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GRAND ROUNDS

Congenital kypho-scoliosis: a case of thoracic insufficiency syndrome and the limitations of treatment

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Abstract

Introduction Congenital spinal vertebral anomalies may present with deformity resulting in congenital scoliosis and kyphosis. This leads to abnormal spinal growth. The latter when combined with associated rib fusions may impair normal thoracic cage development and resultant pulmonary hypoplasia. Most congenital scoliosis can be detected in utero by ultrasound scan or recognized in the neonatal period, but a few spinal defects can remain undetected.
Materials and Methods In this Grand Round, we present the case of a 7-year-old girl with a severe scoliosis and thoracic insufficiency syndrome (TIS). 3D CT reconstruction imaging demonstrated a mixed picture of fusion and segmentation abnormalities. A marked kyphoscoliosis was demonstrated at the thoraco-lumbar junction. Via a left thoracotomy, anterior excision of intervertebral discs was performed together with, interbody fusion, and in situ stabilisation of the kyphosis with double allograft (femur) strut grafts.
Conclusions This article highlights the features of congenital kypho-scoliosis and TIS. The difficulties of treating kyphosis when combined with TIS are discussed together with the limitations of current surgical techniques.

Keyword Thoracic insufficiency syndrome

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Case presentation

A 7-year-old girl presented to the scoliosis clinic with a severe deformity. She was born by caesarean section and the antenatal ultrasound scan at 19 weeks gestation showed multiple congenital vertebral anomalies. She suffered with repeated chest infections from birth. There was a strong family history of scoliosis with her mother, three uncles and one aunt all affected.

On examination she weighed 15 kg, she displayed a severe right-sided kypho-scoliosis with no excursion of the chest wall. On suspension, the kyphosis appeared very rigid (Fig. 1a–c). Oxygen saturations dropped to 85% after walking 40 yards. X-rays demonstrated multiple congenital rib abnormalities together with absent posterior elements 1–10 (Fig. 2a, b). 3D CT reconstruction imaging showed complex congenital anomalies with a combination of fusion and segmental failures. A marked kyphoscoliosis was demonstrated at the thoraco-lumbar junction, with extensive fused ribs and posterior element defects in the mid-thoracic spine. The manubrio–sternal complex was also considerably shortened (Fig. 2c). MRI of the whole spine displayed no cord tether, syrinx or other dysraphic abnormality.

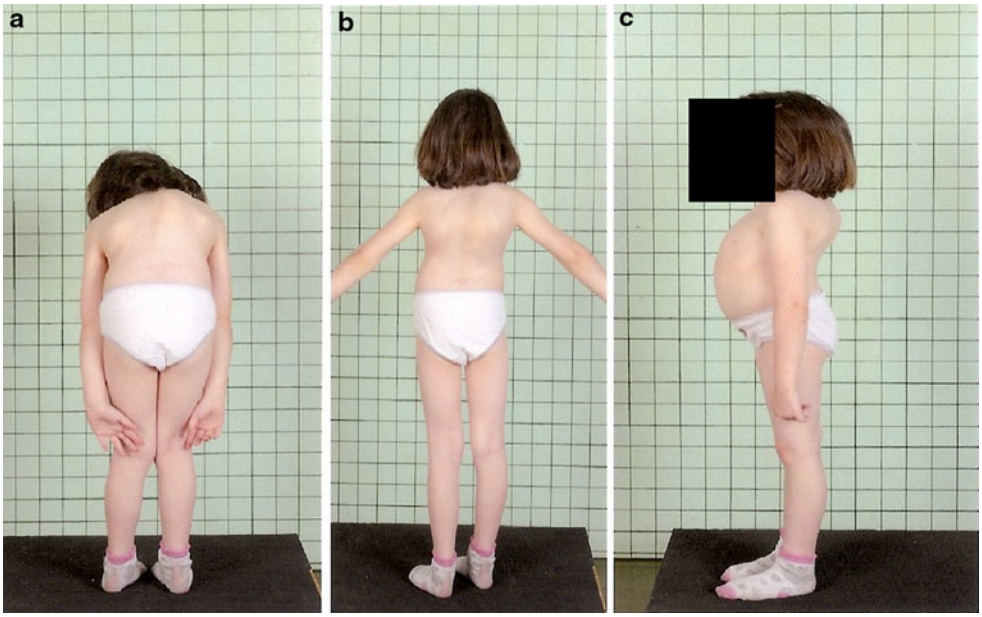

Fig. 1 Demonstrating the rigidity of the kyphosis

Historical perspective and diagnosis of congenital scoliosis

Congenital spinal vertebral anomalies can present as scoliosis or kyphosis or both. A key feature of congenital scoliosis is the presence of one or more abnormally formed vertebrae [1]. The prevalence of the vertebral anomalies is 0.5–1 per 1,000 live births [2].

