

# Spinal Canal Involvement in Solitary Infantile Myofibromatosis: Case Report

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## Abstract

Infantile myofibromatosis involving the spinal canal is very rare; only 11 cases have been reported so far in the literature. The authors present a case of an 18-month-old male child who presented with the history of dribbling of urine and weakness in bilateral lower limbs since 2 months. MRI of spine revealed single intramedullary intradural space-occupying lesion (SOL) at D1 to D2.

## Keywords

- ▶ solitary infantile myofibromatosis
- ▶ spinal canal
- ▶ intramedullary

The patient underwent laminectomy with excision of SOL with biopsy report suggestive of benign nerve sheath tumor, and immunohistochemistry report revealed desmin negative, smooth muscle actin positive, and S-100 focally positive infantile myofibromatosis. The patient gradually recovered and had a clear stream of urine with improved movements and tone of bilateral lower limbs at the time of discharge.

## Introduction

Infantile myofibromatosis is the most common fibrous disorder of infancy and early childhood. It was first described by Stout<sup>1</sup> in 1954 who named this disease congenital generalized fibromatosis. In 1981, Chung and Enzinger<sup>2</sup> renamed the disease as infantile myofibromatosis to emphasize the microscopic resemblance to smooth muscle tissue. It may occur in two distinct forms: multicentric and solitary. In both cases, involvement of the central nervous system is unusual. Spine myofibromas are exceptional, and most of the cases reported in the literature represent secondary locations of visceral lesions. Here, we are reporting good outcome in the case of intramedullary solitary type of infantile myofibromatosis with no visceral involvement.

## Case Report

### History and Examination

An 18-month-old male child presented with history of dribbling of urine and weakness in bilateral lower limbs noticed by his parents since about 2 months for which the patient underwent MRI scan of spine.

MRI revealed a single intradural intramedullary space-occupying lesion at D1 to D2 (see ▶ **Fig. 1**). The patient was then referred to Department of Neurosurgery, M.B.S Hospital, Kota for further management.

The X-ray of long bones, blood investigations, and ultrasonogram of abdomen and pelvis were normal and showed no apparent visceral lesions. Clinically, the child had normal growth with normal body weight but had paraparesis (power ½) with sluggish deep tendon reflexes and intact anal tone.

### Operative Course

C-7 to D-2 laminectomy was done under general anesthesia after localizing D1 in prone position. Dura was identified and midline cordotomy was done with complete excision of encapsulated intramedullary SOL of ~2 × 1.4 cm in size.

### Postoperative Course

On the first postoperative day, the patient was taking oral feeds and passed stools. On the second postoperative day, a catheter free trial was given and a clear stream of urine with improved motor power of bilateral lower limbs

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April 9, 2015

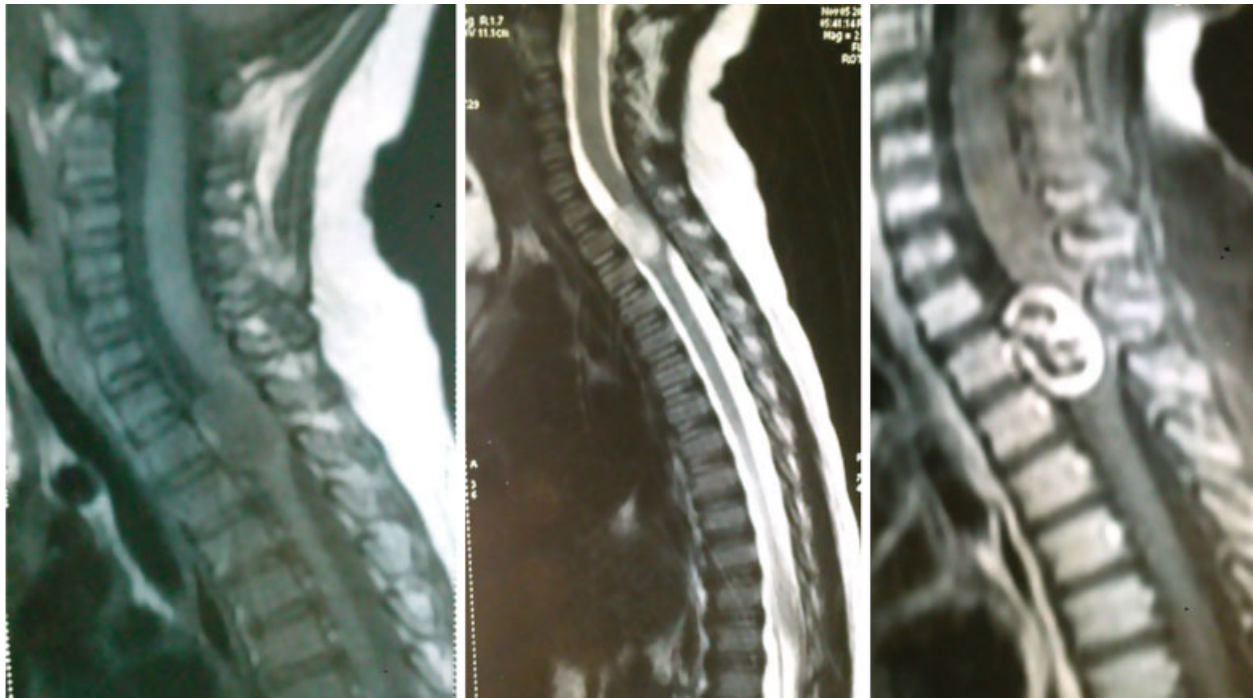
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**Fig. 1** Preoperative MRI images.

