Letter to the Editor

Congenital polymelia of the neck: a case description

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Introduction

Polymelia is a rare congenital condition observed in both humans and animals, although it is more commonly reported in animals such as chicks, calves, frogs, and lambs (1-4). Characterized by the presence of extra limbs, polymelia often results in underdeveloped or nonfunctional appendages that vary in size and structure. Despite the rarity of cervical polymelia, this report provides a detailed description of the deformity, along with an in-depth discussion of the diagnosis and treatment process.

Case presentation

All procedures performed in this study were in accordance with the ethical standards of the institutional and/or national research committee(s) and with the Helsinki Declaration (as revised in 2013). Written informed consent was obtained from the patient for publication of this article and accompanying images. A copy of the written consent is available for review by the editorial office of this journal.

A 30-year-old female presented with a deformed upper limb in the neck, a condition observed since birth. Over the previous year, she had experienced intermittent neck pain, soreness, and discomfort, prompting her to seek medical attention. The deformity leaned toward the left neck and shoulder and had grown slowly and continuously with age. She reported no sensorimotor impairment, unsteady walking, or sensation abnormalities. There was no family history of genetic diseases, consanguinity, maternal exposure to teratogenic drugs, or radiation during pregnancy.

Physical examination revealed limited cervical spine movement and reduced curvature, and the contraction of the left sternocleidomastoid muscle had a palpable cord-like contracture. A deformed and bent upper limb was present on the left side of the neck. The base of the deformed limb was thicker than the distal end. There were skin folds visible at the bend and undifferentiated parts of the fingers and nails at the end. The limb was palpable, but it demonstrated no voluntary or spontaneous movement (*Figure 1A-1C*). There were bulging cervical spinous processes connected to the deformed upper limb. There was no obvious lateral bending or deformity of the thoracolumbar spine and no abnormalities in the patient's other limbs or internal organs. Although the patient could perform daily physical labor, her condition seriously affected her sleep, and her appearance was a constant concern. In order to improve her quality of life and appearance, the patient strongly desired surgical resection.

X-ray (anteroposterior and lateral) showed bony structures on the left side of the neck connected to the second and third spinous processes, indicative of congenital developmental abnormalities (*Figure 2A,2B*).

On computed tomography (CT) with three-dimensional (3D) reconstruction, three tubular bones could be observed in the neck. The spinous processes of C2 and C3 vertebrae were abnormally shaped and connected to the longer tubular bones at the proximal end. The first (proximal) longer tubular bone formed an acute angle with the second (middle) and third (distal) short tubular bones. The three tubular bones were connected by fibers without bony connection. The terminal ossicles of the odontoid process of the C2 vertebrae and the lamina of C4 vertebrae were incompletely closed (*Figure 3A-3D*).

