Unusual association of diseases/symptoms

Syringomyelia, limb hypertrophy and sympathetic overactivity: a rare association

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Correspondence to Dr Arunansu Talukdar, arka_talukdar@yahoo.com**Summary**

A 32-year-old man presented with uniform enlargement of right upper limb for 6 years. Examination revealed painful disorganised elbow joint along with sensory impairment in the affected limb with weakness of small muscles. The patient was given empirical antileprotic therapy from outside without any benefit. Ultrasonography showed pan-hypertrophic nature of local tissues. Although axonal type of sensorimotor neuropathy involving right ulnar and median nerve was detected in nerve conduction velocity study, biopsy of the same failed to confirm any axonal degeneration or evidence of leprosy. Considering the nature of sensory abnormality MRI of cervical spine was done which revealed a syrinx extending from C3 till D2. Tests for vasomotor tone showed positive results on the affected limb. Syringomyelia can rarely give rise to pan-hypertrophy of limb due to sympathetic overactivity, which is infrequently reported in literatures and deficient in logical grounds.

BACKGROUND

Hypertrophy of a single limb is a rare experience and when it combines with sensory impairment and painful Charcot's elbow joint it becomes a diagnostic challenge and often misleads clinicians to an overzealous diagnosis of leprosy, considering the endemicity of the disease, such as in our case. Moreover the classical atrophic nature of syringomyelia blinds clinicians to see its uncommon hypertrophic variety. Incidentally, limb hypertrophy as a consequence of syringomyelia is least to come in mind and the previously published reports are driven in the preconceived notion of sympathetic stimulation. The authors here also tried to objectify the presence of sympathetic over-activity by means of various advanced methodologies.

CASE PRESENTATION

A 32-year-old non-diabetic, immunocompetent right-handed male electrician presented with painless, progressive, non-pitting uniform enlargement of right upper limb (figure 1A,B) over past 6 years without any sensory complaint, weakness, raised temperature, sweating abnormalities or loss of hair in the affected limb. The patient was clinically diagnosed as pure neuritic leprosy 3 years back and treated with rifampicin, dapsone and clofazimine for 6 months without any improvement. The patient had a road traffic accident (RTA) 8 years back which predominantly injured his lower limbs and was recovered uneventfully. Family history was insignificant and he underwent no surgery or radiation exposure earlier. Vitals were stable and no clinically palpable lymph nodes were found. Local examination of right upper limb revealed a gun-stock like deformity of elbow joint with associated pain and restriction on movement but the patient repeatedly denied any local injury. Ulnar and radial cutaneous nerves were thickened and non-tender with no thrills or bruits to suggest any vascular malformation. There was associated skin thickening. Right upper limb measured 28.5 cm at mid arm

(left, 25 cm), 29 cm at elbow (left, 24 cm) and 26 cm at forearm (left, 20 cm) with hairline at C2 level and normal height- neck ratio. Motor examination revealed normal tone but small muscle weakness of right hand with muscles around shoulder and elbow being 5/5 (MRC grading) power and biceps jerk on the right upper limb was diminished. Sensory examination was suggestive of dissociative anaesthesia. Light touch was spared in comparison with pain or temperature sensation. Other components of neurological examination as well as other systemic examinations were unremarkable.

INVESTIGATIONS

His routine haematological and biochemical profiles offered no clue for diagnosis. Ultrasonography of right upper limb pointed marked thickening of skin, subcutaneous tissue, muscles and nerves (ulnar, median and radial cutaneous nerve) with no features of synovitis or joint effusion. Doppler study of right upper limb excluded presence of any vascular tumour or malformations. X-ray of right elbow joint (figure 2) revealed increased bone density, destruction, loose bodies and disorganisation suggestive of Charcot's joint. Nerve conduction velocity study of both upper limbs suggested axonal type sensorimotor neuropathy involving right ulnar and median nerve. Electromyography (EMG) of upper limb revealed denervation pattern of the ulnar innervated muscles. Considering the ulnar nerve thickening, sensory deficit and Charcot's joint pure neuritic type of leprosy was a possibility. We went for biopsy from right-sided radial cutaneous nerve and sent for histopathological examination. Lymphoedema, being the commonest to cause unilateral limb enlargement and when long-standing can produce thickening of skin and subcutaneous tissue, a search was made for microfilaria in the peripheral blood and parasite-specific ELISA study was done considering high endemicity of filariasis in India, but both of which were negative. Radio-nuclear lymphoscintigraphy was

Figure 1 Photograph showing uniform swelling of right upper limb.

also normal negating a possibility of lymphoedema tarda. Presence of Charcot's elbow joint prompted us for an MRI cervical spine (figure 3) which revealed a hyperintense intramedullary signal change extending from C3 till D2 suggestive of syrinx with mild left-sided scoliotic tilt and normal CV junction and brain. In the meantime result of nerve biopsy came which showed no active myelin or axonal degeneration, due to skin thickening and also there was no features to suggest Hansen's disease and no deposits on Congo red stain or Oil red O stain.

DIFFERENTIAL DIAGNOSIS

- ▶ Filariasis
- ▶ Pure neuritic leprosy
- ▶ Syringomyelia
- ▶ Vascular tumour/malformation
- ▶ Amyloidosis
- ▶ Tangier's disease.