(MRC grade 2/5) was observed. The patient gradually recovered and was discharged on 14th postoperative day. The patient was under regular follow-up and within a span of 2 months was able to walk with support.

### Histopathology

Encapsulated SOL was sent for histopathological examination which revealed well-circumscribed spindle cell tumor composed of oval to elongated spindle cells arranged in sheets. Neoplastic cells were desmin negative (see ►**Fig. 2**), smooth muscle actin positive (see ►**Fig. 3**),

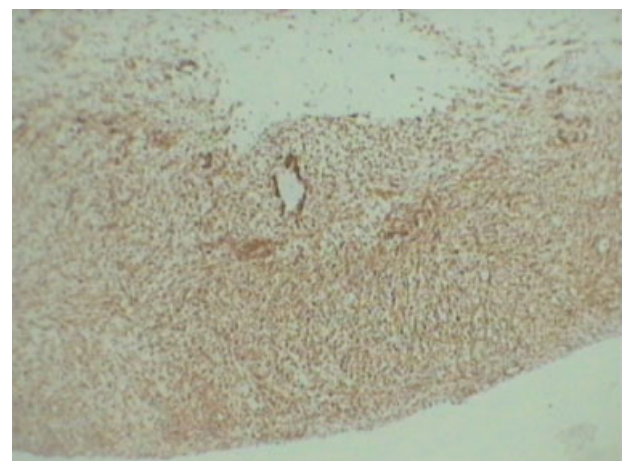
S-100p focally positive, KI-67 positive, 2 to 3%, PAN cytokeratin negative suggestive of infantile myofibromatosis (see ►**Fig. 4**).

### Discussion

Infantile myofibromatosis involving the spinal canal is very rare; 11 cases have been reported so far in the literature including 4 cases of the solitary form and a single case of intramedullary solitary form (►**Table 1**).<sup>3-6</sup> To the best of our knowledge, we are reporting second case of



