Case Report

Lessons learned from surgical management of craniovertebral instability in Morquio syndrome: A series of four unusual cases

ABSTRACT

Morquio syndrome (MS) is an autosomal recessive defect caused by the deficiency of N-acetylgalactosamine-6-sulfatase. Odontoid hypoplasia, periodontoid soft tissue deposition, and cervical stenosis lead to myelopathy and quadriparesis in these patients. Craniovertebral junction instability in MS possesses a surgical challenge as bones are yet to completely ossify. The atlantoaxial dislocation (AAD) is reducible, and the need of transoral decompression for the soft tissue deposition ventral to odontoid is debatable. We present a series of four cases (mean age 4.3 ± 0.4 years) operated through posterior-only approach (n = 2, C1-lateral mass to C2 pars-interarticularis [Goel's technique]; n = 1 sublaminar wiring followed by C1-lateral mass to C2 pars-interarticularis; and n = 1 suboccipital plate with pars-interarticularis of C2 screw and pedicle of C3 and rod fixation). All patients had acceptable outcome and doing well at the last follow-up (12–96-follow-up). None of our patient needed transoral decompression. Patients with MS frequently manifest with spastic quadriparesis at an early age due to reducible AAD. Early surgical fixation with posterior C1–C2 screw and rod technique is recommended for the favorable surgical outcome and long-term stability of the cervical spine.

Keywords: Chondroitin-6-sulfate, Goel's technique, keratan sulfate, Morquio syndrome

INTRODUCTION

Odontoid hypoplasia, periodontoid soft tissue deposition, and cervical stenosis are the prominent causes of myelopathy and quadriparesis in a patient of Morquio syndrome (MS). The associated joint involvement and thoracolumbar curvature anomalies add up to the morbidity. Craniovertebral junction (CVJ) instability in MS possesses a surgical challenge as bones are yet to completely ossify, glycosaminoglycan deposited ventrally compromises the canal diameter, and natural course of the disease is unpredictable. The role of transoral decompression is controversial as the authors support the resolution of soft tissue deposits after posterior stabilization. Added vigilance and understanding of the growth patterns after fusion are desired. In this article, we present four cases of MS who were managed surgically and highlight the surgical outcome of the same.

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CASE REPORTS

The mean age of four patients was 4.3 ± 0.4 years [Table 1]. All the patients had characteristic facial features such as orbital hypertelorism, broad nasal bridge, malar prominence, and flat forehead. Clinodactyly was seen in one patient,

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Table 1: Clinical and radiological details of patients with Morquio syndrome (n=4) in our study

Number	Age (years)	Sex	Preoperative Nurick grade	Clinical association			Radiological association				Surgery	Postoperative Nurick grade	Follow-up months	Follow-up Nurick grade
				Facial anomaly	Acral anomaly	Others	Clivus	Odontoid	Atlas	Axis	Subaxial				
1	4	Male	3	+Coarse	+Platyspondyly	Pectus carinatum	-	Aplasia	Bifid	-	-	C1-C2 fusion	3	96	3
3	4	Male	3	+	+	Pectus carinatum	Hypoplasia	Aplasia	C1 arch stenosis, Bifid	-	-	C1-C2 fusion	2	60	2
4	4	Female	3	+ Coarse	+ Clinodactyly	Pectus carinatum MVP/MR	-	Aplasia	Bifid	-	Beak-shaped vertebrae	C1-C2 sublaminar wiring followed by Goel's C1-C2 fusion	3	72	3
2	5	Male	1	+Coarse	+	Pectus carinatum	-	Aplasia	Assimilated	-	-	OC2-C3 fusion	1	12	1

MVP/MR - Mitral valve prolapse/Mitral regurgitation

and the cardiac anomaly was found in one patient, while pectus carinatum was characteristically present in all four patients. All four patients of MS had odontoid aplasia or agenesis, which lead to C1–C2 instability (translational dislocation) and needed posterior fixation. The stenotic element (mucopolysaccharidosis [MPS] deposition anterior to the cord) additionally compresses the spinal cord; however, in our experience, none of the patients of MS required ventral decompression. Bifid atlas arch was found in one patient. One patient underwent C1–C2 sublaminar wiring; one patient required suboccipital plate with pars-interarticularis of C2 screw and pedicle of C3 and rod fixation. Two patients underwent C1–C2 fixation (Goel's and Harm's technique). Individual consent to use clinical and radiological details for publication was taken at the time of admission, as per our department policy.

Case 1

A 4-year-old male patient, with lower limb deformity in since birth, presented with progressive spastic quadriparesis for the last 4 years (Nurick grade III). Preoperative two-dimensional echo revealed myxomatous degeneration of heart valves and mitral regeneration. He underwent lateral mass of C1 rod and screw fixation with pars-interarticularis of C2 screw (Goel's technique). Postoperatively, there was an improvement in spasticity, and the child discharged on the 7th day. At the time of writing, the patient has completed an 8-year follow-up and doing well [Figure 1].

