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CASE REPORT

Ten year follow-up of Jarcho–Levin syndrome with thoracic insufficiency treated by VEPTR and MCGR VEPTR hybrid

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Abstract
Purpose Jarcho–Levin syndrome patients commonly suffer from repeated respiratory infections and become ventilator dependent due to an insufficient thoracic volume. Multiple congenital vertebral anomalies are associated with this genetic disorder and results in stunted spine growth. The purpose of this case report is to present the results of a hybrid vertical expandable prosthetic titanium rib (VEPTR) and magnetically controlled growing rod (MCGR) construct for the management of a patient with Jarcho–Levin syndrome.
Methods A boy with Jarcho–Levin syndrome undergoes a hybrid VEPTR–MCGR construct to treat his thoracic insufficiency syndrome and spinal deformity.
Results The patient could wean off ventilator and had reduced chest complications with the construct. He was also able to achieve some spine length gain with the distraction device. However, there were limitations in prolonged treatment as no spine height was gained once he reached 12 years old. Once no further growth can be achieved, the MCGR led to progressive kyphosis. Nevertheless, at latest follow-up after the removal of all implants, the patient could maintain satisfactory correction of both thoracic and spine deformities.
Conclusions A hybrid construct is necessary for managing Jarcho–Levin syndrome as the VEPTR deals with the thoracic cage deformity while the MCGR deals with the spine deformity.

Keywords Magnetically controlled growing rod · Vertical expandable prosthetic titanium rib · Distraction · Spinal height · Jarcho–Levin syndrome

Introduction

Jarcho–Levin syndrome (JLS) is characterized by abnormal segmentation of the thoracic spine and irregular fusion of the ribs resulting in early-onset scoliosis (EOS) and restrictive lung disease [1]. This leads to thoracic insufficiency syndrome (TIS) [2, 3], resulting in frequent respiratory infections, and occasionally ventilator-dependency. The associated vertebral anomalies limit spinal growth, leading to a low percentile of expected height.

The vertical expandable prosthetic titanium rib (VEPTR) device allows serial expansion of the thoracic space for alveolar development and improves pulmonary function [4]. Although the safety and effectiveness of VEPTR have been well-reported, [2, 5, 6] increase in thoracic spine height during treatment is only between 24 and 67% of the expected value [6, 7]. Hence, additional treatment is required to promote spinal growth. Furthermore, rib to rib distraction without an increase in spine length can lead to increase in abdominal pressure and distention.

The magnetically controlled growing rod (MCGR) is a novel technology that uses an external remote control (ERC) to distract the spine non-invasively in the outpatient setting. We first reported the successful correction of spinal deformities using MCGR [8], and since then there have

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been several series documenting its safety and efficacy [8–14].

In this case report, we report the feasibility of a hybrid MCGR–VEPTR construct to improve the chest volume and enhance spinal growth in a case of JLS, and report the outcomes and challenges faced in this patient.

Case report

This was the first pregnancy from a non-consanguineous Southern Chinese family. The boy was born at full term by spontaneous vaginal delivery. The antenatal and family history were unremarkable. His mother was aged 22 and enjoyed good past health. The birth weight was 2.38 kg.

At birth, he had a short neck and trunk, and shortly after delivery he developed respiratory distress requiring ventilatory support. Radiographs revealed multiple hemi-vertebrae along the thoracic and lumbar spine, with symmetrical rib crowding and costo-vertebral fusion. He was diagnosed with JLS based on the constellation of clinical and radiographic features.

His pulmonary function deteriorated, requiring a tracheostomy at aged 6.5 months and subsequently became ventilator dependent. A diagnosis of TIS was made and at that time, his computerized lung volumetry was only 197 ml. At the age of 2 years, his body weight and height were below the third percentiles for his age (Fig. 1). To address the TIS, he underwent right and left VEPTR at the age of 38 and 59 months of age, respectively. His lung volume increased by 51% over the course of subsequent 3 years and he no longer required oxygen supplementation. He was able to participate in prolonged ambulatory activities for the first time.

