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Scoliosis in Gordon's syndrome

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Abstract Gordon's Syndrome is described as an autosomal dominant condition with the characteristics of short stature, a stiff spine, camptodactyly (89%), cleft palate (27%) and club feet (73%). The authors present a case report of a patient with this rare entity complicated by an unusual complex spinal deformity. There are no prior reports in the literature concerning operative or nonoperative management of deformity in this patient population. Scoliosis in Gordon's Syndrome shares the characteristics of an arthrogrypotic neuromuscular curve and de-

mands extensive soft tissue release for optimal correction.

Keywords Gordon's syndrome · Arthrogryposis · Scoliosis · Neuromuscular diseases

Introduction

The association of a spinal deformity in musculoskeletal syndromes of genetic or developmental aetiology is frequent. In 1969, Gordon et al. described a syndrome of autosomal dominant inheritance characterised by camptodactyly, cleft palate and club foot [6]. A spinal deformity has not previously been reported as a feature among the individuals affected by Gordon's Syndrome. The authors present a case report of a patient with this rare clinical entity complicated by an unusual complex spinal deformity.

Case report

A 13-year-old girl was referred to our spinal unit for her spinal deformity, which was detected in a routine school screening programme. She had been diagnosed at birth to have Gordon's Syndrome, with deformities including camptodactyly involving all fingers and toes, cleft palate (surgically corrected) and club feet (corrected by serial plasters). She did not have any visceral anomalies

and her spine was normal. Her genetic screening revealed normal chromosomal pattern (44XX). Her amino acid assays did not reveal any specific abnormality. She was physically active and had normal intelligence. On genetic survey of her family tree, two other siblings had similar dysmorphism, though in milder forms, thereby supporting an autosomal dominant transmission. After correction of the cleft lip and club feet, she was followed up until the age of 6 and then discharged from further orthopaedic observation, as it was deemed not necessary.

At the first examination in our scoliosis clinic, she was 2 years post-menarche and had a King type II right thoracic scoliosis extending from T3 to T11 with a Cobb angle of 32° and a compensatory lumbar curve from L1 to L4 with a 20° Cobb angle. There was no evidence of vertebral anomaly. She had normal lung function and was neurologically intact. She was treated with a corrective Cheneau type brace and was followed up every 4 months. In the brace, the curve did correct initially to 20° in the thoracic curve and 15° in the lumbar curve.

At the age of 14 1/2, the thoracic curve had increased to 50° and remained uncorrected on bending views, and her lumbar curve was 35°, which corrected down to 20° on bending views (Fig. 1). Her respiratory function measured 90% of expected values, and she was Risser grade 4 and Tanner grade 4.

Considering the age of onset and the curve morphology, the initial impression of the deformity was of adolescent idiopathic sco-

Fig. 1 Posterior-anterior and lateral preoperative radiograph demonstrating the right thoracic curve of 50° and the lumbar curve of 35°, and a marked hypokyphosis (−10° from T4 to T12)

liosis. However, the significant rigidity as revealed in the bending films was more in support of a neuromuscular curve. We performed a preoperative MRI scan of the whole spine, which showed no evidence of cord cavity or other pathological findings.

The patient underwent one-stage combined anterior release (T3-T11, right fifth rib thoracotomy), with posterior release (facetomy and flavotomy T2-T11) due to the rigidity and the marked thoracic lordosis, followed by instrumentation from T2 to T11. The corrected thoracic curve, measured on postoperative standing film, was 15°, with satisfactory sagittal and coronal alignment (Fig. 2). The notable intraoperative observations included profound tissue bleeding despite hypotensive anaesthesia, routinely used in scoliosis surgery, abnormally tough soft tissues (disc, muscle and fascia) and osteoporotic bones with poor purchase of screws. At 18-months follow-up she was clinically pain free and well balanced, the radiological appearance showed a solid fusion and no loss of correction.

Discussion

In 1969, Gordon et al. reported on a family from Cape Town, South Africa, in which six members were affected

Fig. 2 Posterior-anterior and lateral radiograph at follow-up showing the correction of the thoracic curve down to 15° and the lumbar curve down to 12°, with a satisfactory restoration of the thoracic kyphosis to 20° from T4 to T12

with varying combinations of camptodactyly, cleft palate and club feet, with the pedigree pattern suggestive of a single mutant autosomal dominant gene [6]. Since then, a few other authors have reported cases with similar dysmorphic features and genetic pattern, thereby establishing Gordon's Syndrome as a recognised clinical entity [7, 10, 11]. None of the authors reported spinal abnormalities as a feature in the individuals affected by Gordon's Syndrome.

However, in 1982, Hall et al. proposed a comprehensive classification of distal arthrogryposis and designated Gordon's Syndrome as type IIA variant of distal arthrogryposis. They reported that this group of patients are characterised by additional features, notably spinal and craniofacial abnormalities [8]. There is considerable information available in the literature on the pathoanatomy and natural history of spinal deformities in arthrogryposis

multiplex congenita (AMC), but it is unclear whether spinal deformities of distal arthrogryposis syndrome behave in the same way as in AMC. In general, AMC is known to involve the trunk, predominantly with scoliosis, despite earlier literature suggesting relative trunk sparing. The incidence of spinal deformity in AMC has been reported to be as high as 42% [3]. Most of these curves are noted at birth or within the 1st year of life [9]. Curves are rigid very early in their development, and may require treatment in infancy. Most curves have paralytic characteristics, with associated congenital vertebral anomalies in 11–50% of the cases [2], and have significant potential for early progression [5]. The common curve pattern is of a long C-shaped thoracolumbar curve with or without pelvic obliquity resembling neuromuscular scoliosis [4]. Curve correction is hampered by the rigidity of the scoliosis and currently combined anterior release and posterior fusion techniques are usually recommended [1].

In this reported case, following satisfactory correction and follow-up of her cleft lip and feet, she was not on regular observation for any late-onset deformity because, by description, the deformities of Gordon's Syndrome are non-

progressive and the affected individuals are likely to have normal intelligence and life span. The deformity was recognised in her adolescence, which is an unusual presentation in this rare syndrome.

The curve morphology resembled an idiopathic adolescent scoliosis, but the rapid progression and rigidity raised doubts on the possibility of some neuromuscular elements associated with it. On surgical exploration, the extensive intraoperative bleeding, tough and fibrotic soft tissue elements including disc, muscle and fascia, and the significant osteoporotic bones were more in concordance with arthrogrypotic neuromuscular scoliosis.

Conclusion

In this patient with Gordon's Syndrome, the deformity resembled in its curve morphology adolescent idiopathic scoliosis, but it shared the characteristics of an arthrogrypotic neuromuscular curve and demanded extensive soft tissue release for optimal correction.

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