Case 2

A 4-year-old male child referred from the medical genetics department with a complaint of gradually progressive spastic quadriparesis for the last 2 years (Nurick grade III). Dynamic CT images were suggestive of fixed atlantoaxial dislocation (AAD) and partially occipitalized atlas. Magnetic resonance imaging suggestive of C1 stenosis, C1 lateral mass, and C2 pars interarticularis fixation by classic Goel's

Figure 1: A patient of Morquio syndrome with "dish face" (a), lower limb deformity (e), and pectus carinatum (d) had odontoid hypoplasia and soft tissue deposition (b), cervical canal stenosis at craniovertebral junction (c) and underwent lateral mass of C1 rod and screw fixation with pars-interarticularis of C2 screw (f and g)

technique was performed with excision of the C1 posterior arch. Postoperatively, there was an improvement in spasticity and power, and the child discharged on 7th the day. At the time of writing, the patient has completed a 5-year follow-up and doing well [Figure 2].

In the third case, the sublaminar wiring technique was used for fixation [Figure 3]. After 18 months of follow-up, the patient complained of "click" sounds on the neck movement along with progressively increasing spasticity. This child needed re-surgery, and the sublaminar wires were removed and replaced with C1 lateral mass to C2 pars with rod. At the time of writing, the patient has completed a 6-year follow-up and doing well, and his spastic weakness improved. The fourth case also underwent suboccipital plate with pars-interarticularis of C2 screw and pedicle of C3 and rod fixation [Figure 4].

DISCUSSION

MS is an autosomal recessive defect caused by the deficiency of either N-acetylgalactosamine-6-sulfatase (MPS IV, type A) or β-galactosidase (MPS IV, type B), resulting in the accumulation of keratan sulfate (KS) and chondroitin-6 sulfate.[1] The accumulation of excessive KS and chondroitin-6-sulfate primarily in the lysosomes of bone, cartilage, ligaments, and extracellular matrix of these tissues leads to skeletal spondyloepiphyseal dysplasia. The characteristic clinical features consist of short trunk stature, cervical spinal cord compression, pectus carinatum, kyphoscoliosis, knock-knee, and hypermobile joints. Other rare features are characterized by marked odontoid hypoplasia, platyspondyly (flat vertebra), coxa valga, and laxity of joints.[2] These children are usually healthy at birth, and diagnosis of the syndrome is difficult to make until 2–3 years of age since total urine glycosaminoglycan level is within a normal limit. These glycosaminoglycans accumulate posterior to the dens, further decreasing the space available for the cord at this level.[3] Furthermore, atlantoaxial instability from odontoid hypoplasia is almost universal in this condition. Very few cases of cervical spinal cord compression without dens hypoplasia has been reported in the literature till date.[4]

Odontoid hypoplasia is a characteristic feature of CVJ involvement in MS, which occurs due to impaired ossification of the odontoid process. The AAD is the reducible type with C1 hypoplastic or dysplastic. Moreover, these patients present in early childhood, before complete ossification of the odontoid process, so atlantodental interval could not measure for the objective assessment of AAD. Paraplegia or sudden death secondary to upper cervical spine instability and cord compression is well-documented serious consequences of Morquio A.[5] Prophylactic fixation may prevent this unwanted death. Death commonly occurs by the age of 7 years secondary to cervical myelopathy, as well as effects on the respiratory system and resultant hypoxia.[5] In his series of four patients, Muthukumar showed the technical difficulty in the infixing screw in lateral mass of these patients.[6] He showed the importance of counter-pressure by the assistant surgeon in stabilizing a rotatory dislocation.[6] In one of his patients, he tried Goel's technique, but excessive blood loss prevented the procedure, and subsequently, a translaminar screw was placed. Surgical fixation in an asymptomatic patient is still debatable. Giussani *et al*. proposed delayed surgical treatment until 3 years of age as they believed that bone maturation is sufficient enough to allow infixing screws after 3 years of age.[7] Our series highlights the role of surgery in halting disease progression. Although the median Nurick

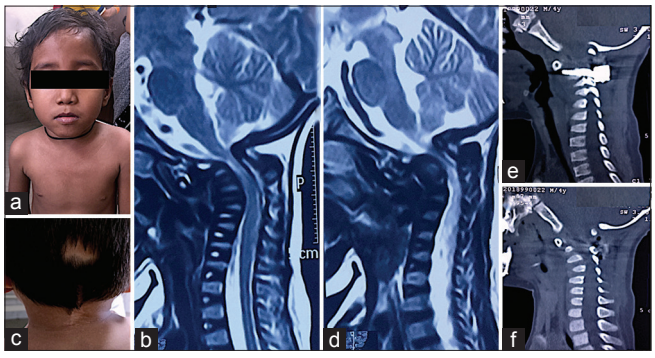

Figure 2: Second case of Morquio syndrome in our series who underwent lateral mass of C1 rod and screw fixation with pars-interarticularis of C2 screw (Goel's technique) (a). Magnetic resonance imaging showing (b and d) a partially occipitalized atlas, suggestive of C1 stenosis, bullet-shaped vertebrae. Postoperative surgical wound is shown (c). C1 lateral mass and C2 pars-interarticularis fixation by classic Goel's technique was performed with excision of the C1 posterior arch (e and f)

