International Orthopaedics (2019) 43:159–167
https://doi.org/10.1007/s00264-018-4143-0

REVIEW ARTICLE

Spine challenges in mucopolysaccharidosis

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Received: 7 July 2018 / Accepted: 3 September 2018 / Published online: 14 September 2018

Abstract

Purpose Mucopolysaccharidosis (MPS) are rare inherited metabolic diseases, causing lysosomal storage of mucopolysaccharides; clinical presentation involves skeletal system and particularly the spine. Anomalies include developing kyphosis at thoracolumbar junction, that can causes nervous symptoms, and dens hypoplasia with associated atlantoaxial subluxation that can cause myelopathy. We present our experience in the treatment of spine pathology in MPS.

Methods Medical treatments of MPS seem to have little impact on spine disease: treatment of cervical instability often includes surgical decompression and stabilization, as in patient MPS1 that we present, while thoracic lumbar kyphosis is treated by bracing and, in severe cases, with surgery. Bracing is more effective in kyphosis under 40° Cobb. Our surgical cases with thoracic lumbar kyphosis over 40° Cobb, treatment include the first one ever described by only posterior approach with vertebrectomy in MPS and a case of lateral costo-transverse approach instrumented correction.

Results Surgical patients had no major complications after surgery and CT scan at follow-up showed complete fusion without loss of correction, even if in a cervical case we used an adult rigid instrumentation in a four year-and-six month-old girl (11 years follow-up) and in thoracic lumbar kyphosis case treated by vertebrectomy due to diminutive anatomy we positioned interbody cage in suboptimal position.

Conclusions Bracing is a viable treatment strategy in thoracic lumbar kyphosis and can obtain good clinical results at medium terms follow-up even if kyphosis deformity remains in radiographs. Surgical treatment is effective in severe evolving cases both at cervical and thoracic lumbar level, main difficulties arose from unavailability of dedicated instrumentation in very young patient, as even smallest devices available are often too big.

Keywords Spine surgery in mucopolysaccharidosis · Thoracic lumbar kyphosis · Cranio cervical stabilization · Thoracic lumbar kyphosis surgical treatment

Introduction

Mucopolysaccharidosis (MPS) are a group of rare inherited metabolic diseases (overall incidence about one in 22.000 births) caused by mutation of enzymes involved in the catabolism of long-chain carbohydrates called glycosaminoglycans (GAGs) or mucopolysaccharides: GAGs compose proteoglycans, proteins that provide function and integrity of connective tissues, granting hydration and swelling pressure to tissues [1]. Enzyme deficiency causes accumulation of GAGs within lysosomes of cells of parenchymal and mesenchymal tissues, leading to multiorgan dysfunction.

Skeletal manifestations in MPS, known as "dysostosis multiplex" [2], are a wide range of abnormalities [3]. The introduction of haematopoietic stem cells trasplantation in 1981 [4] improved the disease progression and patients survival in MPS I and IV. Successful engraftment provide a source of enzyme by donor cells resulting in improved cardiopulmonary function, decreased hepatosplenomegaly and improved neurologic status, and patients survive to adult age [5–7].

As life expectation increases in patients with MPS treated by haematopoietic stem cells transplantation the treatment of orthopaedic problems becomes more important, as the majority of musculoskeletal deformities seem largely unresponsive to bone marrow transplantation, presumably due to insufficient penetration of the donor enzyme in the skeletal tissue [8–10].

Bone abnormalities in MPS presumably arises from a lack of primary ossification at several sites, a lack of secondary

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bone remodeling, and dysfunction of ligamentous structures and joint capsules [11].

In relation to spinal involvement, vertebral, and skull base anomalies and GAG deposits in the meninges and in the supporting ligaments often cause atlantoaxial subluxation and instability that can result in spinal cord compression with or without signs of compressive myelopathy [12–15]. The thoracolumbar spine and more commonly the craniocervical junction are the two major locations of spinal cord compression.

The hallmark orthopedic feature in MPS I and IV is thoracolumbar kyphosis resulting in gibbus deformity, which has historically played a significant role in the diagnosis of MPS [13, 14]. Gibbus deformities occur in nearly all children with MPS I [15].

