

Unilateral limb hypertrophy and shoulder weakness in a 37-year-old woman

Clinical record

A 37-year-old woman presented with a 2-month history of progressive restriction of movement of the left shoulder joint, associated with episodic pain over the region. She was unable to raise her left arm above the shoulder. The pain was mild, deep-seated, non-radiating and relieved by supporting the left elbow. She denied any weakness of the arm muscles.

The patient also complained of diminished pain and temperature sensation over the affected limb, as evidenced by painless scalds of the fingers of her left hand — the first about 4 years previously, and the second, 7 days before presentation. She had had mild pain in the left shoulder region infrequently for the previous 17 years, and had noticed enlargement of her left arm since childhood.

She had not noticed any lymphadenopathy. She had normal facial sensation and denied any history of diplopia, dysarthria, dysphagia, hoarseness, vertigo, sphincter abnormalities or weakness and ataxia of any extremity. There was no history of neck pain, trauma, radiation or prolonged fever. She had no hypertension or diabetes, and her family and obstetric history were unremarkable.

Examination: The patient's left upper limb was larger than the right (Box, A). The other limbs appeared normal. Girth of the left upper limb was 51.6 cm at the shoulder (right, 46.5 cm); 35.8 cm at the mid-arm (right, 31.4 cm); and 29.1 cm at the forearm (right, 25.2 cm).

Wrist and fingers also had wider girth on the left compared with the right, but were equal in length. The swelling of the left upper limb was painless, non-pitting, with no colour change or abnormal sweating. Temperature of the limb was normal. Apart from evidence of a recent scald injury, no other superficial skin changes, vascular prominences or nerve thickening were observed.

Loose bodies were present in the left shoulder joint, as evidenced by palpable crepitus. Left shoulder abduction was restricted beyond 90°. No hum, bruit or machinery murmur could be heard on auscultation in any part of the affected limb. The right breast was slightly larger than the left.

Neurological examination revealed that pain, temperature and tactile sensation were diminished on the left side from C2 to T6 dermatomes anteriorly, and from C2 to T4 dermatomes posteriorly.

Position and vibration senses were well preserved in these segments. All modalities of sensation in other parts of the body were normal. The patient had normal speech and higher functions. The hairline was at C2 level and the height-neck ratio was 11.67 (reference range, < 13). No meningism or cranial or spinal deformities could be identified. Examination of the cranial nerves gave results within normal limits.

Apart from a mild weakness of the small muscles of the left upper limb, as evidenced by a positive card test and Grade 4/5 power in the left supraspinatus and deltoid muscles, the motor system appeared normal. Deep tendon reflexes in the left upper limb were present but grossly diminished. Plantar reflexes were bilaterally flexor, with preserved abdominal reflexes. The patient had normal coordination, stance and gait, with no evidence of nystagmus or Horner syndrome.

Investigations: Complete blood count and routine biochemistry tests gave normal results. Fasting blood glucose level was 5.4 mmol/L (reference range [RR], < 6.1 mmol/L), and results of oral glucose tolerance were in the reference range. A serum VDRL (Venereal Disease Research Laboratory) test was negative.

An x-ray of the left hand showed soft tissue swelling and a slight increase in the size of the metacarpals and proximal phalanges compared with the right (Box, B). X-ray of the left shoulder joint showed loss of normal architecture, multiple bony fragments and loose bodies (Box, C). Skeletal imaging showed no abnormalities of the skull, spine or other limbs. No abnormalities were seen in the affected limb on arteriovenous doppler examination, computed tomography, magnetic resonance imaging, angiography, lymphangiography and skin biopsy.

Magnetic resonance imaging of the brain and spine showed a fairly large (12 mm) cerebellar tonsil dipping in the posterior portion of the foramen magnum, with a large syrinx involving the whole of the cervical and thoracic spine to level T8 (Box, D).

Management: The patient was advised to undergo posterior fossa decompression, but was unwilling to have surgical intervention and is currently receiving regular follow-up. ♦

A syrinx is a cavity in the spinal cord (syringomyelia) or brain stem (syringobulbia).¹ Syringomyelia is a chronic progressive degenerative or developmental disorder of the spinal cord, characterised clinically by brachial amyotrophy, cutaneous analgesia and thermoanaesthesia, with sparing of tactile, joint, position and vibration senses, and loss of deep tendon reflexes. Frequently, there are associated developmental abnormalities of the vertebral column, base of the skull, and particularly of the cerebellum and brain stem. About 90% of patients with syringomyelia have type I Chiari malformation (downward displacement of cerebellar tonsils).