Magnetic resonance imaging (MRI), including, T1-

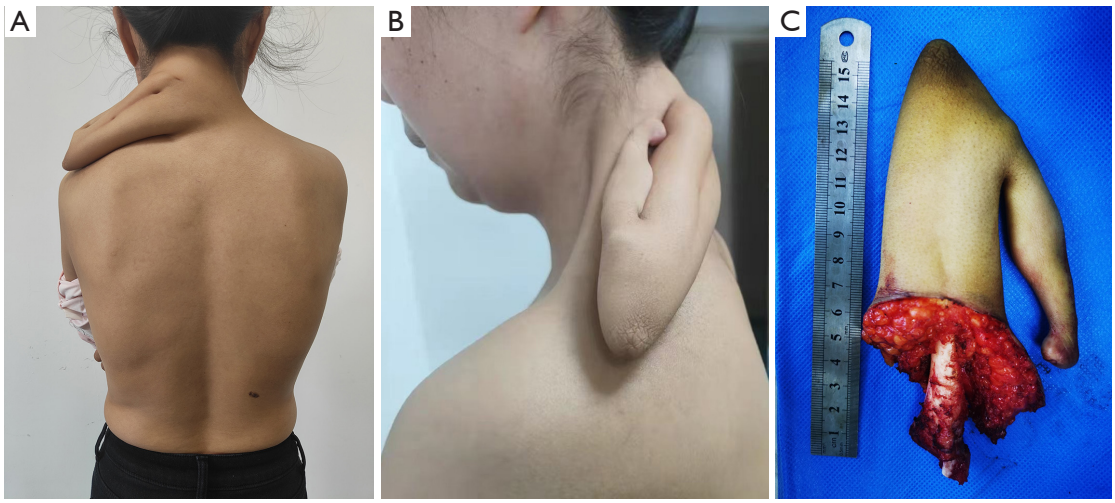

Figure 1 Preoperative and resected gross specimen images. (A) Preoperative orthographic image. (B) Preoperative lateral image. (C) Resected gross specimen images show a deformed upper limb on the left side of the neck.

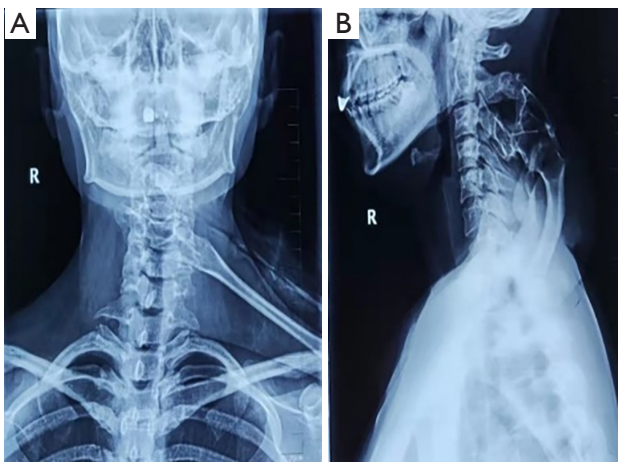

Figure 2 Preoperative cervical X-ray images. (A,B) The anteroposterior and lateral X-rays showed bony structures on the left side of the neck, connected to the second and third cervical spinous processes.

weighted imaging (T1WI), T2-weighted imaging (T2WI), and short tau inversion recovery (STIR), revealed an epidural lipoma at the level of C2–C3 vertebrae, closely related to subcutaneous fat on the posterior cervical spine (*Figure 4A-4C*). No abnormalities were noted in the thoracic spine.

Neck computed tomography angiography (CTA) revealed a small branch originating from the left subclavian artery supplying the abnormal limb. Head and neck CTA showed no additional abnormalities.

The deformed limb was surgically removed under general anesthesia. An arc-shaped surgical incision, approximately 10 cm in length, was made adjacent to the C2 and C7 vertebrae. The skin, subcutaneous tissue, and fascia were incised sequentially, followed by dissection of the muscle and periosteum to expose the bony connection between the C2 and C3 spinous processes, as well as the proximal end of the deformed limb. The deformed limb and the bony connection between the C2 and C3 spinous processes were carefully severed with an ultrasonic bone knife. The blood vessels and nerves supplying the deformed limb were isolated, electrocoagulated, and ligated before the limb was completely excised. No intervention was performed on the epidural lipoma or the soft tissue fat at the back of the neck, and no additional blood vessels or nerves were altered. The exposed ends of the C2 and C3 spinous processes were trimmed, and the incision was thoroughly irrigated to ensure hemostasis. After the absence of active or significant bleeding was confirmed, the incision was closed in layers. The deformed limb of the neck was completely removed, there was no obvious bulge on the neck, there was little bleeding during the operation, and the patient returned to the ward safely. The amputated limb measured approximately 15 cm in length and resembled the left upper limb, consisting of a humerus, elbow joint, and indistinct structures of the ulna, radius, and phalanges (*Figure 1C*). Postoperatively, there were no abnormalities in cardiac ultrasound, abdominal ultrasound, electrocardiography, or laboratory tests. The patient recovered well, with no