Early development is due to a lack of primary ossification [16]: abnormal growth creates a defect of the anterosuperior development of the vertebral body. Weight bearing forces worsen growth disturbance and lead to retrolisthesis of the hypoplastic vertebra, resulting in trunk anterior shift. Progression of the sagittal deformity and associated scoliosis is the result of both growth-plate dysfunction and architectural modifications of ligamentous and capsular joints [17]. The affected vertebra shows a typical "beaked" appearance on radiographs [18] and anterior and posterior disc bulging on magnetic resonance imaging [19].

Sagittal alignment imbalance enhances spinal deformity: vertebral dysplasia, hip, and knee flexion contracture and weak abdominal muscle tone cause anterior shift of the trunk. To restore trunk balance, compensatory mechanism occurs, including thoracic lordosis and pelvic retroversion; as pelvic incidence in these patients remain low compared with general population, pelvic retroversion acts in a limited way to compensate for sagittal imbalance [17]. Weak abdominal muscle tone increases the flexion moment of the lumbar spine; deformity progression maintains the pelvic incidence in a low range, closing the vicious circle. Kyphosis is always located in the thoracolumbar junction, as it is a weak area more likely to suffer from axial hypotonia [17]. The apex of the kyphosis is usually located at the L2 vertebra [15].

Left untreated, the kyphosis is likely to progress causing progressive clinical deformity and possible neurological deterioration [10, 15, 20, 21].

Almost all patients affected by mucopolysaccharidosis IVA (Morquio-Brailsford or Morquio A syndrome) presents dens hypoplasia or os odontoideum associated with some degrees of cervical cord compression due to periodontoid soft tissue mass, composed by GAG [22].

Dens hypoplasia and associated laxity of transverse and alar ligaments lead to instability between anterior arch of the atlas and the dens causing atlantoaxial subluxation [23].

Atlantoaxial subluxation cause a pincer-like effect between atlas posterior arch and the axis, indenting the dorsal aspect of the spinal cord and causing cord compression when, during flexion of cervical spine, the anterior arch of the atlas moves anteroinferiorly while the odontoid process tilts posteriorly [24].

On lateral radiographs of the cervical spine, an atlantodental interval over 3 mm in adults and over 5 mm in children is abnormal and when it differs by more than 2 mm on functional flexion-extension lateral radiographs; it is a sign of atlantoaxial instability [25]. Atlantoaxial subluxation that exceeds 9 mm atlantoaxial interval is more likely to cause spinal cord compression [26].

Spinal stenosis is described in MPS patients, due to focal or multilevel thickening of the connective tissues within the spinal canal, caused by GAGs deposition in the meninges, posterior longitudinal ligament, and flavum ligament [24].

Spinal cord compression is a common complication of spinal involvement in MPS: it may involve multiple cord levels, but it is more common at the craniocervical junction and in thoracolumbar spine. There are two main etiopathologic mechanisms involved in the compression at the craniocervical junction: the central spinal canal stenosis due to GAGs deposition in the peri-odontoid tissue, supporting ligaments and meninges, and the atlantoaxial subluxation and instability related to dysplasia of the odontoid process and ligamentous laxity. At thoracolumbar level, spinal cord compression is secondary to kyphosis due to deformed "beaked" vertebra that causes spinal canal narrowing.

Plain radiographs in anterior posterior and lateral view are usually the initial imaging assessment of spinal involvement of the spine in MPS patients, and sometimes in patients with suspect MPS the presence of characteristic vertebral anomalies, such as beaked vertebra, addresses the diagnosis.

MPS patients' spine typically presents a combination of anterior "beaking" and posterior "scalloping" of vertebral bodies, but in imaging findings is impossible to differentiate between MPS I, IV, and VI, the forms that tend to have greater spinal involvement in MPS. Patients with MPS IV appear healthy at birth and spinal abnormalities appear in early childhood [27]. Beaked vertebra bodies of thoracolumbar junction are present in both MPS I and MPS IV patients' spine: MPS I patients, however, often shows anterior beaking of the inferior aspect of vertebral body [18], while MPS IV patients shows anterior beaking of the midportion of the vertebral body [25].

Functional extension-flexion lateral views of cervical spine assesses vertebral instability, but in some patients with basilar invagination or enlarged mastoid processes (typical in MPS IVA) [28], C1–C2 level is not clearly visible limiting usefulness of this exam.

CT scan is more accurate than plain radiographs in detecting vertebral bony changes, particularly in characterization of the odontoid process and atlantoaxial articulation.