Most congenital scoliosis is often recognized at birth, but a few spinal defects can remain undetected. In view of the strong family history, there is a 1 in 100 risk of a first degree relative having a single vertebral malformation and a risk of 1 in 10 for multiple vertebral anomalies in either siblings or children of a patient [3]. Patients with congenital scoliosis frequently have associated non-spinal anomalies. Presence of vertebral anomalies leads to a disruption in the longitudinal growth of the spine. Restricted pulmonary function is a major problem in those patients with curves greater than 90°. There may be evidence of hypoplastic lung development [4].

Thoracic insufficiency syndrome (TIS) is the inability of the thorax to support normal respiration or lung growth [5]. The syndrome demonstrates two important deficiencies. First, the thorax is unable to support normal respiration with compensation by an increased respiration rate and inability to maintain adequate levels of oxygen saturation. Secondly, the thorax is unable to support lung growth. A deformed thorax that may be adequate in early life may not be able to support normal lung development in the future. Campbell et al. [5] developed a volume-depletion deformity classification system. Type 1 and Type 2 deformities are asymmetric, with unilateral volume-depletion deformities. These require unilateral surgical expansion to

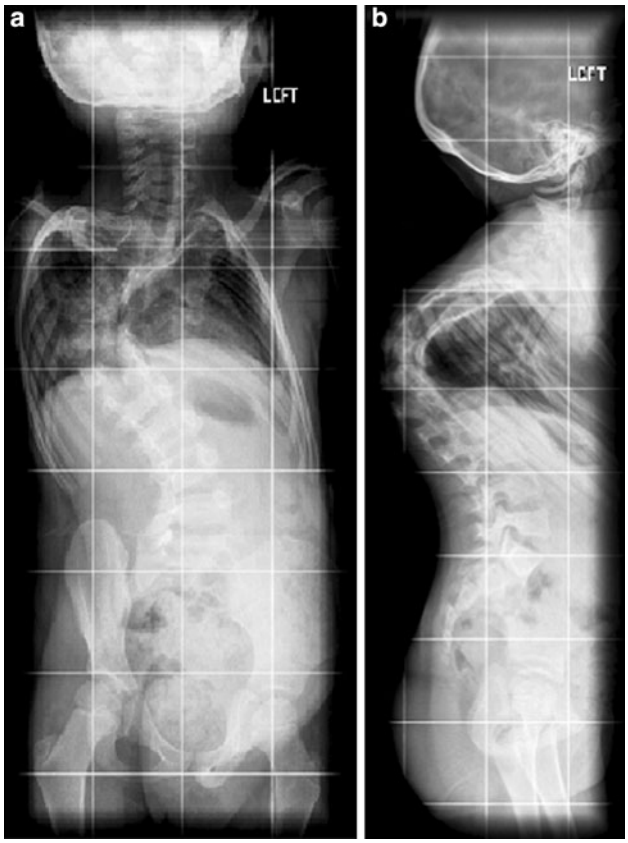

Fig. 2 X-rays demonstrated multiple congenital rib abnormalities together with absent posterior elements

restore thoracic volume and symmetry. Type 3 deformities have a global thoracic deficit which can either be a symmetrical longitudinal constriction such as Jarcho–Levin syndrome or a symmetrical transverse constriction such as in Jeune asphyxiating thoracic dystrophy.

Campbell et al. treated more than 150 children with a type of expansion thoracoplasty. For severe progressive congenital thoracic scoliosis, an opening wedge thoracostomy was developed which directly treats the segmental hypoplasia of the hemithorax while correcting the scoliosis. The correction is stabilised with a chest wall device; a vertical, expandable prosthetic titanium rib (VEPTR). Correction during growth is maintained by serial expansion of the VEPTR by limited incisions. The aim of this procedure is to restore lost thoracic volume by straightening the angulated thorax. *Periodic prosthetic lengthening preserves thoracic spine growth so that the thorax can gain additional volume through increases in spinal height.*

Opening wedge thoracoplasty with VEPTR has been shown to increase lung volume in congenital scoliosis children with fused ribs by CT scan [6]. The volume of the convex lung increased by 60% with an average density decrease of 6%. Decrease in density improves the aeration of the lung. There are now several articles highlighting the benefit of VEPTR and lung expansion for children with severe congenital scoliosis. The main problem arises is that the majority of the data refers to scoliosis and that the device is kyphogenic. Ramirez et al. [7] also noticed a thoracic kyphosis maintained at anatomically normal values with sufficient space available for lung expansion. Therefore, there is poor data representation related to a kyphoscoliosis as in this Grand Round presentation.

