CASE REPORT

Sprengel's deformity: association with musculoskeletal dysfunctions and tethered cord syndrome

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SUMMARY

Sprengel's deformity is a rare and complex congenital deformity of the shoulder girdle. The deformity commonly occurs sporadically, though in combination with other congenital anomalies, such as congenital scoliosis, fusion of cervical vertebrae, and conditions like Klippel-Feil syndrome may coexist. We report a case of a 14-year-old girl with bilateral Sprengel's deformity presenting with a progressive bilateral lower limb weakness and gait abnormality. Radiological investigations demonstrated multiple musculoskeletal abnormalities on x-ray and lumbar spina bifida occulta causing tethering of the cord on MRI. The patient consulted neurosurgeons and orthopaedic surgeons, who recommended no operative intervention and conservative management till the end of growth spurt. Therefore, we prescribed a home-based exercise regimen to strengthen the periscapular and intrinsic foot muscles. Although rare, Sprengel's deformity can be associated with other musculoskeletal abnormalities including lumbar spina bifida and comprehensive neurological examination should not be ignored as it is much more than a cosmetic problem.

BACKGROUND

Sprengel's deformity is a rare and complex congenital deformity of the shoulder girdle. The disorder occurs due to inadequate caudal movement of scapula during development resulting in an abnormally high placed scapula.^{1–3} The scapular involvement may be unilateral or bilateral and associated changes in axial rotation, shape, and size are normal.⁴ More women are affected than men with a 3:1 ratio.⁵ The deformity commonly occurs sporadically, though in combination with other congenital anomalies, such as congenital scoliosis, fusion of cervical vertebrae, and conditions like Klippel-Feil syndrome, kidney abnormalities and cleft palate may coexist.^{5–6} It is also frequently associated with other bone and soft tissue anomalies,^{5–7} but to the best of our knowledge, its association with neurological abnormalities leading to lower limb deformities has not been reported from the Indian subcontinent.

CASE PRESENTATION

A 14-year-old girl visited our outpatient clinic with her father and presented with chief complaints of limitations of bilateral shoulder movements in abduction and overhead elevation, bilateral shoulder asymmetry, limitations of neck movements,

neck deformity and head deviation to the right side, first noticed by her parents when she was about 3 years of age. The patient had fresh complaints of weakness of bilateral lower limbs with increase in medial longitudinal arch height and elevation of the medial foot border during stance phase of walking since 18 months which were insidious in onset, progressive in nature, not associated with back pain, radiation to lower limbs or bladder and bowel deficits. Familial history for any congenital disorder was unremarkable. On clinical examination, both shoulders were elevated than the normal level in the standing position with right lying higher than left (figure 1). No clear torticollis was noted but head was deviated to the right, shortened clavicle was present on the left side with normal bilateral acromian processes. There was a presence of mildly exaggerated thoracic kyphosis with a cervico dorsal 'S'-shaped scoliotic curve having dorsal convexity to the right side and compensatory cervical convexity to the left side. Spirometric measurements could not be performed because the patient did not cooperate; therefore we evaluated thoracic expansion by means of chest excursion measurements. The patient's chest excursion measurements showed a range of 1 cm from complete expiration to maximal inspiration at the axillary, epigastric and subcostal landmarks, respectively, revealing insufficient excursion. Restriction of the bilateral shoulder range of motion in forward flexion (active–90° and passive –130°) and abduction (active–90° and passive –140°) was seen with a normal range of motion in other movements at shoulders and power of all the

Figure 1 Image showing bilateral Sprengel's deformity with elevated shoulders (rt>lt) and hypoplastic left clavicles.

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Figure 2 Chest x-ray showing bilateral mild high riding scapulas (black arrows), hemivertebrae at T3 (blue arrow), bifida anterior right second and third ribs (white arrow) and a hypoplastic left first thoracic rib (red arrow).

muscle groups at shoulders was 4 bilaterally on a Medical Research Council scale. On neurological examination of lower limbs, both feet were in attitude of pes cavo varus with diminished bilateral ankle reflexes and weakness of intrinsic muscles of both feet (Lumbricals and interossei). Rest of the neurological examination was unremarkable.

INVESTIGATIONS

Diagnostic measures performed included chest x-ray, shoulder x-rays, spine x-rays, abdominal sonography and MRI LS spine. Radiological investigations demonstrated an obvious skeletal deformity with chest x-ray (figure 2) showing mild high riding bilateral scapulas, hemivertebrae at T3, bifid anterior right second and third ribs, and hypoplastic left first thoracic rib. x-Ray of lumbosacral spine showed spina bifida occulta at L5 level. Findings of lumbosacral spine x-ray were confirmed on MRI LS spine (figure 3) which showed abnormally a low-lying spinal cord with conus medullaris at L4 level, thickened terminal filum and a spur from spina bifida occulta at L5 causing tethering of the spinal cord. Electrodiagnostic studies conducted for the bilateral lower limbs showed reduced amplitude of sural sensory nerve on the right side with normal bilateral tibial and common peroneal nerves, and F wave and H reflexes. Electromyography of intrinsic foot muscles and trunk muscles

Figure 3 MRI LS spine (sagittal T1/II slice) showing conus medullaris at L4 level (white arrow), thickened terminal filum, spina bifida occulta with spur (black arrow) causing traction of the spinal cord.

was not done due to patient's non-cooperation and those of L4, L5 and S1 myotomes revealed no significant abnormality.