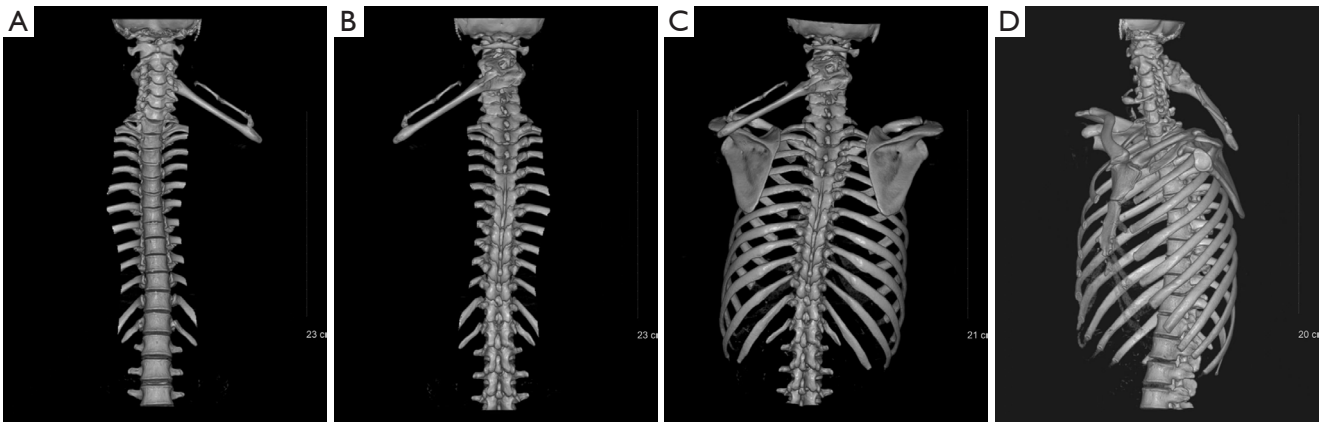

Figure 3 Computed tomography volume rendering images. (A-D) Three tubular bones were observed in the neck. The spinous processes of the C2 and C3 vertebrae were abnormally shaped and connected to the longer tubular bones at the proximal end. The first (proximal) longer tubular bone formed an acute angle with the second (middle) and third (distal) short tubular bones. The three tubular bones were connected by fibers without bony connection. The terminal ossicles of the odontoid process of the C2 vertebra and the lamina of C4 vertebra were incompletely closed.

