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Case Report

A girl with spina bifida, an extra leg, and ectopic intestinal loops – a "foetus in foetu" or a whim of the neural crest?

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Summary

This article describes a girl with an extra leg attached to her lower back, combined with a spina bifida and a myelomeningocele. Despite lacking sensory or motor functions, the leg grew proportionately with the rest of the body. The bony structures were almost normal. A cross section showed fat tissue with some centrally situated blood vessels, nerve bundles, and muscular fragments. Proximally, an isolated colon loop was found. The extra leg and intestine respected the dorsal fascia, without connection with the peritoneal or retroperitoneal compartments. The finding is discussed with reference to existing hypotheses for limb formation.

Keywords: Foetus in foetu; maldevelopment; neural crest; parasitic twin; teratoma.

Introduction

"Foetus in foetu", also called "parasitic twin", is a well-known but rare condition that occurs at 3 weeks of gestation [18]. Grant and Pearn indicate an incidence of 1 per 500 000 births [15]. It was first described by Banks [3] and Meckel [26] and defined further by Willis [33]. A PubMed search for the term revealed 118 publications dating back to 1950. Several recent articles also offer reviews of previously published cases [2, 7, 16, 24, 27, 32]. A majority of the described cases have appeared in relation to the peritoneum, most often retroperitoneally [16].

During our work in Addis Ababa, Ethiopia, we encountered a patient with an uncommon constellation: spina bifida, a myelomeningocele, an extra leg attached to the back and a stray loop of intestines. Owing to lack of expertise and financial resources, the girl had reached the age of 1 year and 2 months before the referral process started. A year later, she was finally referred to us for surgery. Details of the case are presented below.

Case report

This 2-year-old girl was born with an extra leg attached to her back, in the lower thoracic-lumbar region (Fig. 1). She also had pronounced bilateral, distal pareses of the two otherwise normal lower limbs, most pronounced in the right leg, as can be seen in patients with a myelomeningocele. Her psychomotor development had otherwise been normal, without any signs of hydrocephalus. The extra leg was of approximately the same size as the two ordinary legs and was covered by normal skin. The leg grew proportionately with the lower limbs and the rest of the body from the time of birth to the time

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Fig. 1. Preoperative picture of the patient at the age of 2 years. Note the extra leg held by the investigator, and the distal pareses of the lower limbs

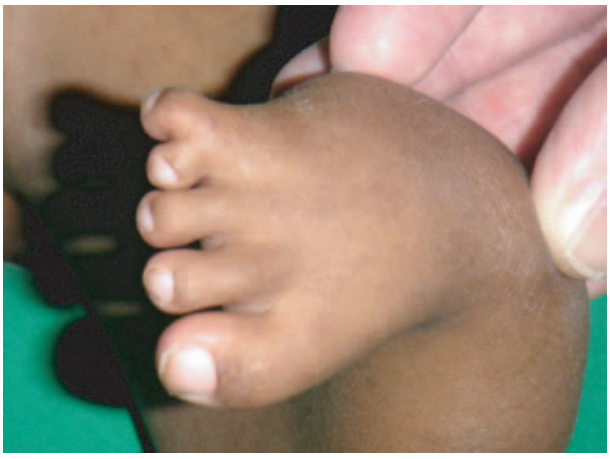

Fig. 2. Details of the foot of the extra leg

of operation at the age of 2 years and 5 months. An X-ray revealed a thoraco-lumbar spina bifida as well as the bony elements of the extra leg, which appeared to have an almost normal appearance.

The pregnancy was uneventful, except that her mother tried to provoke an abortion during the first month of the pregnancy by taking a total of 20 contraceptive pills distributed over 6 days. The exact type of contraceptives is unknown. When the mother could not feel any normal movement and the date of the term was past 2–3 weeks, she delivered by Caesarean section. The extra leg had not been diagnosed prior to birth.

As a consequence of the pareses associated with the myelomeningocele, the child's right, normal foot was in an equinus position with pareses of all the foot muscles (extensors, flexors, invertors and evertors), and the forefoot was in a varus position. In her left foot, the hindfoot was in equinus, the forefoot in varus and the midfoot in cavus position (Fig. 1). All the foot muscles of the left foot had the same pareses as in the right foot.

She had spontaneous urination and defecation, but her bowel function was somewhat slow. She only defecated once or twice a week. She had not had any urinary tract infections and her bladder seemed to function normally.