MRI is used in monitoring nervous complication of MPS spinal involvement: spinal canal stenosis, spinal cord compression, and myelopathy [28]. Signal hyperintensity in T2-weighted images at the level of spinal compression shows

edema or myelomalacia on ischemic base, but MRI myelopathy not always correlate with nervous symptoms, as in most cases the nervous deficit are less severe than suggested by imaging [25]. When open MRI system is available, flexion-extension MR imaging of the cervical cord shows elements useful to investigate cervical spine instability and cord compression in pre-surgical planning [29].

Material and methods

Some MPS types (mainly MPS I and MPS VI) are treated by haematopoietic stem cell transplantation while intravenous enzyme replacement therapy (ERT) is available for MPS I, II, and VI [30, 31]. Haematopoietic stem cell transplantation avoids cardiac and nervous impairment of MPS, but seems to have no impact on skeletal deformities. Surgery therefore remains the standard treatment for MPS spine disease in two basic issues: a) spinal cord compression with myelopathy at craniovertebral junction and thoracolumbar level is clearly and indication for intervention; b) surgical correction is also required in MPS patients with progressive and symptomatic kyphosis to prevent progression and nervous impairment.

Aims of spine surgery in MPS patients are restore vertebral alignment, protect nervous structures and stabilize the spine maintaining the best possible range of mobility.

Intervertebral instability at craniovertebral junction is treated by luxation reduction and stabilization by Halo traction and occipitocervical fusion; decompression before fusion could be mandatory in case of severe spine cord compression. In MPS IV (Morquio), atlantoaxial instability and spinal cord compression caused by GAGs deposits in peri-odontoid tissues are so common that some authors recommend prophylactic fusion, eventually associated with cord decompression, at an early stage of disease in these patients [32, 33]. Posterior occipitocervical fusion is the most common procedure in treating MPS occipitocervical junction instability, but patients with severe spinal cord compression may require decompression and posterior occipital cervical fusion [34]. In our experience, we surgically treated a four year-six month-old girl affected by MPS I, with severe cervical spine stenosis and occipitocervical junction instability, tetraplegia in patient with hydrocephalus treated by ventricular derivation. Patient has been treated by posterior occiput-cervical decompression and occipital cervical stabilization by two rods, screws at occipital level and hooks at cervical level. As there was no dedicated cervical pediatric instrumentation, we used the adult rigid instrumentation available (Fig. 1).

Thoracolumbar kyphosis is a common spine deformity associated to Hurler syndrome (MPS I) and some authors consider thoracolumbar kyphosis almost universal in these patients once they are able to walk [20]. The deformity does not improve with medical treatment due to lack of skeletal penetration of the replaced enzyme and does not always respond to orthesis treatment. Untreated kyphosis is likely to progress. Various surgical strategies have been proposed and, at present, there is no clear evidence of the best indication in the treatment of the deformity as for surgical technique and for intervention timing.

Some authors sustain that thoracic lumbar kyphosis in MPS patient should be treated by bracing as soon as the child acquires sitting position [17]. In our experience, we treated by bracing thoracic lumbar kyphosis in 15 patients with Hurler syndrome, mean age six years three months (range 4 years 9 months–9 years), mean kyphosis at beginning of treatment 41° Cobb (range 36°–50° Cobb). Mean follow-up is three years three months (range 2 years 8 months–5 years). At final follow-up, mean kyphosis was 25° Cobb (range 20°–40° Cobb).

Bracing should relieve hypoplastic vertebra from weight forces [17], but it is not sure to prevent deformity progression [35], also in cases with good results at medium terms (Fig. 2). Thoracolumbar kyphosis exceeding 40° Cobb has propensity to progress [20, 35].

When kyphosis is progressive, surgery is indicated to avoid neurological deficits and impairment, in analogy to the surgical treatment of progressive congenital kyphosis [36]. We had no good results from previous experience with in situ fusion, and we wanted to reduce the morbility related to combined approach, anterior-posterior, as in treatment of congenital deformities. We applied our previous experience on the treatment of progressive congenital scoliosis, kyphoscoliosis, and kyphosis in young children by posterior approach only vertebra resection, to the problem of thoracolumbar kyphosis in Hurler syndrome. We have used posterior approach only hemivertebra resection from 2006 [41]. To our knowledge, our patient is the first case of thoracolumbar kyphosis in MPS 1 treated by posterior only approach and total vertebrectomy.