During this treatment period, there was minimal increase in spinal height, and his body height was below the third percentile for his age. Hence, rib-to-pelvis MCGR devices were sequentially implanted bilaterally to distract and lengthen the spine when he was aged 8 (Fig. 2a, b). The VEPTRs were kept in situ, and serial distractions were performed to maintain the thoracic cage volume. Over the course of the subsequent 4 years, he underwent 24 MCGR lengthenings on the left and 21 on the right using an external magnet in the outpatient setting. The total length distracted was 26.4 cm on the left rod and 23.6 cm on the right rod. There was an increase in body height by 15.3% during this period from 106.8 to 115 cm; lateral spine height by 5% from 140 to 151 cm; C7 plumb line-S1 distance by 93% from 34.6 to 67 cm, and thoracic width by 8%. He maintained a satisfactory respiratory function with a forced vital capacity (FVC) of 0.42 l, forced expiratory volume in one second (FEV_1) of 0.37 l (18% of normal), and his CT lung volumetry increased from 250 cm^3 preoperatively to 640 cm^3. He continued to be free from respiratory infections during this period.

At aged 12, 4 years after MCGR implantation, no further distraction could be achieved (Fig. 2c, d). No further gains in body height and spinal length were recorded (Fig. 3), and serial lateral radiographs showed progressive kyphosis. At the same time, he began to suffer from repeated chest infections. The triradiate cartilage was closed but the Risser sign was still grade 0. Since the MCGRs were no longer functional and his pulmonary development was complete, all VEPTR and MCGR

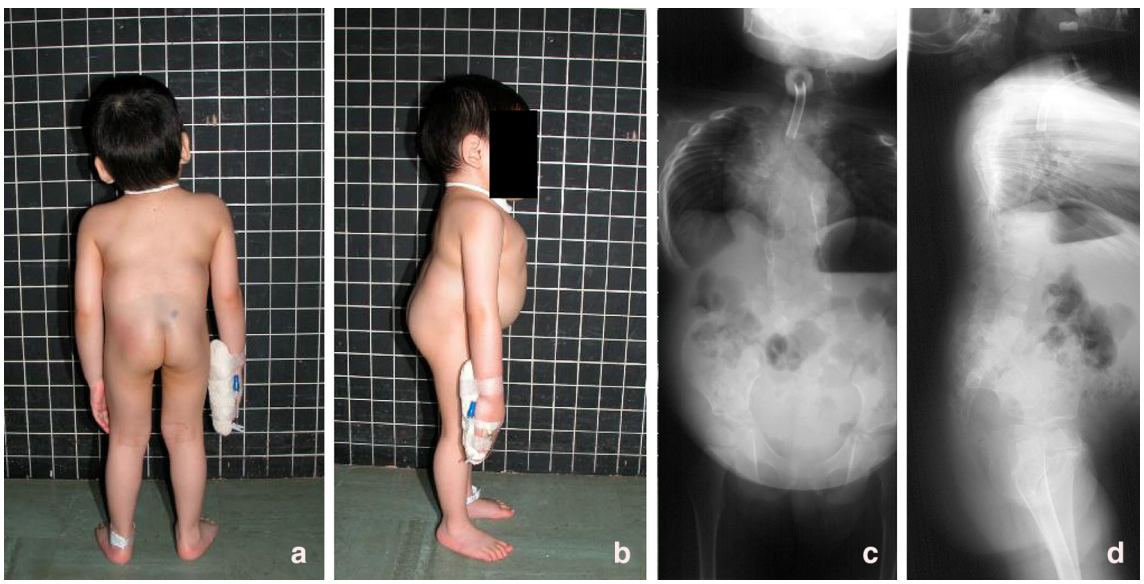

Fig. 1 Preoperative clinical photo showing frontal (**a**) and lateral (**b**) views. Corresponding preoperative posteroanaterior (PA) (**c**) and lateral (**d**) views