X-rays remain the primary imaging study in the diagnosis of congenital scoliosis. The magnitude of the curve, progression and assessment of potential growth are all assessed utilising this imaging modality. Three-dimensional reconstruction CT displays the morphology of the spine along with any other abnormalities present especially in patients with TIS [8] (Fig. 3a, b). MRI scanning is essential pre-operatively in order to determine any cord abnormalities [9].

Rationale for treatment and evidence-based literature

There is a general acceptance that an untreated congenital scoliosis can be potentially lethal in terms of TIS, neurological complications and there has been a reported death from cor pulmonale [10].

In this Grand Round case, several factors had to be considered when planning surgical treatment. At the time of presentation, the patient was 6 years old and alveolar development was more likely than not completed—it was decided that surgery would be best at this stage in order to preserve lung function, maintain rib mobility and chest excursion by improving or preventing progressive kyphosis. In addition, there was concern that progressive kyphosis may lead to neurological deterioration [11].

Several studies have produced good results showing the efficacy of VEPTR-based treatment strategy. One study

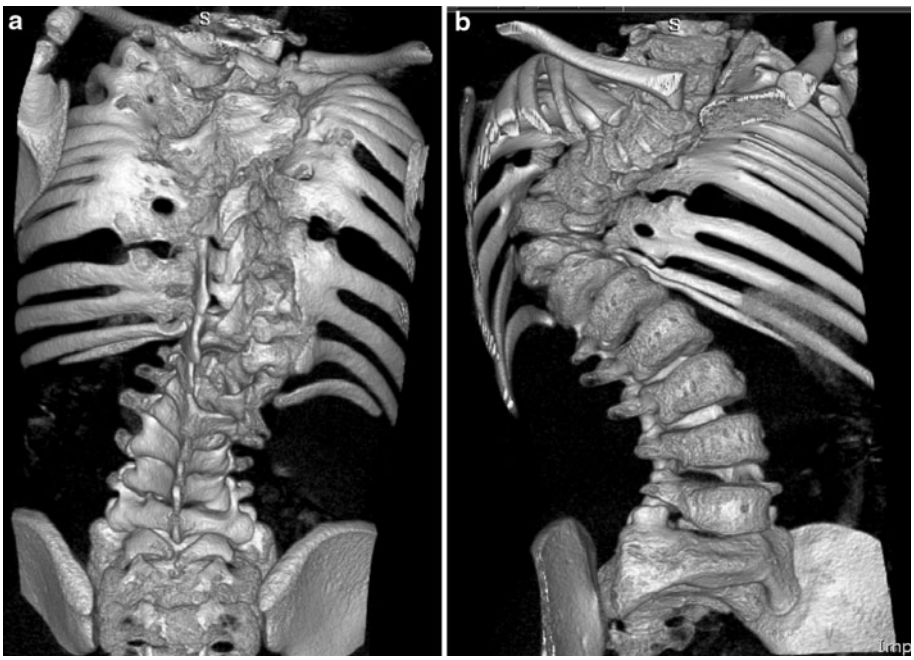

Fig. 3 Three- dimensional reconstruction CT displaying the morphology of the spine in our patient with thoracic insufficiency syndrome

does highlight a limitation of the VEPTR strategy. It may not be a correct treatment strategy where the vertebral segment may be too long for resection or where there is some flexibility in the anomalous segment [12]. In this case, the bilateral VEPTR devices (and adjuvant thoracostomies) would need to be placed sufficiently anteriorly to correct the kyphosis (assuming there was flexibility in the curve which was not the case) and anchorage to the anterior pelvis. This has not been previously described and it is questionable if the small pelvis would accommodate the implant. The soft tissue cover over the antero-lateral chest is also poor given the relative bulk of the VEPTR device.