TREATMENT

Neurosurgical opinion was taken but the patient refused to undergo posterior decompression of syrinx considering the uncertainty to revert the hypertrophy of the upper limb. Contrary to common belief, our orthopedic colleagues encouraged to use elbow joint instead of immobilisation which is essential for reducing articular damage while retaining an optimally functioning joint. Decision against replacement of elbow joint was taken considering loss of pain sensation and reflexes, presence of osteopenic bone and weakness of surrounding bone and ligaments. Plastic surgeons explained the existence of subclinical ulnar neuropathy due to distortion of cubital tunnel as a consequence of deformity of elbow joint and to prevent further progression of the problem, a subcutaneous ulnar nerve transposition was performed without complication.

OUTCOME AND FOLLOW-UP

At his latest follow-up examination, 6 months after the surgery, the patient was continuing physical therapy and had a functional, pain-free elbow. Although there was hypertrophy of right upper limb with varus deformity of right elbow joint and bone fragments creating audible

Figure 2 Skiagram of right elbow joint antero-posterior view showing density (sclerosis involving the medial aspect of Humerus), debris (loose bodies), disorganisation, dislocation and destruction suggestive of Charcot's joint.

crepitation, a repeat EMG revealed normal study of ulnar innervated muscles on the right side.

DISCUSSION

Syringomyelia, which literally means 'tube within marrow' was first coined by Ollivier d'Angers in 1827,¹ is a degenerative disease characterised by abnormal fluid filled cavity within spinal cord with annual incidence of 9 per 100 000 and more among young males.² Half of the patients of syringomyelia have Chiari malformations³ and although the idiopathic variety is most common, important causes include trauma, tumour, arachnoiditis and tuberculosis of spinal cord.⁴ Asymmetric cape-like distribution of dissociative anesthesia, areflexic weakness, wasting of upper limb along with neuropathic joints are the usual manifestations of syringomyelia which often results in facial numbness

Figure 3 MRI of cervical spine (sagittal section) shows hyperintense intramedullary signal changes extending from C3 till D2 suggestive of syrinx with no Chiari malformation.

and Horner's syndrome and rarely produces bulbar features (syringobulbia) but pan-hypertrophy of a single limb in syringomyelia is a very under-reported presentation and the mechanism is claimed to be stimulation of preganglionic sympathetic neurons.^{5–7} Sympathetic overdrive resulting in hyperemia and increased osteoclastic activity leading to bone resorption is well known to produce neuropathic joints but to involve elbow joint is rarely reported^{8–10} and contrary to the classical belief, is often painful in the early hypertrophic phase.¹¹

To substantiate the presence of increased sympathetic tone, we performed a simple cold pressor test,¹² where the affected right hand (blood pressure (BP): right 134/88, left 130/80) was immersed in ice-cold water and measurement of BP after 5 min revealed a sharp rise on the right side (right 160/120, left 130/80). The same was conducted on the opposite (BP: right 128/100, left 110/90) and there was uniform rise of BP in both upper limbs (right 150/112, left 130/90), which corroborates with an increased vasomotor tone on right side. A rarely employed bedside method in present time, known as Minor's test¹² revealed area of hyperhidrosis involving the entire right upper limb which feeds the possibility of increased sympathetic activity.

Next, we performed a sympathetic skin response (SSR) study¹³ using right ulnar and median nerve stimulating digit II and IV which showed increase amplitude of SSR on right side compared to left. Presence of orthostatic tachycardia in the absence of orthostatic hypotension also gives clue to autonomic dysfunction.¹⁴ Recent technological advances including Quantitative Sudomotor Axon Reflex Test (QSART), Silastic Sweat Imprint, Thermoregulatory Sweat Test (TST) and Quantitative Direct and Indirect Axon Reflex Test (QDIRT)^{15–17} if available, would be more specific to judge the nature of sympathetic stimulation.

Albeit, the proper aetiology of syrinx in our case is difficult to ascertain, a subclinical spinal trauma as a result of RTA 8 years back may produce this non-communicating extra-canalicular syrinx¹⁸ which is strategically placed around right intermediolateral column to produce autonomic features only resulting in the hypertrophy of right upper limb.¹⁹ The patient was empirically given antileprotic therapy based on the ulnar neuropathy and Charcot's elbow joint but on recognising the disease as an atypical syringomyelia, subcutaneous transposition of right ulnar nerve prevented progress of ulnar neuropathy²⁰ but the right upper limb hypertrophy and Charcot's elbow joint on the right side was unabated till the recent visit.

Learning points

- ▶ It is important to suspect and recognise syringomyelia in a country where Hansen's disease is highly endemic in order to avoid overzealous empirical treatment.
- ▶ Syringomyelia, however uncommonly, can present as chiromegaly and rarely can involve and stimulate the sympathetic neurons of intermediolateral column in absence of dissociative anesthesia.
- ▶ Syringomyelia presenting as pan-hypertrophy should be tested for sympathetic overactivity by means of advanced methodologies (SSR, QSART, TST and QDIRT).

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Competing interests None.

Patient consent Obtained.

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