The extra leg was covered by normal skin which was indistinguishable from that covering the rest of her body (Fig. 1). The leg appeared to contain normal long bones, and the foot had four toes, the fourth toe, however, was divided distally into two toes with two separate nails (Fig. 2). In the cranial and proximal portion of the ''thigh'' of the extra leg, overlying the cranial part of the spinal defect, a soft swelling could be palpated to the left, which we anticipated to be the myelomeningocele (MRI not available). The leg could be passively moved in all directions, indicating that there was no bony attachment to the girl's skeleton. The extra limb did not seem to have any sensory connections with the girl's central nervous system, as it could be pinched or otherwise stimulated with pain stimuli without any reaction. The deep tendon reflexes or the plantar reflexes could not be elicited. Voluntary or spontaneous active movements were not observed.

She was operated on at the age of 2 years and 5 months. In order to reduce intra-operative blood loss, a tight circular bandage was wrapped around the extra limb before the operation. The separation started with a cranio-caudal, leaf-shaped incision around the base of the limb, leaving enough skin for an adequate wound closure. Underneath the skin of the stray limb, only fat tissue, a few blood vessels, the bony elements, but no musculature, could be observed. Proximally in the leg, we found an apparent iliac bone that was attached to the left edge of the lumbosacral spine with strong bands of connective tissue, however, without a real joint (Fig. 3).

A postoperative X-ray of the removed limb revealed almost normal bony elements along the entire length of

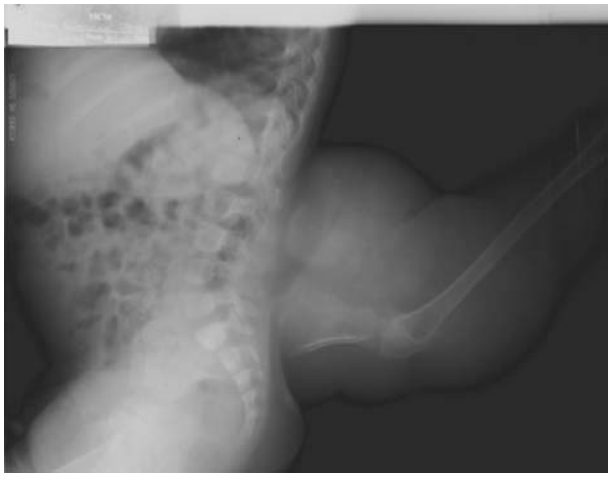

Fig. 3. Preoperative X-ray (lateral view) showing the attachment of the extra leg. Note the loose, non-bony attachment of the extra iliac bone to the patient's spine

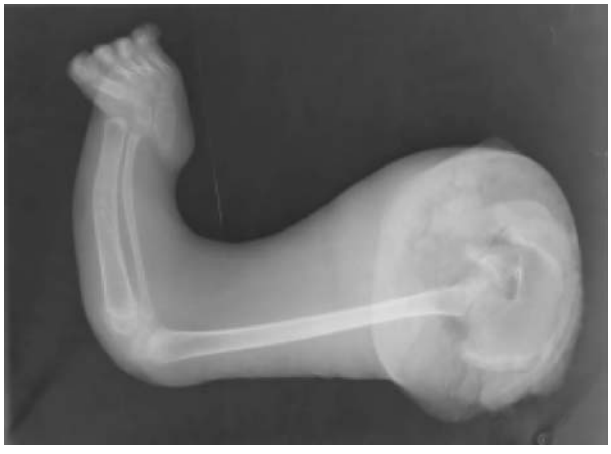

Fig. 4. Postoperative X-ray of the removed leg. Note the almost normal appearance of all the bony elements, including the hip joint