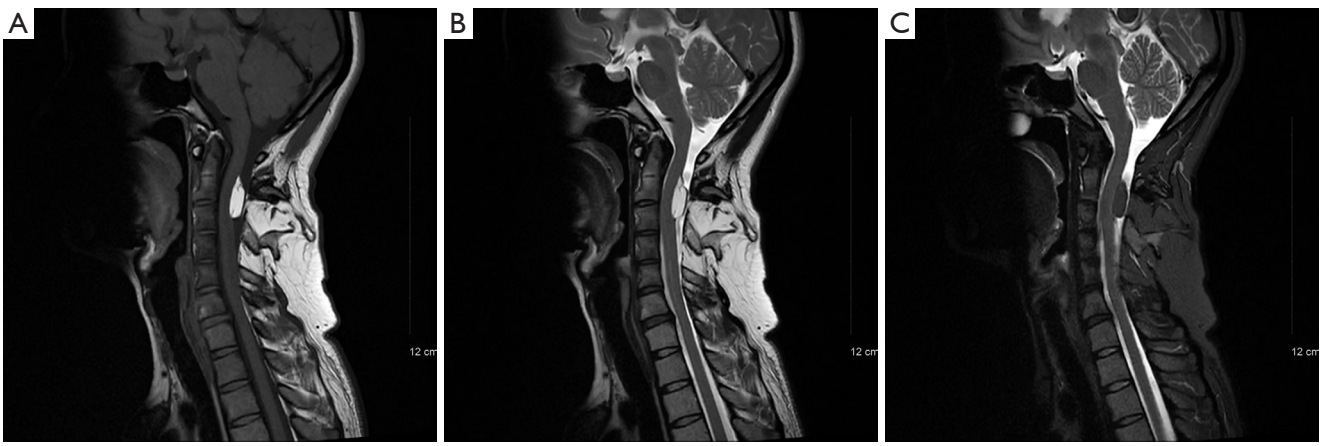

Figure 4 MR images. (A) Epidural lipoma on T1-weighted imaging showed high signal intensity. (B) T2-weighted imaging of the epidural lipoma showed high signal intensity. (C) A short tau inversion recovery sequence of the epidural lipoma showed high signal intensity. An MR plain scan of the cervical and thoracic spine showed an epidural lipoma at the level of the C2–3 vertebrae, closely related to the subcutaneous fat on the posterior side of the cervical spine. MR, magnetic resonance.

significant complications noted during follow-up. The postoperative follow-up X-ray is shown in *Figure 5A,5B*.

Discussion

Polymelia is a rare congenital anomaly characterized by the presence of an accessory limb, typically attached to the spine. It is rare in humans, with a worldwide incidence of approximately 1.47/100,000 (5). In humans, polylimb deformities are usually found on the chest, back, lumbosacral region, inner thigh, or acetabulum (6-15). Polylimb deformity is largely considered to be an asymmetrical conjoined twin, specifically a parasitic fetus. Parasitic fetuses appearing internally are considered to be endoparasitic, while those attached to the surface are considered ectoparasitic (15). The pathogenesis of this congenital malformation remains unclear. Studies employing experimental models have found that various mutagenic drugs (aspirin, acetazolamide, cadmium, adenine, and aminophylline), retinoids (16), mechanical damage (17),

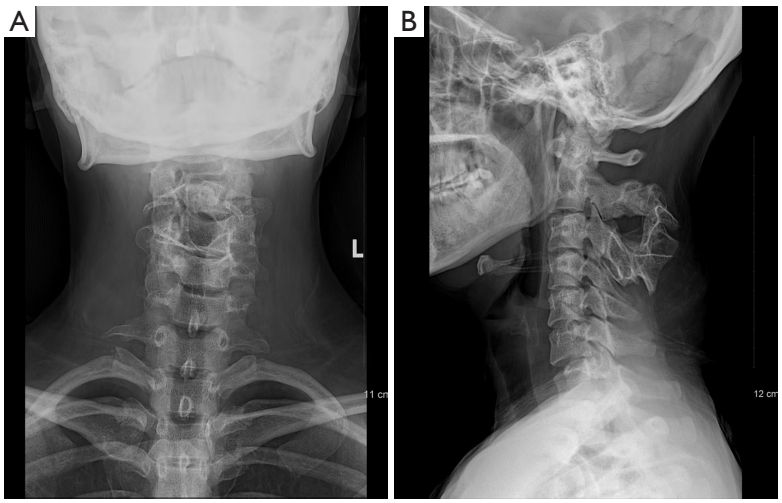

Figure 5 Postoperative cervical X-ray images. (A,B) Postresection X-ray of the cervical spine showed bony structures of the spinous processes of the C2 and C3 vertebrae.

familial inheritance, radiation, or environmental factors can induce this condition, but these findings cannot be entirely extrapolated to humans. Otsuji *et al.* (18) reported that administration of busulfan (butyl dimethyl sulfonate) to maternal rats caused limb deformities. More recent research has found that the monoallelic and biallelic variants in *LEF1* are the genetic underpinnings of limb deformities (19).

Limb development is a complex process regulated by numerous genes (20). Genetic testing on a child with congenital malformations of three lower limbs identified variations in three collagen-related genes that may be critically involved in multiple malformations (21). Our patient had polymelia attached to the spinous processes of cervical vertebrae 2–3. Reports of cervical polylimb deformities are rare. The patient, a 30-year-old married woman, had abnormal limb buds on her neck from birth that gradually developed. She had experienced no intellectual or other abnormalities over the previous 30 years, and her daily activities were not affected. There was no family history of such conditions, nor was there a history of maternal radiation exposure or teratogenic drug use during pregnancy. Genetic testing was not performed on this patient, so genetic causes could not be investigated.