Our patient, with type 1 mucopolysaccharidosis, was affected by thoracic-lumbar kyphosis caused by hypoplastic L1 and L2 vertebras. Deformity increased even with brace treatment for 36 months (Fig. 3). At 6 years of age, when the patient came at our observation, he had 65° Cobb kyphosis T11-L3 on standing X-rays. At six years and one month of age, 105.5 cm height and 19 kg weight, the patient was operated by posterior approach only total L2 vertebra resection, obtaining progressive deformity correction by two rods and pedicle screws in T11-T12-L1 and L3 and filling the gap left by resection with one cage with autologous bone (Fig. 4). We used the smallest adult cervical cage available, 1 cm in diameter. During surgery intraoperative neurophysiologic monitoring was used. Intervention lasted eight hours, time strictly related to difficulties in cage positioning; total blood loss was 300 ml.

Kyphosis was reduced to 37° Cobb on standing X-rays; patient had no major complication (infection, instrumentation failure, nonunion, nervous, visceral, or vascular injuries). For

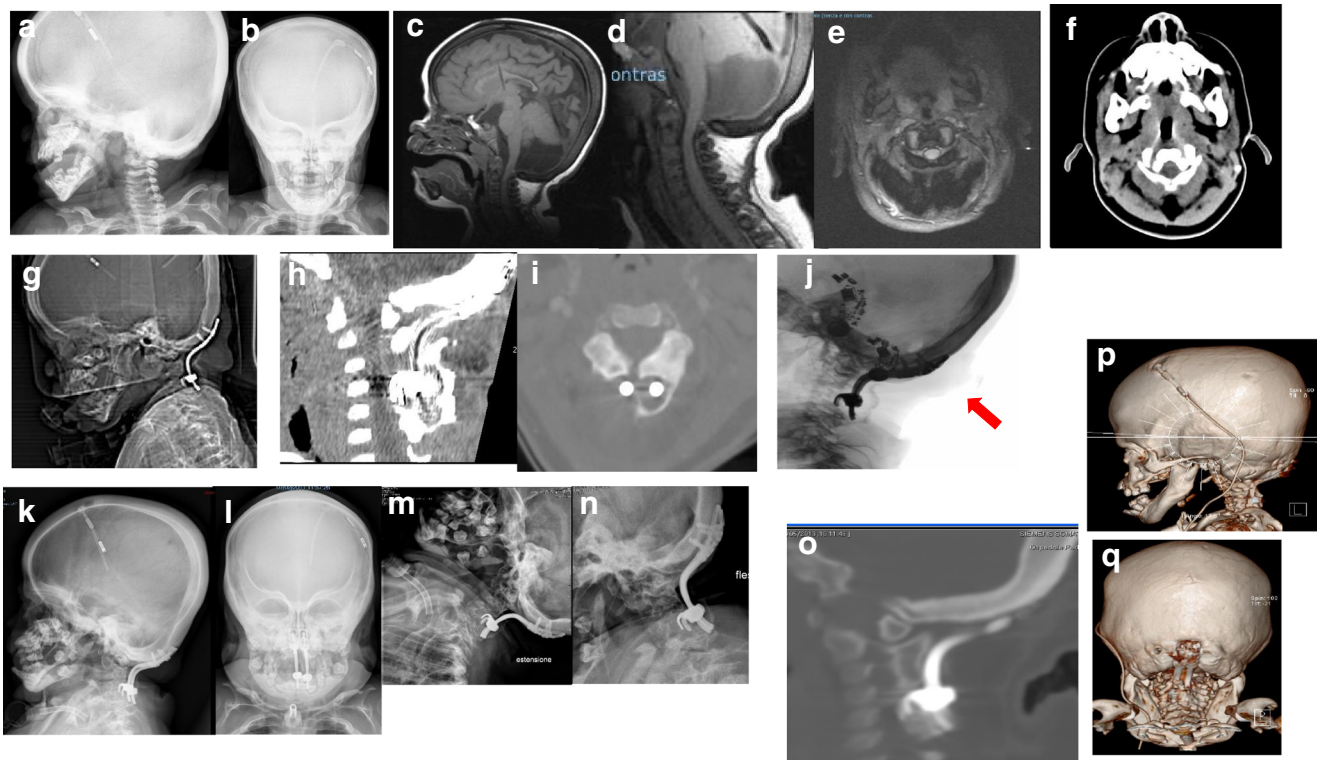

Fig. 1 I.V., a 4-year-6-month-old girl affected by severe MPS I; **a** and **b**: treated by ventricular derivation for hydrocephalus, developed tetraplegia; **c**, **d**, and **e** MRI and **f** CT show cervical canal stenosis; she has been treated by posterior cervical decompression and occipital cervical stabilization with rods, occipital screws, and cervical hooks; **g** lateral radiograph after surgery, **h** and **i** CT scan after surgery; **j** at 24 months FU she developed soft tissues swelling by protruding instrumentation; swelling solved by anti-inflammatory drugs; **k** and **l** anterior posterior and lateral radiographs at 11 years FU; **l** and **m** lateral dynamic radiographs show full stability at 11 years FU; **o**, **p**, and **q** CT scan with 3D reconstruction at 11 years follow-up shows complete fusion, with instrumentation completely embedded in fusion mass

about 4 weeks after surgery patient has paresthesia in his right leg. Symptom disappeared with steroid treatment. Patient was braced for four months after surgery. Eight months follow-up CT scan showed complete L1-L3 arthrodesis, and another CT scan was performed at 25 months follow-up, showing complete cage osteointegration. At four years follow-up, there was no instrumentation failure and no loss of correction (Fig. 4).