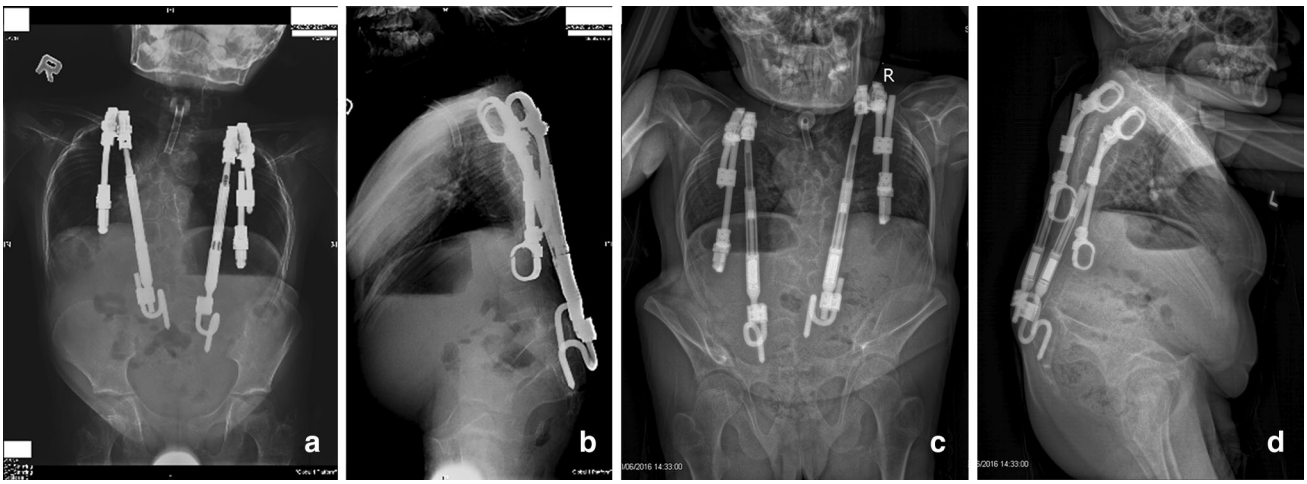

Fig. 2 Immediate postoperative PA (**a**) and lateral (**b**) radiographs after insertion of bilateral magnetically controlled growing rods (MCGR). PA (**c**) and lateral (**d**) radiographs at the end of maximal MCGR distraction

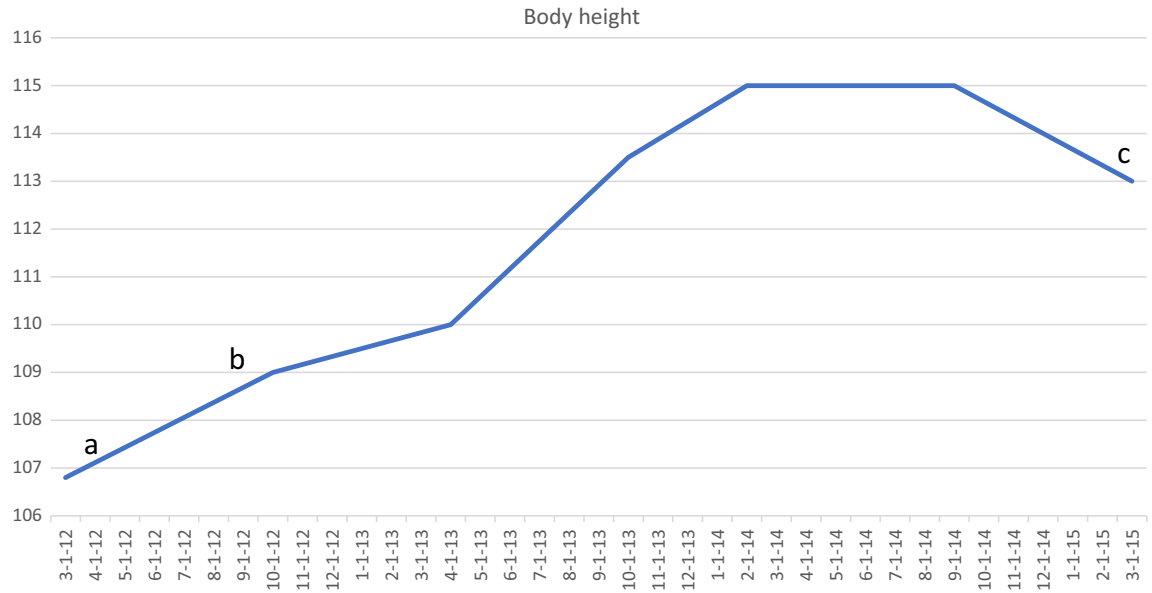

Fig. 3 Graph showing the change in body height from the beginning of treatment to final follow-up. The following time points are denoted: **a** insertion of left MCGR; **b** insertion of right MCGR; **c** end of MCGR distraction before removal of implants