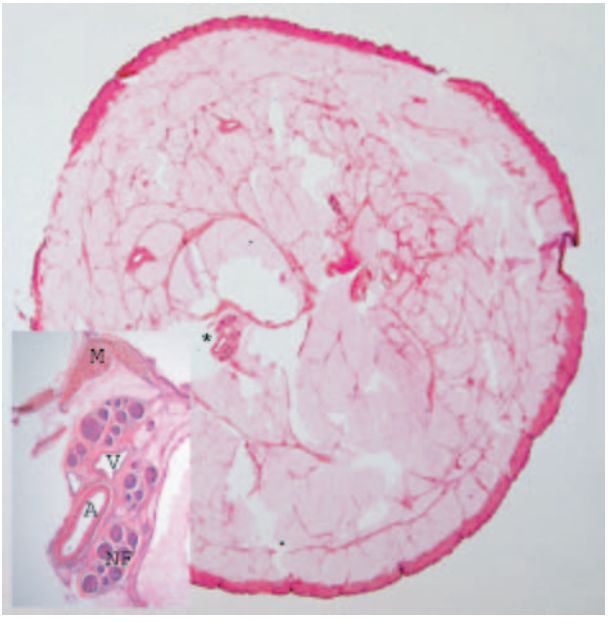

Fig. 5. The central bone has been removed from this H & E stained cross section (thickness 10μ) of the extra leg which is covered with normal skin (epidermis, dermis and adnexal structures). The interior consists of fatty tissue, which is compartmentalised by connective tissue. The * marked area is presented in the inset, which shows triads of artery (A), vein (V) and nerve fascicles (NF). Skeletal muscle fibres are marked with M. H&E, original magnification of inset: $\times 20$

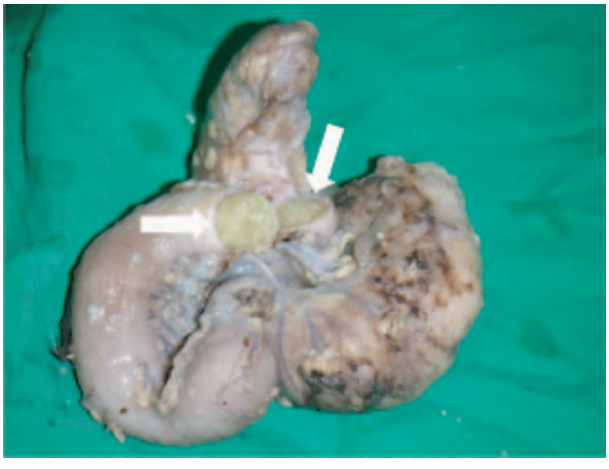

Fig. 6. The removed, stray intestinal loops. Note the granulated mucous content that appears when the intestine is transected (white arrows)

the leg: a hip joint with a femur head, femur, tibia and fibula, an aberrant ankle joint and small bones of the foot (Fig. 4). A cross section of the removed thigh showed mostly fat tissue all the way from the skin to the femur, with some centrally situated blood vessels, a few nerve bundles, and muscle tissue in a very small area (Fig. 5).

Cranial to the iliac bone, where we had palpated the soft, fluctuating mass anticipated to be the myelomeningocele, we found instead isolated intestinal loops. This stray intestine was lying in relation to the ''inside'' of the iliac bone, and had a seemingly normal segmental arterial supply. When opened, the contents proved to be a greyish-white, granulated mucous, apparently sterile (Fig. 6). The histological examination revealed a microscopic appearance identical to colon (Fig. 7). The intestinal loops could easily be dissected away from the underlying dorsal fascia and the myelomeningocele. During the dissection, the myelomeningocele was inadvertently opened. The content was inspected, and the opening was closed, followed by closure of the wound

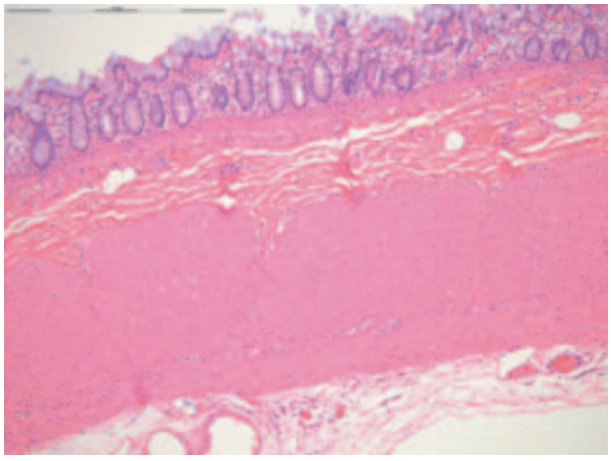

Fig. 7. Section of the stray intestinal loop, showing a colon-type intestine without pathological changes. H & E, original magnification: $\times 10$ (scale bar 500 µm)

in several layers with emphasis on covering the MMC sac. The postoperative course was uneventful.