In our patient, critical aspect of operation is cage positioning, as limited vertebras dimensions (vertebral plate diameter was more or less 1 cm) did not allow an adequate positioning even of the most minimal device available, resulting in an oblique position of the cage. Even if in sub-optimal position, however the bone, obtained from the resected and morcellized vertebral body filled into cage, allowed a complete interbody fusion, as showed by CT scan at follow-up. We achieved an incomplete kyphosis resolution, with good cosmetic results and stability in the time, and may be this incomplete correction could be reason of no occurrence of proximal junctional kyphosis.

We treated by different technique another patient with severe thoracic lumbar kyphosis: the patient was seven years and seven months old boy affected by MPS I with worsening thoracic lumbar kyphosis, unsuccessfully treated by brace since four years of age. He had 70° Cobb kyphosis T10-L4. He had nervous symptoms complaining paresthesia in both legs and showing limbs somatosensory evoked potentials slowed down. His kyphosis was quite stiff as in the case of the boy treated by vertebrectomy, but partially reducible in extension lateral view radiographs. In this patient, we decided a different surgical approach from the first case, to avoid problems of cage positioning, as in the case previous described, and to avoid blood loss from vertebral osteotomy. We treated him by right lateral costal transverse approach, T11-T12 T12-L1, and L1-L2 discectomy; T11-T12, T12-L1, and L1-L2 interbody fusion with autologous bone graft from resected rib; and posterior instrumented arthrodesis T10-L4. We used pedicle screws and hooks in "claw fashion" at upper extremity, to obtain a more flexible construct, in our opinion, with lesser risk of developing proximal junctional kyphosis. T10-L4 thoracic lumbar kyphosis was reduced to 20° Cobb. (Fig. 5).

Results

A four year-six month-old girl treated by occipito cervical decompression and stabilization had recovery of the neurological status after surgery, and, at six month follow-up, she regained partial use of upper extremities. She had no complication in the immediate post-operatory period: at 24 months follow-up, she