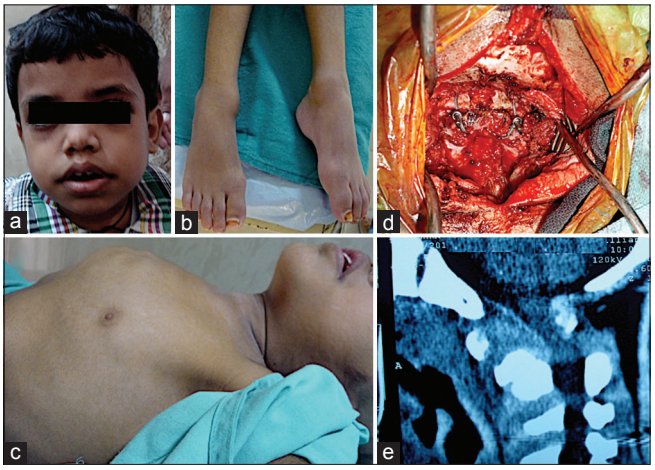

Figure 3: Third case of Morquio syndrome with broad facies (a), lower limb deformity (b), and pectus carinatum (c). Patient had odontoid hypoplasia and mucopolysaccharide deposition (e) for which he underwent sublaminar wiring (d)

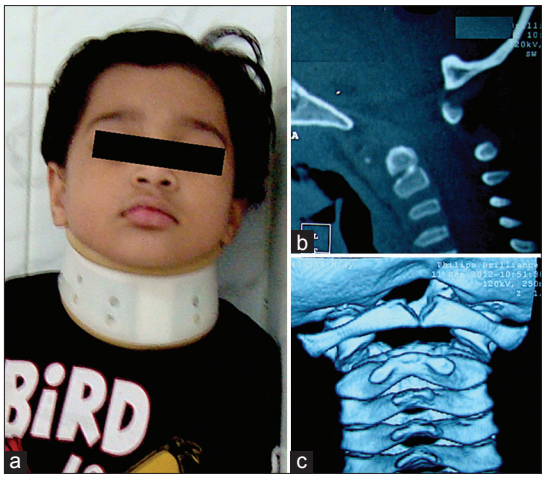

Figure 4: Fourth case of Morquio syndrome (a) with odontoid hypoplasia (b and c) suboccipital plate with pars-interarticularis of C2 screw and pedicle of C3 and rod fixation

grade did not change, the requirement of antispasticity drugs, neck pain, and self-care was improved in our study. According to Stevens *et al*., every patient of MS should be investigated for spinal cord compression between the ages of 3 and 8 years.[8] Treatment aims to avoid neurological damage or at least to arrest the progression of neurological disability. Prophylactic fusion was reported to have better neurologic consequences compared with fusions performed after neural compromise.[9] Ain *et al*. recommended instrumentation even for the asymptomatic patient if the spinal cord space is 8 mm and for patients with 5–8 mm of cervical instability with evidence of spinal cord impingement or damage on flexion–extension radiographic imaging and magnetic resonance imaging.[10] Posterior occipitocervical fusion results in the disappearance of the ventral soft tissue thickening and normalization of dens development in MS.[11] All the four patients in our study were operated by posterior fusion approach only. Goel's technique of direct opening the joints and manual manipulation of facets is beneficial for rotatory dislocations also (common in MS).[12] Giussani *et al*. suggested early surgical fusion in a symptomatic patient because of the disease progression in almost inevitable.[7] In another series of three patents, Nehete *et al*. reported C1 stenosis in MS, and the excision of the C1 arch along with fixation was performed.[13] He also postulated that C1 arch stenosis in MS is either due to a soft tissue mass or incomplete fusion of the C1 arch. According to our experience, transoral decompression is recommended only if the patient fails to show clinical improvement and radiological persistence of soft tissue deposit even after posterior fusion. In posterior fixation, screw and rod technique (Goel's and Harm's technique of C1/C2 fusion) is preferable over C1–C2 wiring due to its ability to provide immense stability. In the case of a congenital disability in the anterior and posterior arch, fixation of titanium connector between two rods will help yield extra stability in the postoperative rotational movement at the C1–C2 level. In a follow-up, there was a mild restriction of neck movements, and bony fusion was seen among all the patients. Three patients with long-segment follow-up showed 30° flexion, 15° of extension, 15° of lateral bending, and 15° of rotation each side.

CONCLUSION

Patients with MS frequently manifest with spastic quadriparesis at an early age due to reducible AAD. Myelopathy is usually attributed to the combination of odontoid hypoplasia with C1–C2 instability and retro-dens soft tissue collection. Early surgical fixation with posterior C1–C2 screw and rod technique is recommended for the favorable surgical outcome and long-term stability of the cervical spine.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patients have given their consent for their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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