There have been many distraction growing rod systems namely the spinal jack of Wenger and the original Harrington rod used without fusion. These techniques may be used in early onset scoliosis but there are no proven benefits from the rod system. Akbarnia et al. [13] have reported problems of implant loosening and rod dislodgement, which may be exacerbated by the patient's pre-existing kyphosis. There have also been no reports of improvement of pulmonary function and three reports have stated a decrease in normal vital capacity at the time of follow-up [14–16]. In this patient, such device would be kyphogenic and the extent and number of the fused ribs would in all probability prevented clinically useful distraction.

Correcting the kyphosis does have theoretical advantages in expanding chest volume; however, limitations similar to VEPTR strategies do arise. A Type 3 deformity with a global thoracic deficit requires multiple Ponte osteotomies, which require some anterior column flexibility which may not be possible in the presence of multiple vertebral fusions. Moreover, multiple anterior releases (or vertebral column resection—VCR) risks compromising the cord blood supply which in this case was most probably anomalous in keeping with the multiple posterior element defects. There has been a case report whereby segmental vessel ligation during anterior spinal surgery has caused paraplegia [17]. This report raises the question of neurological risk especially in complex congenital spinal deformities occurring mainly in the thoracic spine where vascular supply to the cord may be abnormal.

A posterior approach to hemi-vertebrae (HV) excision has reported correction results of 23°–36° with an average loss at follow-up of 3.7° [18, 19]. In a long-term study for single, fully segmented HV with single stage excision via posterior approach alone accounted for 54.3% scoliosis correction and 67.4% kyphosis correction [20]. Single stage anterior and posterior HV excision had mean postoperative correction of 67% [21]. The results of a lumbar HV resection and short-segment fusion through a lateral-posterior approach reported postoperative curve correction of 60.9% [22]. In a study of 10 patients with thoracic and