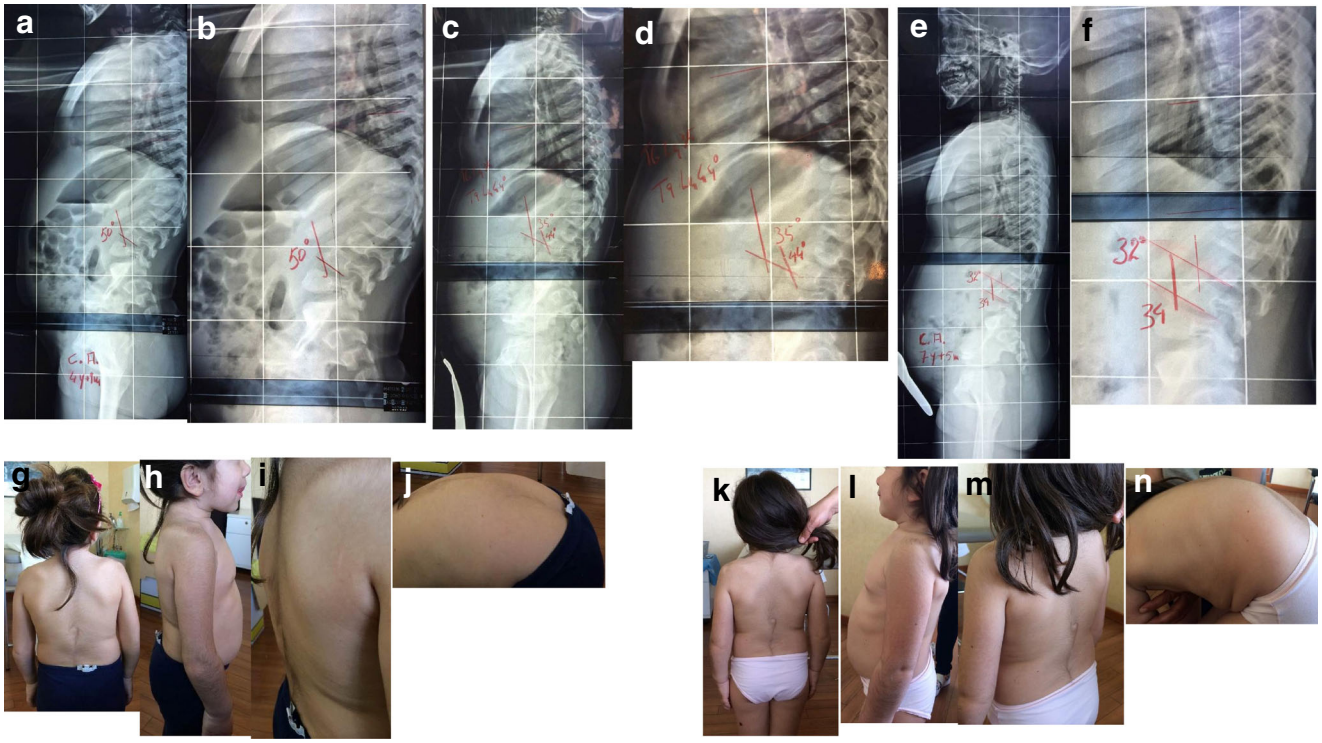

Fig. 2 C.A., MPS-affected girl, thoracic lumbar kyphosis sustained by beaked vertebra; **a** and **b** 50° Cobb thoracic lumbar kyphosis at 4 years 9 months of age, treated by bracing; **c** and **d** 5 years 9 months of age, 1 year brace treatment follow-up, total thoracic lumbar kyphosis reduced to 35° Cobb, segmental kyphosis 44° Cobb; **e** and **f** 7 years 5 months of age, 2 years 8 months brace treatment follow-up, total thoracic lumbar kyphosis reduced to 32° Cobb, segmental kyphosis reduced to 34° Cobb; **g**, **h**, **i**, and **j** clinical appearance at 5 years 9 months of age; **k**, **l**, **m**, and **n** clinical appearance at 7 years 5 months of age: optimal cosmetic result, no "gibbus" deformity

