Case Report

POLYMELIA ASSOCIATED WITH LIPOMYELOMENINGOCELE: A CASE REPORT


Polymelia is a congenital defect involving limbs, in which the affected individual has more than the usual number of limbs. The supernumerary limb can be either shrunk or deformed and can be attached to various regions of the body. Supernumerary limb attached to a lipomyelomeningocele is very rarely reported and we report a case of 4 months old male baby born to parents of non-consanguineous marriage presented with supernumerary lower limb of length 9cm attached to lipomyelomeningocele at the lumbosacral region of size 18x13cm. Surgery was performed with untethering of the spinal cord, debulking of the lipoma and excision of the supernumerary limb. On histopathological examination lipomatous tissue was confirmed and on meticulous dissection of the supernumerary limb, it had bone exposed at one end and the other end had two digits of length 1.4 and 1cm. The limb had features of sole with partially developed plantar aponeurosis, muscles and blood vessels. Thigh and leg were not fully formed. These malformations can have multifactorial etiology and antenatal diagnosis can be done by ultrasonogram. Early surgical management can prevent neurological complications.

KEY WORDS: Polymelia, lipomyelomeningocele, supernumerary, congenital.

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INTRODUCTION

Number of rare congenital anomalies are reported in human and animals. One such rare anomaly in human and most land-dwelling animals is polymelia which means having five or more limbs. The extra limb or supernumerary limb is most commonly deformed and it can be attached to various regions of the body. It has been demonstrated that the majority of human and animal malformations are due to multifactorial etiology. Lipomyelomeningocele is a form of Occult Spinal Dysraphism in which a subcutaneous fibro fatty mass traverses the lumbodorsal fascia, causes a spinal laminar defect, displaces the dura, and infiltrates and tethers the spinal cord [1]. Polymelia attached to lipomyelomeningocele is very rarely reported.
CASE REPORT

We report a case of a 4 months old male baby born to parents of non-consanguineous marriage and with no family history of congenital anomalies presented with supernumerary lower limb associated with lipomyelomeningocele. Computed tomography confirmed the presence of lipomyelomeningocele at the lumbosacral region and it was of size 18x13cm. A supernumerary lower limb which was 9cm in length was found attached to the lipomyelomeningocele (Fig.1). Surgery was performed with untethering of the spinal cord, debulking of the lipoma and excision of the supernumerary limb, the specimen was sent for histopathological investigation. Lipomatous tissue was confirmed and on meticulous dissection of the supernumerary limb, it had bone exposed at one end and the other end had two digits of length 1.4 and 1cm (Fig.2&3). The limb had features of sole with partially developed plantar aponeurosis, muscles and blood vessels. Thigh and leg were not formed fully. Histo-pathological examination of the supernumerary limb showed incomplete limb bud with endochondral ossification (Fig.4).

DISCUSSION

Limb malformations occur in approximately 6 per 10,000 live births with 3.4 per 10,000 affecting the upper limb and 1.1 per 10,000 affecting the lower limb. Abnormalities of the limbs can be meromelia, amelia, phocomelia or polymelia. These defects are often associated with other birth defects involving the craniofacial, cardiac and genitourinary systems. Although these abnormalities are rare and mainly hereditary, cases of teratogen – induced limb defects have been documented [2].

Polymelia is an extremely uncommon congenital entity rarely reported in humans, though it is frequently reported in animals. Polymelia is called as cephalomelia- attached to head, notomelia- attached to backbone, thoracomelia - attached to thorax and called as pyromelia if the extra limb is attached to pelvis [3]. Cases with partial duplication of lower limb and renal agenesis have been reported [4, 5]. A case of lower limb bud on the left posterior thigh was reported [6].