implants were removed. His latest radiograph showed that his sagittal alignment had improved (Fig. 4), and he is being observed for recurrence of spinal deformity or need for spinal fusion.

Discussion

We presented a novel approach in the management of TIS with complex spinal deformities using a hybrid MCGR–VEPTR construct in a patient with JLS. It illustrated a combined approach of increasing thoracic cage volume and pulmonary function using VEPTR and successful distraction and lengthening of the spine without the need for repeated surgeries and wound re-openings using MCGR. Overall the patient enjoyed improved pulmonary function, increased spinal length, body height with better quality of life.

The aim of management of TIS is to increase the thoracic volume in early childhood to encourage normal alveolar development and improve respiratory biomechanics. The VEPTR expansion thoracoplasty was developed to address directly the segmental hypoplasia of the hemithorax resulting from fused ribs associated with congenital scoliosis in TIS [3]. In the initial presentation, the over-riding clinical problem was bilateral restricted lung

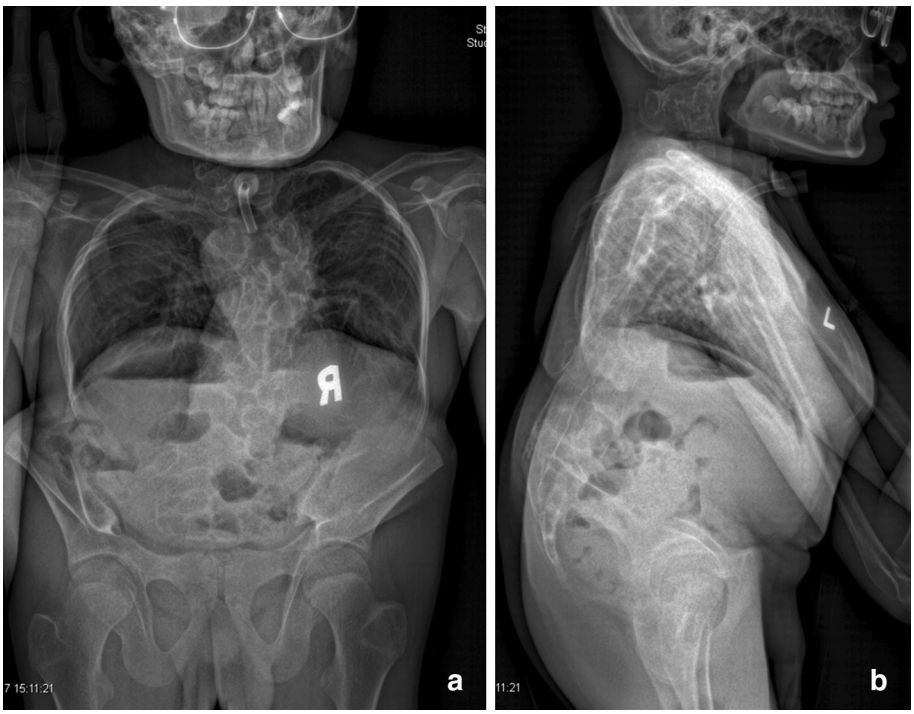

Fig. 4 PA (**a**) and lateral (**b**) radiographs at final follow-up after removal of all implants

volumes due rib fusion, causing TIS which was associated with a significant mortality [3]. Hence a rib-to-rib VEPTR was inserted which substantially increased his vital capacity and improved his quality of life, allowing him to ambulate without respiratory support during childhood. Spinal growth, which was variable in such syndromes, was a secondary issue after the chest condition improved. When alveolar growth was complete and lung volume reached its expected size, further rib-to-rib VEPTR distractions would not be beneficial or affect the spinal length.