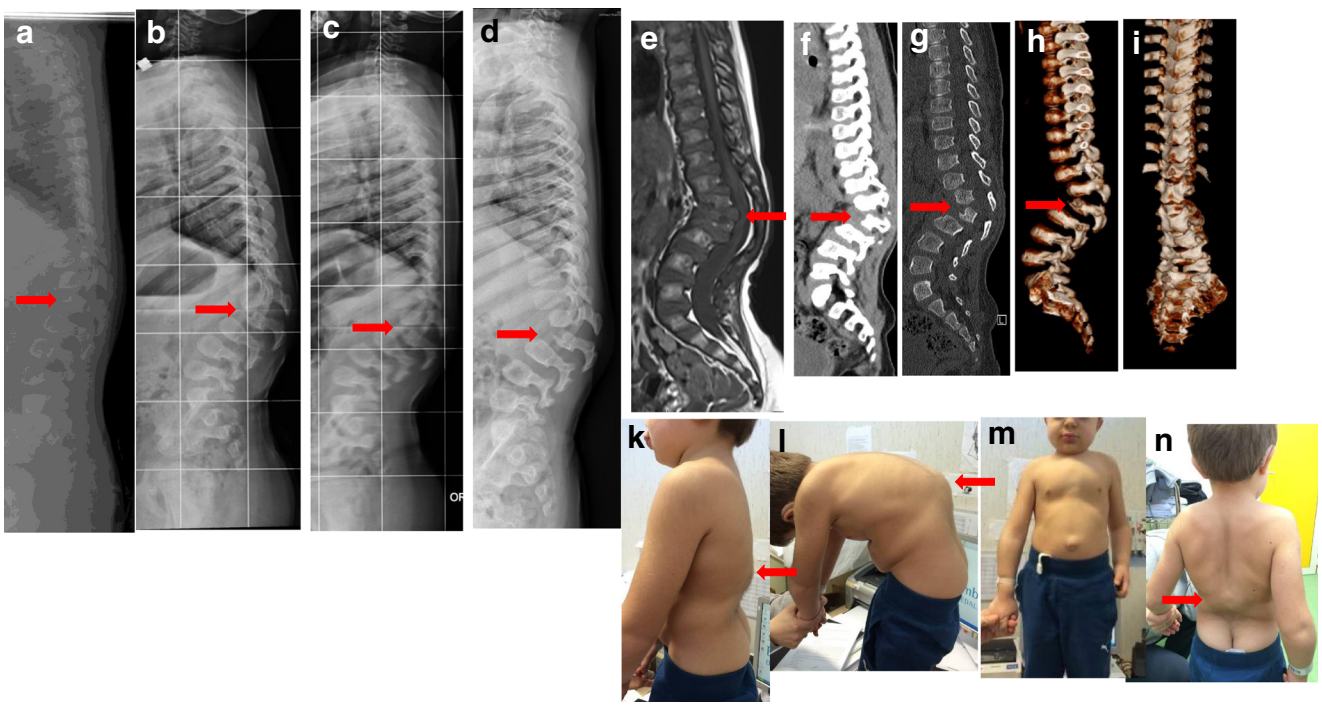

Fig. 3 C.R. MPS I-affected boy, treated by haematopoietic stem cells transplantation; progressive thoracic lumbar kyphosis not responsive to brace; kyphosis progression, spine lateral X-ray at: **a** 10 months, **b** 3 years and 7 months, **c** 5 years, **d** 6 years and 1 month; **e** MRI showing L2 protruding in vertebral canal; **f** and **g** CT scan showing typical beaked as **h** and **i** 3D CT reconstruction; **h**, **l**, **m**, and **n**: clinical appearance, arrows point typical thoracic lumbar "gibbus" deformity

