commonly attached to the skin of the lumbosacral region; the skin which completely envelopes the mass is deficient in hair follicles and sweat and sebaceous glands.<sup>4,15,17</sup> However, there is one case in the literature of a neonate born with a true tail which contained bone.<sup>18</sup> True human tails are sometimes capable of spontaneous or reflexive motion, while voluntary contract-

ions are rare.<sup>15,19</sup> True human tails are not inherited; however, a single report has been published in which three female generations within one family were born with true human tails.<sup>20</sup> The incidence rate of true human tails is twice as high among males in comparison to females.<sup>15</sup>

Unlike true tails, a pseudotail is a caudal protrusion composed of other normal and abnormal tissue such as bone, cartilage and remnants of notochord.<sup>3,15</sup> During neurulation, non-fusion of the neural tube due to focal premature dysfunction can lead to the formation of a pseudotail.<sup>1</sup> The non-fused neural tube exposes the paraxial mesoderm to the dorsal aspect of the neural ectoderm and induces the formation of fatty elements. This prevents neural tube fusion and the attachment of these fatty elements to the neural structures, which leads to a tethering cord and tethered cord syndrome.<sup>2</sup> Early untethering of the cord is essential to prevent the development of catastrophic neurological *sequelae*.<sup>9,14,21</sup> The incidence of cutaneous lesions along the craniospinal axis in the general population is approximately 3% and the most common lesions are deep dimples.<sup>22</sup> However, cutaneous signs are present in 76% of patients with occult spinal dysraphism, including tails, dermal sinuses, lumbar lipomas, port wine stains, deviated gluteal furrows, hypertrichosis, haemangiomas and hamartomas.<sup>1</sup> Lu *et al.* reviewed 59 patients with human tails and noted that 49% of cases had spinal dysraphism and 20% had tethered cords.<sup>2</sup>

## Conclusion

The current case describes a neonate who was found to have a true human tail as evidenced by the absence of bony elements or underlying spinal dysraphism. It is very important that clinicians differentiate true tails from pseudotails as the latter entity is associated with underlying spinal lesions. Accordingly, MRI of the spine is crucial as it outlines the underlying pathology and helps in surgical planning. A true human tail is a benign condition and can be treated via simple surgical excision.

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