Abnormal vertebral segmentation or formation defects in JLS cause short trunk dwarfism, and JLS patients have limited growth potential. The spinal height and overall size of these patients may be as low as 25% of normal [6]. MCGR was developed to correct spinal deformities and maintain spinal growth in early-onset scoliosis non-invasively using an external remote controller. We first described the extended application of MCGR as an internal distractor for gradual correction in a girl with severe kyphoscoliosis and concurrent syringomyelia and Arnold-Chiari type I malformation using noninvasive techniques [15].

In this case, the rib-to-rib VEPTR was left in situ to maintain the volume of the thoracic cavity, whilst a rib-to-pelvis MCGR was applied to maximize spinal length and body height gain during the growth period. Although complications have been reported with the use of Dunn–McCarthy S-hook in rib-to-pelvis construct [16], there was no implant-related adverse event or unplanned return to the operating room in this case. In the present application, MCGR was superior to traditional growing rod (TGR) systems because it not only mitigated the need for repeated surgeries, but the rate and frequency of MCGR distractions could also be varied according to the patient's response, thus maximizing the visco-elastic properties of the spine and chest wall compliance. We found that after each distraction, the child experienced some pain that spontaneously settled within 24 h. This suggested that the soft tissues were tight, and each distraction episode had maximized the remaining growth potential. Since the natural rate of growth in JLS patients was undetermined, the ability to vary the length and frequency of distraction according to the clinical response was important. We postulated that using MCGR rather TGR, a more gentle but persistent distracting technique could be applied to achieve the increased spinal length. We previously also found that frequent small distractions with MCGR could avoid the law of diminishing returns [17]. A rib-to-pelvis rather than a spine-to-pelvis was chosen since the pedicles were significantly deformed, and placing downgoing hooks in the upper instrumented vertebra for MCGR distraction was not advisable, so using rib anchors in the proximal foundation was safest in this situation. The VEPTR was left in situ when the MCGR was implanted in case further spinal growth may require additional distraction of the thoracic cage.

Nonetheless, our case also highlights some challenges in the management of patients with combined TIS and short trunk dwarfism. During the first 4 years of MCGR distractions, the patient gained significant spinal length and height. However, continual distraction with a posterior-based construct eventually led to sagittal malalignment and our patient developed progressive kyphosis without an

increase in spinal length or body height. We postulated that the rigidity of the posterior implants restricted the natural lumbar lordosis, and continual rib-to-pelvis distraction resulted in pelvic retroversion, both of which contributed to the sagittal malalignment. The subsequent kyphosis led to reduced thoracic volume and inefficient respiratory mechanics, and the patient began to develop pulmonary complications again. Increase in thoracic kyphosis associated with VEPTR treatment has been noted in other series [7, 18, 19], although none has reported symptomatic progression. At this stage, the Risser sign was still grade 0 and the distal radial physis was still open, indicating skeletal immaturity. Although in a normal child at this chronological age, definitive fusion could be considered, in this case we elected to wait longer because any residual gain in height would be more significant in a congenitally short child. Furthermore, as the current magnitude of the curve is small and if no significant deformity develops, there is a possibility that the child may not need definitive fusion. Alternately, autofusion may have already occurred and final fusion is not needed [20]. After removal of all implants without final fusion, the spine appeared to return to its natural sagittal alignment, and the pulmonary function also improved. He will be monitored for any further spinal deformity until skeletal maturity.

Conclusions

This case report highlights the challenges of improving lung function and maintains spinal growth and height in a JLS patient. A rib-to-rib VEPTR addressed the TIS. Subsequent addition of a spine-to-pelvis MCGR allowed non-invasive distraction with variable frequency and distraction length, thereby making use of the viscoelastic properties of the spine and soft tissues to maximize spinal length and height of in a child with limited growth potential. This innovative use of VEPTR and VEPTR MCGR hybrid construct can be considered in early-onset scoliosis patients suffering from similar clinical problems.

Compliance with ethical standards

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

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