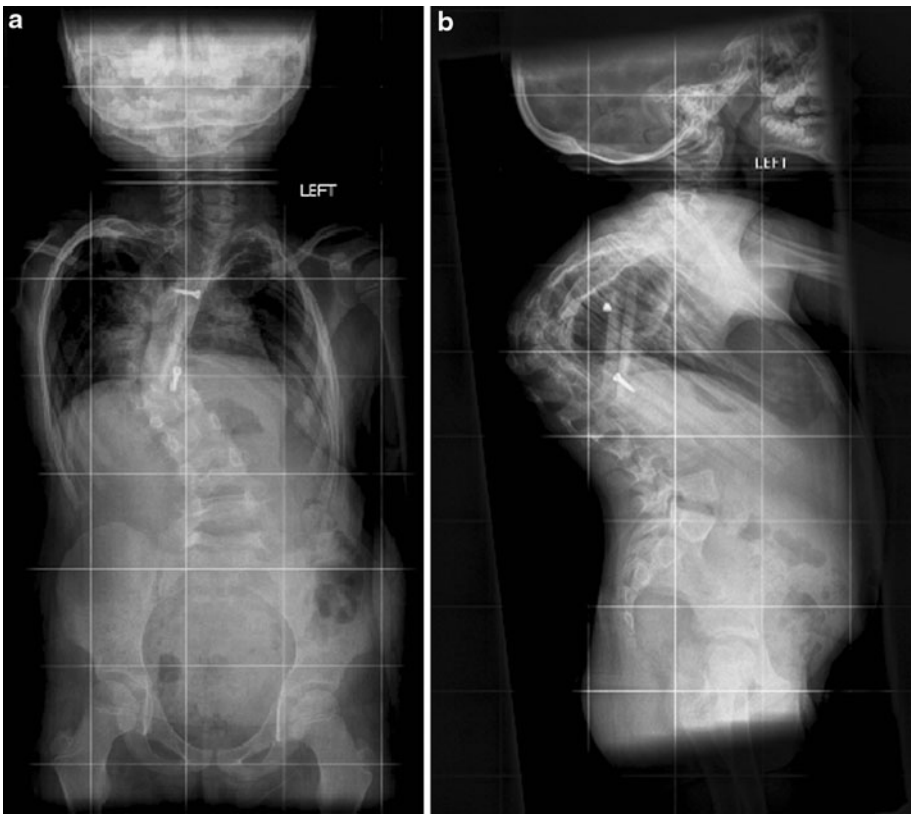

Fig. 4 Post-operative check X-rays showing satisfactory position of the fixation

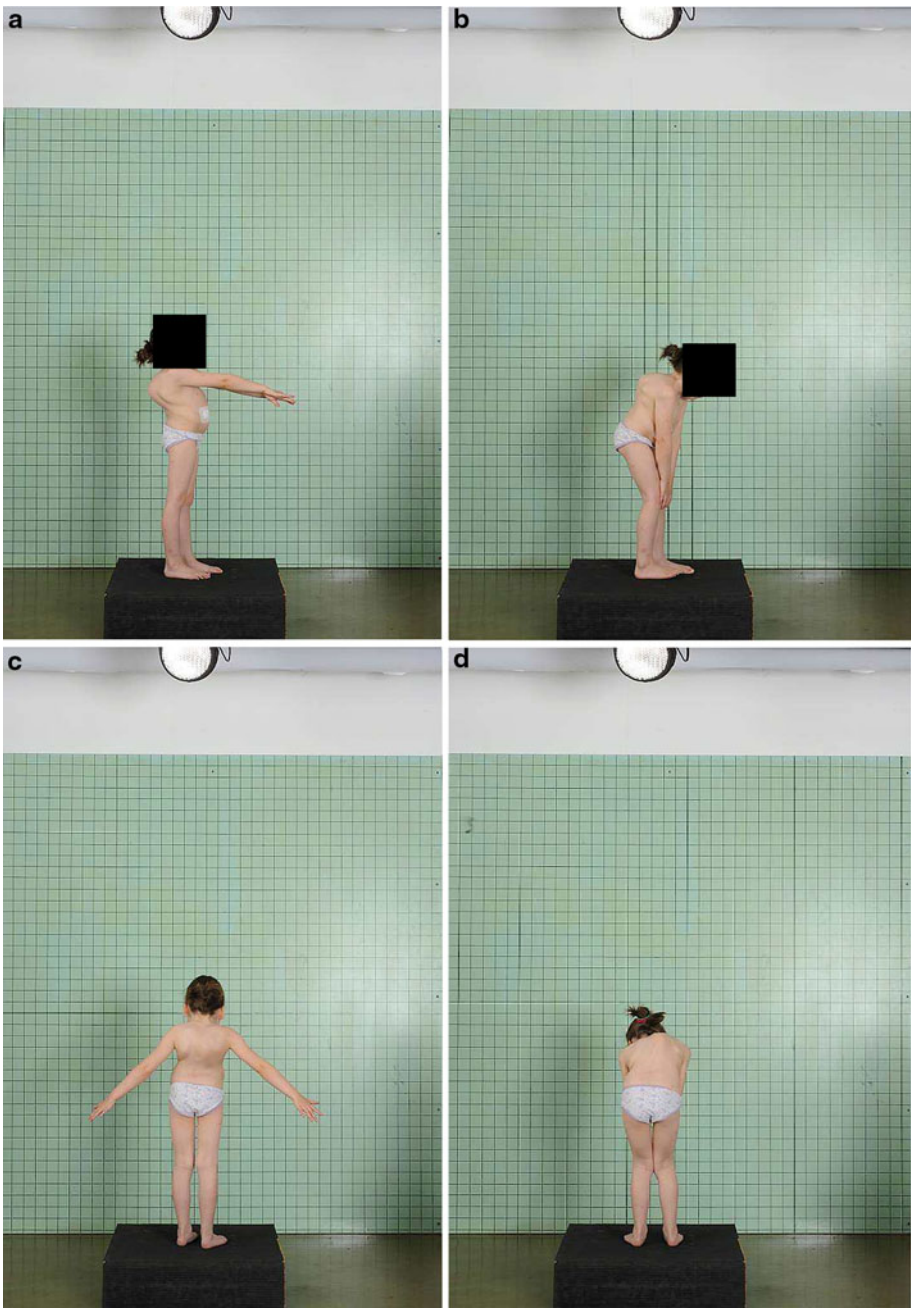

Fig. 5 Photographs one year on demonstrating the clinical progress

thoracolumbar HV excision, the mean curve improvement was 59% [23]. This approach was inappropriate in this patient as there were too many vertebral anomalies and the hazards of VCR have been highlighted above.