Discussion

''Foetus in foetu'' or parasitic conjoined twins is in itself an extremely rare, and remarkable condition [15]. Several aspects make the present case even more remarkable. First, the obvious relation between the limb and a spinal defect is rare, although it has been described before [11, 17, 19]. The place of the attachment, the dorsal lumbar region, with no visible fistula connecting the parasitic elements to the abdominal or retroperitoneal compartments, thus respecting the dorsal fascia, is also rather unique. Most of the dorsal limbs that have been reported previously have had some connection to peritoneal organs or compartments [11, 17, 19]. In fact, the great majority of previously reported ''parasitic'' elements have been located intraperitoneally or retroperitoneally [6, 12, 14, 16, 21]. Moreover, most reported cases seem to have had a rather bizarre appearance, whereas the extra leg in our patient, with a normal skin covering that was continuous with that of the child's back, seemed almost normal, were it not for the flaccidity caused by the almost complete lack of muscles. A case has recently been published that bears some resemblance to ours. That child had supernumerary, however grossly malformed, and partially duplicated lower limbs, malformed and partially duplicated pelvis, spina bifida, as well as malformations in organs derived from all of the germ layers [9]. Such anomalies have some similarity with the mouse mutant disorganisation as pointed out by Donnai and Winter [10] and has not been observed as recurrent in families [30]. It is therefore possible that the syndrome is caused by a mutation occurring at a very early stage within the individual, possibly as a mosaic phenomenon.

Until recently, little has been known about the molecular basis for the initiation of limb bud outgrowth, except that it is very complex and not fully understood [5]. This applies also to the duplication of limbs. Some knowledge exists, however. The apical epidermal ridge (AER) plays an essential role via the expression of fibroblast growth factor (FGF) molecules that are necessary for limb formation [28], sometimes in interplay with other factors [1]. Moreover, it is known that a total of 39 human HOX genes appear to be involved in disorders of limb formation [22]. Crossley *et al.* have provided data indicating that FGF8 is a key regulator of limb development that not only induces the limb bud formation but also *sustains* its subsequent development [8]. Interestingly, FGF2 and other factors have been shown to result in the induction of an ectopic limb in animal experiments [13, 31]. For a general review of the mechanisms behind limb formation and malformations, see [23, 30].

The lack of skeletal muscles may be explained by a deficient innervation, as immature myoblasts may fail to differentiate into mature skeletal muscle cells in the absence of appropriate nervous impulses. As previously discussed by Stephens *et al.* [29], the lack of innervation and skeletal muscles in parasitic limbs is significant, and may also bear some important implications for how limbs are formed. Innervation is apparently not a prerequisite for limb morphogenesis, and a parasitic limb may grow even in the absence of skeletal muscles or sensory functions. Other authors have also observed a similar growth of a parasitic limb [20].

There apparently exists a wide variety of related conditions, from real conjoined twins to parasitic twins, ''foetus in foetu'', or teratomas [4, 14]. Stevenson indicates that some of the cases with supernumerary lower limbs in fact may be due to partial twinning [30]. Whether all these conditions represent variations of the same phenomenon (a continuum) or are separate conditions remains to be decided. One may in fact question whether the presented case is a real ''foetus in foetu'', or a maldevelopment caused by errors in the neural crest. Matsuoka *et al.* have investigated the role of the neural crest in the development of the upper limb [25]. It appears possible that neural crest cells also have the potential to induce formation of the lower limb.

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Comments

Fetus congenital malformation as spina bifida is a rare disease. In this particularly case, the relation between the extra limb and the spinal defect is very interesting. Other reported cases have had quite a bizarre appearance, whereas the authors of this article stress the normal aspect of the skin and subjacent tissues related to this malformation, which may support the idea of the potential of neural crest to induce formation of the lower limb.

This article is a very interesting reported case about an extremely rare and complex malformation: the association of an extra leg and spina bifida.

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The authors describe in detail a rare case of a girl with an extra leg attached to her back, combined with a spina bifida and a myelomeningocele.

Never in my life I have seen such a case and probably most of the readers of Acta Neurochirurgica haven't either. The authors provide us with all the necessary information and the relevant pictures to get an impression of this kind of malformation as well as with a thorough discussion about the existing hypotheses.

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Kassel