TREATMENT

The patient consulted orthopaedic surgeons, who recommended no operative intervention for Sprengel's deformity. The patient was also referred to a neurosurgeon for opinion and was kept on watchful observation for worsening of symptoms and advised conservative management till the end of growth spurt. Therefore, we prescribed a home-based exercise regimen to maintain the range of motion at shoulders and to strengthen the periscapular and intrinsic feet muscles, deep breathing and stretching exercises for scoliosis.

OUTCOME AND FOLLOW-UP

At 6 months follow-up, the neurological condition of the patient was the same and did not show further worsening.

DISCUSSION

The scope of abnormalities that were present in this case is different from what has been described by other authors. Although Klippel-Feil syndrome is commonly associated and usually responsible for the limitation of neck and shoulder motion in these patients, we did not find any vertebral fusion in our patient and dysplastic neck muscles seemed to be responsible for this problem. Cervical spina bifida⁸ has been associated with Sprengel's deformity but spina bifida causing tethered cord syndrome is rare and this combination of congenital high scapula, multiple rib and spine deformities including lumbar spina bifida with tethered cord syndrome has not been reported before, to the best of our knowledge.

The surgical management of the tethered cord remains controversial. Although treatment in a patient with tethered cord syndrome (TCS) and evidence of neurological decline are straightforward, the natural history of TCS suggests a progressive decline when managed conservatively. Thus, in a symptomatic patient, any change in routine urodynamic or manual motor testing should be closely monitored. With surgery, the initial approach is aimed at dealing with the local pathology. Over time, however, recurrent tethering in an adolescent due to growth spurt can produce symptoms. In the adult population, the rate of retethering has been quoted to be as high as 25%.⁹ Lee *et al*¹⁰ found that motor weakness will stabilise or improve in only 27 and 64% of patients, respectively. Sensory deficits remained unchanged in 50% of patients. Urological abnormalities improved in 50% of patients undergoing untethering and remained stable in 45%. The patient was thus planned to be managed conservatively till the end of growth spurt with watchful monitoring on change of motor function or bladder urodynamics.

Although this congenital abnormality is rare and those with minor cosmetic deformity or functional impairment may not need surgery comprehensive neurological examination should not be ignored and we must find these cases and solve their problem either as a functional or a cosmetic aspect.

Learning points

- ▶ Association of Sprengel's deformity with other multiple neural tube defects (spina bifida, Klippel-Feil syndrome, diastematomyelia) must always be kept in mind.
- ▶ Comprehensive neurological examination should be carried out in a patient with Sprengel's deformity.

Contributors NM collected and interpreted the patient data and was involved in preparation of the manuscript. RM obtained and interpreted radiological studies. RM and SC reviewed the literature. MA approved the final draft of the manuscript. All authors read and approved the final draft of the manuscript.

Competing interests None.

Patient consent Obtained.

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REFERENCES

- 1 Lungu SG. Undescended scapula. *Med J Zambia* 2009;36:136–9.
- 2 Crenshaw AH. *Campbell's operative orthopedics*. 5th edn. St Louis, MO: CV Mosby Company, 1971.
- 3 Leibovic SJ, Ehrlich MG, Zaleske DJ. Sprengel deformity. *J Bone Joint Surg* 1990;72A:192–7.
- 4 Blatt ML, Scandiffio M. Sprengel's deformity. Failure of the scapula to descend. *Can Med Assoc J* 1934;31:596–8.
- 5 Cavendish ME. Congenital elevation of the scapula. *J Bone Joint Surg Br* 1972;54B:395–408.
- 6 Carson WG, Lovell WW, Whitesides TE. Congenital elevation of the scapula. *Bone Joint Surg Am* 1981;63A:1199–207.
- 7 Grogan DP, Stanley EA, Bobechko WP. The congenital undescended scapula. Surgical correction by the Woodward procedure. *J Bone Joint Surg Br* 1983;65:598–605.
- 8 Thacker MM. Sprengel deformity. *Medscape Reference*, 17 Feb 2012. <http://www.emedicine.com/Orthoped/topic445.htm> (accessed 10 Oct 2012).
- 9 Filler AG, Britton JA, Uttley D, *et al*. Adult postrepair myelomeningocele and tethered cord syndrome: good surgical outcome after abrupt neurological decline. *Br J Neurosurg* 1995;9:659–66.
- 10 Lee GY, Paradiso G, Tator CH, *et al*. Surgical management of tethered cord syndrome in adults: indications, techniques, and long-term outcomes in 60 patients. *J Neurosurg Spine* 2006;4:123–31.

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