Limb differentiation occurs roughly between the fourth and fifth weeks of embryonic development (22). The severity and manifestations of limb deformities can help determine when teratogenic events have occurred during development. According to Swanson *et al.*'s classification of limb deformities (23), the deformed limb of this patient was similar to the normal upper limb on one side and thus could be classified as type III, a duplicate deformity. Saaiq *et al.* (6) proposed a morphological classification of accessory lower limbs based on the literature on lower limb polysomy deformities. Although the deformity in our case appeared in the neck, it could be classified as category Ib (the shape of the amputated limb was similar to the left upper limb, the proximal part was similar to the upper arm, and the distal part was similar to the forearm and palm), which includes the accessory limb being accompanied by spinal insufficiency and moderate development. Polylimb deformity is more prevalent in women, and the female-to-male ratio is 4:1 (6,24). Theories explaining the higher prevalence in females include genetic predispositions and hormonal influences, although these theories have not been verified.

Imaging examinations are crucial in diagnosing and planning treatment for cervical polylimb deformities. CT scans with 3D reconstruction and neck CTA can show cervical vertebral lesions, attachment points of the deformed limbs, and blood supply arteries. MRI is essential for evaluating related conditions within the spinal canal, such as lipomas and meningoceles. Studies point to a strong association between accessory limbs and spina bifida (25,26). In our case, CT showed that the spinous processes of C2 and C3 vertebrae were attached to a deformed limb, the vertebral lamina of the C4 vertebra was incompletely closed, forming spina bifida, and the malformed limb was supplied by a small branch from the left subclavian artery.

MRI revealed an epidural lipoma at the C2–C3 vertebrae, likely a meningocele, similar to previous reports of lower limb polylimb deformities.

Prior to surgery, the patient underwent CT and MRI examinations, which facilitated 3D printing of models for surgical planning. The proximal tubular bone of the posterior polymelia was connected to the spinous process, facilitating a posterior approach for the removal of the polymelia's base and separation from the surrounding tissue structures. MRI also indicated that the epidural lipoma and posterior cervical lipoma were not directly related to the polymelia, and the patient showed no symptoms of lower limb numbness, so no specific treatment was required. Echocardiography was performed to assess potential cardiac abnormalities before surgery. Although Doppler ultrasound is not essential, it was considered helpful given the anatomical variations associated with polymelia. This imaging technique enabled the identification of the deformed vascular pedicle, significantly enhancing the safety and probability of success for the procedure.

Surgical excision is the only effective treatment for multilimb deformity. There are no established guidelines on the timing of surgery. Acharya *et al.* (25) reported a case of an 18-month-old girl with multiple limb deformities preventing her from standing, highlighting the need for early intervention to improve appearance and quality of life. Kubaszewski *et al.* (27) proposed that the goal of surgical treatment is to restore spinal stability. Meanwhile, Tykocki *et al.* (28) suggested that among various cervical spine lesions, attention should be paid to clinical and imaging outcomes, biomechanical instability, and related complications after surgery. Preoperative imaging is vital to assessing the extent of the deformity and any associated complications, such as teratomas, internal organ duplication, scoliosis, or urinary system diseases. In this case, the patient had a cervical polymelia. The surgery was successful, resulted in no postoperative complications, and significantly improved the patient's quality of life. At the time of writing, the range of motion in the neck has increased, pain has been relieved, and aesthetic appearance has improved. Additionally, the physician actively guides the patient in neck rehabilitation exercises to improve coordination, with long-term follow-up being scheduled to monitor various postoperative indicators and functions.

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Footnote

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Conflicts of Interest: All authors have completed the ICMJE uniform disclosure form (available at https://qims.amegroups.com/article/view/10.21037/qims-24-938/coif). The authors have no conflicts of interest to declare.

Ethical Statement: The authors are accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved. All procedures performed in this study were in accordance with the ethical standards of the institutional and/or national research committee(s) and with the Helsinki Declaration (as revised in 2013). Written informed consent was obtained from the patient for publication of this article and accompanying images. A copy of the written consent is available for review by the editorial office of this journal.

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