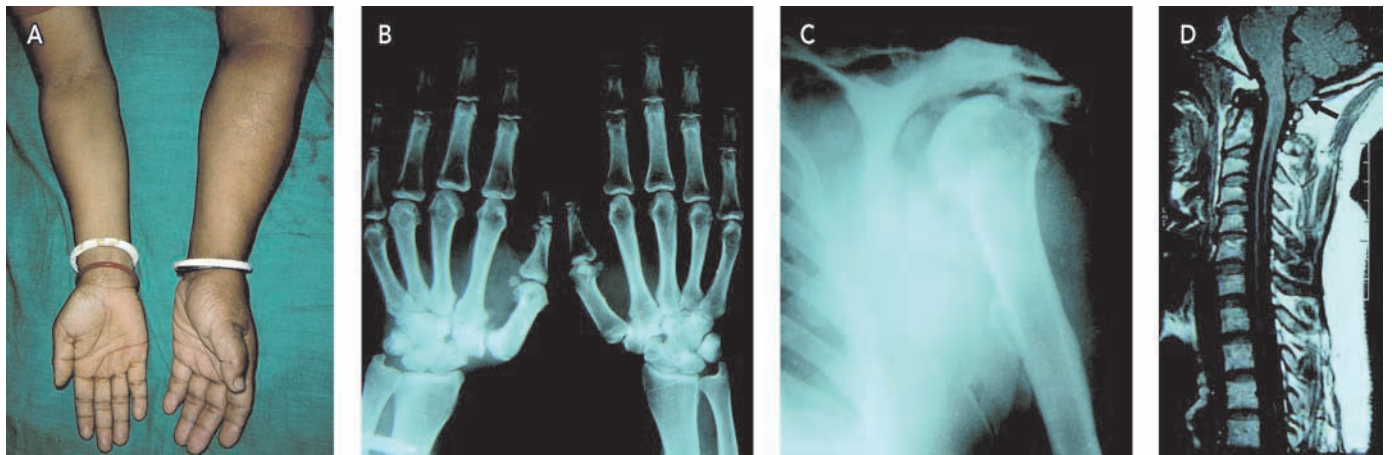
Patients with syringomyelia typically present with lower motor neurone signs at the level of the lesion, dissociated sensory loss in a cape or hemi-cape distribution on the arms or upper trunk, and spinal long tract dysfunction below the level of the lesion. Few patients show the complete picture, and the

clinical features vary with the size, location and shape of the cavity, and the associated neurological condition.¹ Common early signs are wasting and weakness of the small muscles of the hand, or loss of feeling in the hand and resulting injuries. Surprisingly, our patient — despite a relatively large syrinx — had no lower motor neurone signs in the upper limb, apart from diminished deep tendon reflexes and mild weakness, and no spinal long tract signs.

Syringomyelia can also cause “neuropathic joint”, most commonly in the shoulder, elbow or wrist. The affected joint is often enlarged but painless, and movements evoke loud crepitus,² as in our patient.

While syringomyelia is more commonly associated with limb atrophy, hypertrophy can occur, involving bones, muscles and other tissues in one limb, one half of the body or even the tongue.² Trophic changes in the skin can include cyanosis,

A patient with syringomyelia



A: Hypertrophy of the left upper limb.

B: X-ray of the hands showing bony and soft tissue enlargement on the left side.

C: X-ray of the left shoulder showing gross disorganisation with multiple bony fragments.

D: Magnetic resonance imaging of the cervical spine (T1-weighted) showing a large syrinx with type I Chiari malformation (arrow). ◆

hyperkeratosis and thickening of the subcutaneous tissue, leading to a swelling of the fingers described as “la main succulente”.² Other findings associated with syringomyelia include increased ratio of arm to body length, size differences between the breasts, and curved fingers,² (the latter two features documented in our patient).

An association with hypertrophy of the limbs, hands or feet is also recognised in syringomyelia,³ and has been reported in four patients from Japan, including three of a series of 26,^{4,5} and in two patients from India. However, the latter two, in contrast to our patient, had prominent neurological features.^{6,7} Localised hypertrophy of the hand is also seen in syringomyelia: a radiological study of hand bones in four patients with syringomyelic chiromegaly found enlarged hand bones on the affected side.⁸ This feature was also evident in our patient.

A range of mechanisms have been postulated to explain the segmental hypertrophy in syringomyelia, including stimulation of the sympathetic nervous system, causing defective circulation and oedema.⁹ An in-vitro study of muscle cells from patients with syringomyelia and muscle hypertrophy found the cells had accelerated growth in the presence of patient serum, suggesting involvement of serum factors and molecules released in response to neural lesions.¹⁰

To our knowledge, there are few other documented cases of syringomyelia presenting with both neuropathic joint and limb hypertrophy. Our case highlights this rare clinical presentation of a common spinal cord anomaly. It reminds clinicians that, although conventional teaching commonly associates syringomyelia with limb atrophy, it can be associated with limb hypertrophy.

Hence, syringomyelia should be considered as a differential diagnosis in all cases of limb enlargement. Moreover, this case further emphasises the importance of considering a spinal cord anomaly as the aetiology of upper limb neuropathic joint.

Partha P Chakraborty,* Dipanjan Bandyopadhyay†
Sanjay K Mandal,† Subhasis Roy Chowdhury*
Ramtanu Banerjee,† Shounak Majumdar*
Rana Bhattacharji‡

* Postgraduate Trainee, † Assistant Professor

‡ Resident Medical Officer and Clinical Tutor, Department of Medicine
Medical College, 88 College Street, Kolkata, West Bengal 700073, India
docparthapc@yahoo.co.in

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