Limb differentiation occurs roughly between 4th
and 5th weeks of embryonic development and it follows a dorsal to ventral and proximal to distal pattern. Initially, two pairs of limb buds – anterior and posterior – protrude from both sides of the embryo, and comprise cells of ectoderm and mesoderm. Their interaction is responsible for cell positioning and limb differentiation. The covering ectodermal layer of the limb bud is termed the apical ectodermal ridge (AER). The zone of proliferating activity (ZPA), another group of cells is located subjacent to the apical ectodermal ridge. Both are necessary for limb development. Mesodermal cells in the ZPA stimulate AER formation and the AER maintain the ZPA. The level and manifestation of limb deformity can thus be used to determine the approximate timing of the teratogenic event that occurred during limb development. As the AER grows more distal, the induced mesoderm cells, comprising rudimentary parts of the limb, can continue to grow without any developmental interference even if the AER is transplanted to the adjacent region. This leads to an assumption that duplication of the limb arises from the influence of the AER with abnormal splitting creating two sets of limbs [3].

Limb development is a very complex process involving precise gene regulation fundamental to normal growth. Limb development involves a very large number of genes. One gene widely associated with the development of supernumerary limbs is the mouse mutant disorganization Ds gene [OMIM: 223200], which is a semi dominant gene with variable penetrance in heterozygotes and lethality in homozygotes; 67% of heterozygotes have multiple defects and the rest have single defects, in which polymelia is prominent [7].

In experiments with chick embryo, fibroblast growth factor-1 (FGF-1) FGF-2, FGF-4, FGF-8 is able to stimulate development of additional limb [8].

Sometimes an embryo starts developing as a conjoined twin, but one twin degenerates completely except for one or more limbs, which end up attached to the other twin resulting in the formation of supernumerary limb. Women taking hormone like progesterone, during pregnancy have chances of the structural anomalies like polydactyl, polymelia and other congenital defects [3]. Antenatal screening with ultrasonography can be a useful to diagnose supernumerary limb in utero.

Lipomyelomeningocele is a form of Occult Spinal Dysraphism. The diagnosis of tethered cord is made when the tip of the conus is below the level of the lower border of the L1 vertebral body. The embryogenesis of lipomyelomeningocele is that during neurulation, the ectoderm on either side of the neural plate come close together as the neural tube closes. When neural tube fusion is complete the ectoderm detaches on either side in an event called disjunction and fuses. If, however, the disjunction occurs before the closure of neural tube is complete, mesenchymal cell gain access to the central canal of neural tube. The mesenchymal cells then differentiate into fatty tissue to form lipomyelomeningocele, which is essentially a mass of fat extending from the conus medullaris to the subcutaneous plane underly-ing an intact skin. An error in the canalization of caudal cell mass may be another explanation in the origin of a lipomyelomeningocele [9].

There are two main variants of lipomyelomeningocele with a spectrum of intermediate forms. The dorsal variant consists of lipomatous mass extending from the dorsal aspect of the cord to the subcutaneous tissue immediately above. The nerve roots arise from the ventral and lateral aspect of the cord, all within the subarachnoid space and the nerve traverse the lipomatous mass situated dorsally. This variant is surgically more amenable to a safe excision of lipomatous mass, identification and division of the filum terminale and reconstitution of the neural tube. In the caudal variant, the lipoma follows the filum terminale and nerve roots are enmeshed within the fatty tissue making excision hazardous [9].

Congenital lumbosacral lipoma can be responsible for progressive defects. The general feeling is that tethering of roots, filum, or cord probably explains this evolution, and that untethering of these structures could prevent late deterioration. Therapeutic objectives were spinal cord untethering and decompression, sparing of functional neural tissue and prevention of retethering. Procedures used to achieve these goals were subtotal removal of the lipoma,
intraoperative monitoring, duroplasty, and sometimes closure of the placode [10].

Majority of patients with lipomyelomeningocele have bladder dysfunction. Urodynamic assessment is helpful as part of the neurological evaluation and in directing patient care[11]. Older children are more likely to present with urological and neurological complaints. Surgical correction in infancy provides a degree of reversibility not seen in older children [12]. Electromyographic studies and uroflowmetry are required to assess the true sphincteric outcome following surgery [13].

CONCLUSION

Successful management of a patient with spinal dysraphism demands a thorough understanding of the pathologic anatomic characteristic, aided by appropriate preoperative studies, is necessary. Radiologist plays a significant role in patients with polymelia to assess for additional congenital anomalies before surgical intervention. As only few reports are available regarding polymelia, more detailed study on it is needed.

ABBREVIATIONS

ZPA - zone of proliferating activity
AER - apical ectodermal ridge

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REFERENCES


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