**Fig. 2** Desmin—negative.



**Fig. 3** Smooth muscle actin—positive.

ACCESSION NO : 0002ML010526	AGE : 1 Years	SEX : Male	DATE OF BIRTH :
DRAWN : 02/12/2013 00:00	RECEIVED : 04/12/2013 11:13	REPORTED : 10/12/2013 17:20	
REFERRING DOCTOR : DR. S N GAUTAM	CLIENT PATIENT ID :		
<b>Test Report Status</b> Final	<b>Results</b>		
<b>HISTOPATHOLOGY</b>			
<b>CUSTOM IHC PANEL</b>			
<b>CUSTOM IHC PANEL</b>			
ADDITIONAL COMMUNICATION	DECEMBER 06, 2013.		
SPECIMEN	ADDITIONAL IHC MARKERS ARE BEING CARRIED OUT. REPORT WILL FOLLOW.		
MICROSCOPIC EXAMINATION	RECEIVED ONE PARAFFIN BLOCK REFERENCE NO. 5870/13. OF SACRAL MASS.		
	SECTIONS SHOW WELL CIRCUMSCRIBED SPINDLE CELL TUMOUR COMPOSED OF OVAL TO ELONGATED TO SPINDLY CELLS ARRANGED IN SHEETS. THE CELLS IN THE CENTRE OF THE LESION ARE PREDOMINANTLY SPINDLE SHAPED AND ARE SET IN HYALINIZED STROMA. THERE IS A TRANSITION IN THE CELL TYPE AT THE PERIPHERY WHICH ARE MORE PLUMP, OVOID TO ELONGATED WITH OVAL NUCLEI AND ARE SET IN THE MYXOID STROMA. THE STROMA ALSO SHOWS COMPRESSED SINUSOIDAL BLOOD VESSELS. NO ATYPIA OR ATYPICAL MITOSIS IS SEEN.		
<b>DESMIN IHC, TISSUE/PARAFFIN BLOCK</b>			
DESMIN IHC	NEGATIVE.		
<b>SMOOTH MUSCLE ACTIN IHC, TISSUE/PB</b>			
SMOOTH MUSCLE ACTIN, IHC	POSITIVE.		
<b>S-100P, TISSUE/PARAFFIN BLOCK</b>			
S-100 P	FOCALLY POSITIVE.		
<b>KI-67, TISSUE/PARAFFIN BLOCK</b>			
KI-67	POSITIVE, 2-3%.		
<b>PAN CYTOKERATIN IHC, TISSUE/PB</b>			
PAN CYTOKERATIN (IHC)	NEGATIVE.		
<b>INTERPRETATION:</b>			
DIAGNOSIS	INFANTILE MYOFIBROMATOSIS.		
Comments			
THE COMMENTS PRINTED IN "LETTER" ARE DEFINED ACTIONS UNDERTAKEN BY LABORATORY PRIOR TO RELEASE OF THE FINAL DIAGNOSIS.			
IHC carried out on formalin-fixed paraffin-embedded sections. All positive controls show appropriate positive immunostaining. Negative control slide does not show immunostaining.			
Detection system used is - HRP POLYMER.			

Fig. 4 Immunohistopathology report.

intramedullary solitary type IM localized within the spinal cord without any visceral involvement.

The etiology and pathogenesis of IM are still obscure. The solitary form of infantile myofibromatosis mostly occurs in the soft tissues of the head and neck, followed by the upper extremities and trunk. Patients usually have a poor clinical symptomatology, except for occasional pain caused by compression of adjacent nerves.<sup>7-9</sup>

Our patient's symptoms of dribbling of urine and weakness in bilateral lower limbs pointed toward compressive symptoms of tumor in spinal canal.

The usual clinical course of the solitary form is initial rapid growth followed by spontaneous regression within the first 2 years.

Conservative management is usually adopted for those without visceral involvement and complications. We did surgical decompression that was probably the correct

decision in our case as literature says that younger patient age at diagnosis is associated with a more rapid expansion rate for infantile myofibromatosis.<sup>10</sup>

In aggressive cases, there is limited experience of success with radiation therapy, different combination of chemotherapy,<sup>11,12</sup> steroid injection,<sup>11,12</sup> and  $\alpha$  interferon.

## Conclusion

Given the rarity of this condition, correct preoperative diagnosis could not be made initially until after histopathological examination. Though a rare disorder, IM must be suspected when evaluating children who present with either multiple lytic bone lesion or solitary/multiple tumors in the soft tissues, particularly during the neonatal or infancy period. Early surgical decompression of spinal cord led to improved neurological outcome in the patient with infantile myofibromatosis.