In conclusion, this case significant kypho-scoliosis challenged the usual paradigms for surgical treatment of congenital spinal deformity. Her pulmonary development was in all probability complete and the lung function (and possibly neurological status) was going to be adversely affected by progressive deformity. The VEPTR device has not been described in this scenario and pelvic fixation, adequate soft tissue cover and ability of thoracostomies to expand the chest volumes with such shortened manubrium was questionable. Growing rod systems were similarly rejected because of their kyphogenic nature and well-documented problem of loosening especially in the presence of kyphosis. Even more radical surgery to correct the kyphosis (possibly including VCR) was ruled out due to the potential threat to an anomalous blood supply. Therefore, the chosen approach was an anterior strut graft to prevent worsening kyphosis, accepting that the lung volumes would not improve but avoiding neurologic risk in some of the other techniques outlined above. Early fusion was advised to prevent long-term rigid deformities [24].

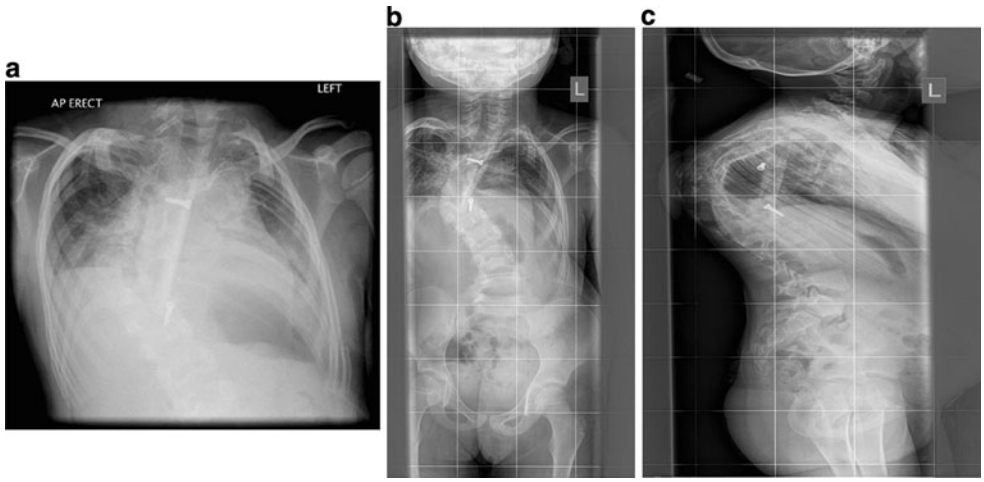

Fig. 6 X-rays one year on showing growth in the child as well as satisfactory fixation

Procedure

A left lateral thoracotomy with rib excision allowed a discectomy at L1/2. A femoral allograft was cut into strips. This was done through a left thoracotomy with anterior excision of intervertebral disc L1/2 and a double allograft (femur) strut graft.

The first strut was inserted and held proximally with an interference screw and the second graft was distally inserted into the disc space and body of L2 with interference screw. There was rapid post-op extubation and recovery. A chest drain was inserted and removed 2 days later. Check X-ray was satisfactory (Fig. 4a, b).

Outcome

This Grand Round case demonstrates the need for a thorough pre-operative clinical assessment especially when children are involved. The initial presentation of the child showed a mixed picture of fusion and segmentation of the spine along with respiratory problems. It was important to stage the procedure for this child in terms of dealing with the respiratory problems first. There are few cases in the literature, which highlight a case such as this Grand Round presentation. The non-fusion strategies are either difficult or there is insufficient data to inform the decision-making process. There is adequate evidence in the literature regarding anterior strut grafting for progressive kyphosis [25–27]. This approach is not widely described in congenital kyphosis with only a few reports mentioned [28, 29]. It is also important to note that some form of definitive treatment was required so as to prevent the progression of neurological deterioration in the future. This has been achieved with the respiratory function of the child clinically much improved 1 year on since the procedure in terms of activity (Fig. 5a–d). Before the procedure her FVC measured between 0.34 and 0.36 with a predicted value of 0.29. One year on after the procedure this has reduced to 0.31 with a predicted value of 0.34. This is in line with a reduction in FEV 1 from 0.34 to 0.25 over a 1 year period. It is important to state that the child is now able to play and run with ease one year post surgery compared to her saturations dropping after walking 40 yards a year previously (Fig. 6a–c). It can take up to 2–3 years for lung function tests to return to normal in the adolescent idiopathic scoliosis group hence these results may not be reliable at the present time. This, therefore, shows a clinical improvement and may also dispute the reliability of lung function tests after 1 year.

Conflict of interest None of the authors has any potential conflict of interest.

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