**Table 1** Reported patients with infantile myofibromatosis involving the spinal canal

Authors and year	Age, sex	Symptoms	Disease type	Lesion location	Spinal cord invasion	Management	Outcome
Davies et al, 1994	Birth, F	Brachial palsy	Multicentric	Cervical spinal cord, left side neck	No	Chemotherapy	Good
Giannini et al, 1995	33 years, F	Paralysis of left leg	Multicentric	Iliac bone, posterior vertebral element, left side vertebral column	No	Chemotherapy	Good
Wada et al, 1998	Birth, F	Paraplegia, bladder and bowel dysfunction	Multicentric	Bone, left side of vertebra, intrapelvic, spinal cord (L3-5)	No	Partial removal	Good
Dimmick and Wood, 1983	Birth, M	Quadripareisis	Multicentric	Skin, tongue, spinal cord (C3-T3)	Yes	Supportive care	Good
Christensen et al, 1961	Birth, M	No spontaneous movement, respiratory failure	Multicentric	Bone, skin, heart, liver, brain, thymus, gut wall, kidney, spinal cord (T2-4, T8-10)	Yes	Supportive care	Died (10 days)
Altamani et al, 1985	Birth, F	Quadripareisis, dyspnea	Multicentric	Bone, right side of neck	Yes	Supportive care	Died (75 days)
Stewart et al, 1989	Birth, M	Paraplegia, respiratory failure	Multicentric	Skin, muscle, lung, spinal cord (T6-8), myocardium, GI tract	Yes	Supportive care	Died (45 days)
Beyer et al, 1990	5 years, M	Local pain	Solitary	Sacrum	No	Surgical removal	Good
Asirvatham et al, 1994	18 years, M	Local pain	Solitary	Spinal cord (C2)	No	Surgical removal	Good
Tamburrini et al, 2003	4 months, M	Spastic paraparesis	Solitary (intramedullary)	Spinal cord (C7-conus)	Yes	Partial removal	Good
Eun Ji Kim, et al, 2013	8 months, F	Paraplegia	Solitary	Spinal cord (T6-conus)	Yes	Partial removal	Good
Present case	18 months, M	Paraparesis, bladder dysfunction	Solitary (intramedullary)	Spinal cord (T1-2)	Yes	Complete removal	Good

**References**

- 1 Stout AP. Juvenile fibromatoses. *Cancer* 1954;7(5):953–978
- 2 Chung EB, Enzinger FM. Infantile myofibromatosis. *Cancer* 1981; 48(8):1807–1818
- 3 Beyer WF, Kraus J, Glückert K, Goldmann AR. [Solitary infantile myofibromatosis of the sacrum]. *Z Orthop Ihre Grenzgeb* 1990; 128(5):473–476(Ger)
- 4 Asirvatham R, Moreau PG, Antonius JI. Solitary infantile myofibromatosis of axis. A case report. *Spine* 1994;19(1):80–82
- 5 Tamburrini G, Gessi M, Colosimo C Jr, Lauriola L, Giangaspero F, Di Rocco C. Infantile myofibromatosis of the central nervous system. *Childs Nerv Syst* 2003;19(9):650–654
- 6 Kim EJ, Wang KC, Lee JY, et al. Congenital solitary infantile myofibromatosis involving the spinal cord. *J Neurosurg Pediatr* 2013;11(1):82–86
- 7 Cardia E, Molina D, Zaccone C, la Rosa G, Napoli P. Intracranial solitary-type infantile myofibromatosis. *Childs Nerv Syst* 1993;9(4):246–249
- 8 Kaplan SS, Ojemann JGM, Grange DK, Fuller C, Park TS. Intracranial infantile myofibromatosis with intraparenchymal involvement. *Pediatr Neurosurg* 2002;36(4):214–217
- 9 Rutigliano JM, Pollack IF, Ahdab- Barmada M, Pang D, Leland-Albright A. Intracranial infantile myofibromatosis. *J Neurosurg* 1994;81:539–543
- 10 Roggli VL, Kim HS, Hawkins E. Congenital generalized fibromatosis with visceral involvement. A case report. *Cancer* 1980;45(5):954–960
- 11 Holzer-Fruehwald L, Blaser S, Rossi A, Fruehwald-Pallamar J, Thurnher MM. Imaging findings in seven cases of congenital infantile myofibromatosis with cerebral, spinal, or head and neck involvement. *Neuroradiology* 2012;54(12): 1389–1398
- 12 Sinha AK, Jesudason EC, Mallucci CL, Losty PD, Kokai G, Pizer BL. Infantile myofibromatosis of the craniovertebral junction. *Br J Neurosurg* 2008;22(4):575–577