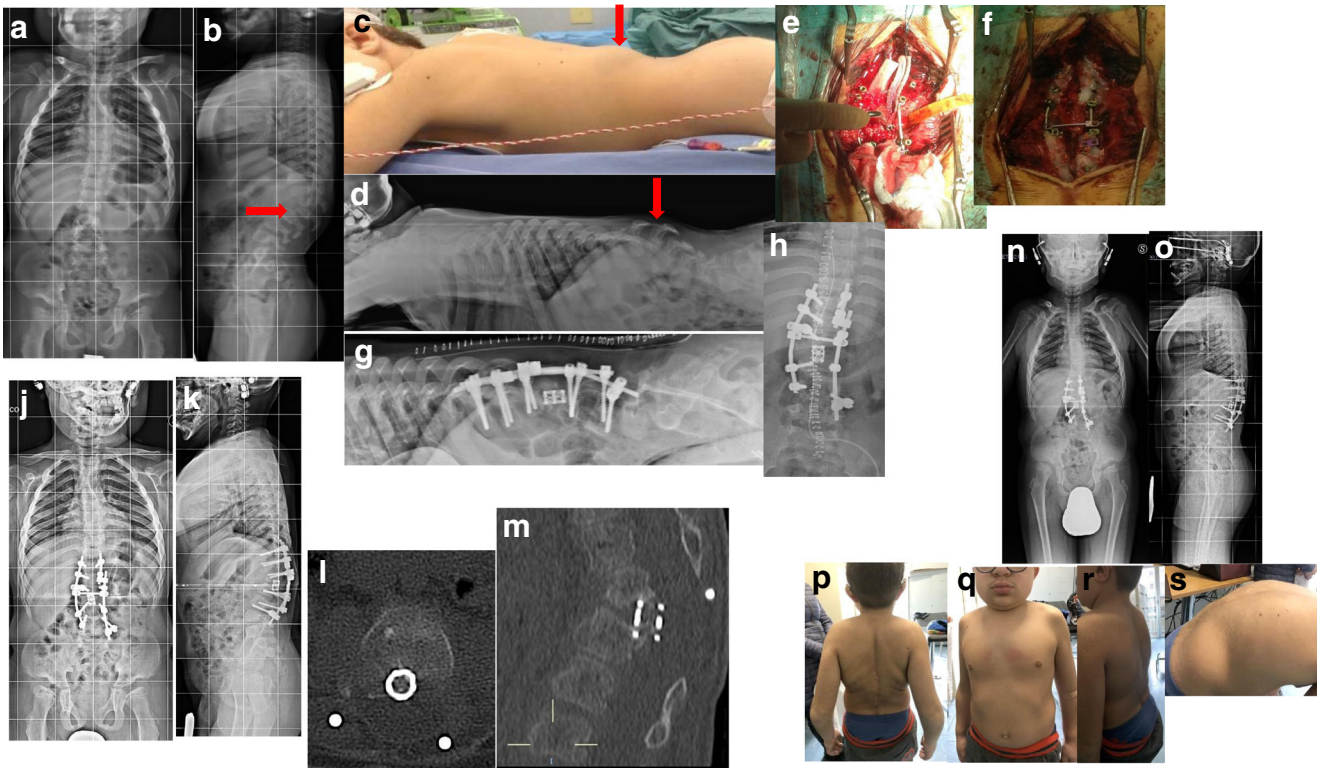

Fig. 4 C.R. 6-year-and-1-month-old **a** and **b** standing radiographs; **c** and **d** thoracic lumbar "hump," kyphosis is fixed and very stiff and does not modify in prone position; **e** and **f** intervention: provisional stabilization rods were mounted and we proceed to complete L2 vertebrectomy by posterior access only: as L2 has been completely removed, the gap between L1 and L3 was filled by cage cut to measure, filled with autologous bone insert obtained from vertebrectomy: **g** definitive rods, adequately contoured to restore thoracolumbar lordosis, were applied obtaining progressive correction. T11-T12-L1 and L3-L4 posterior arthrodesis was performed. The instrumentation was completed by single rod connector at vertebrectomy level, and posterior artrodesis is completed by autologous bone insert, **g** and **h** radiographs after surgery **j** and **k** 24 months follow-up standing radiographs, no loss of correction; **l** and **m** 25 months follow-up CT, CT scan shows complete bone fusion maturation; **n** and **o** 4 years follow-up standing radiographs, stable correction; **p**, **q**, **r**, and **s** 4 years follow-up clinical aspect: optimal cosmetic result, "gibbus" deformity disappeared

developed soft tissue swelling in the occipital area due to reactive bursitis from protruding instrumentation, and need to keep in mind that the only available instrumentation are those for adult patients, do not exist any paediatric device. Bursitis was solved by anti-inflammatory drugs. At 11 years follow-up patient occipito cervical arthrodesis is stable, she has no more problems by protruding instrumentation and her neurologic status is stable, with hardware completely embedded in fusion mass (Fig. 1).

Patient treated by posterior approach vertebrectomy was braced for four months after surgery. Eight months follow-up CT scan showed complete L1-L3 arthrodesis, and another CT scan was performed at 25 months follow-up, showing complete bone-cage osteointegration. At four years follow-up, there was no instrumentation failure and no loss of correction (Fig. 4).

In our patient, critical aspect of operation is cage positioning, as limited vertebras dimensions did not allow an adequate positioning even of the most minimal device available (the smaller adult cervical cage available is 1 cm in diameter, still bigger than patient vertebral plates) resulting in an oblique position of the cage. Even if in sub-optimal position, still the cage filled by autologous bone obtained from morcellized vertebral body allowed a complete interbody fusion, as showed by CT scan at follow-up (Fig. 4). The incomplete kyphosis resolution, with good cosmetic results and stability in the time, in our experience could reduce and avoid the occurrence of proximal junctional kyphosis.

Patient that has been treated by costo-trasversectomy access for interbody vertebral fusion had no complications after surgery, and has been braced by cast for eight weeks and then by brace for other three months. At five months follow-up, CT scan exam shows complete posterior and lateral arthrodesis, and limbs evoked somatosensory potentials come back to normal values. In this patient, obtained good cosmetic and functional results, and slight proximal junctional kyphosis even at one year follow-up (Fig. 5).

Discussion

Cervical instability/stenosis in MPS is a pathology that can require surgical treatment, but certainly thoracic lumbar kyphosis is main treatment issue in MPS patients' spine.

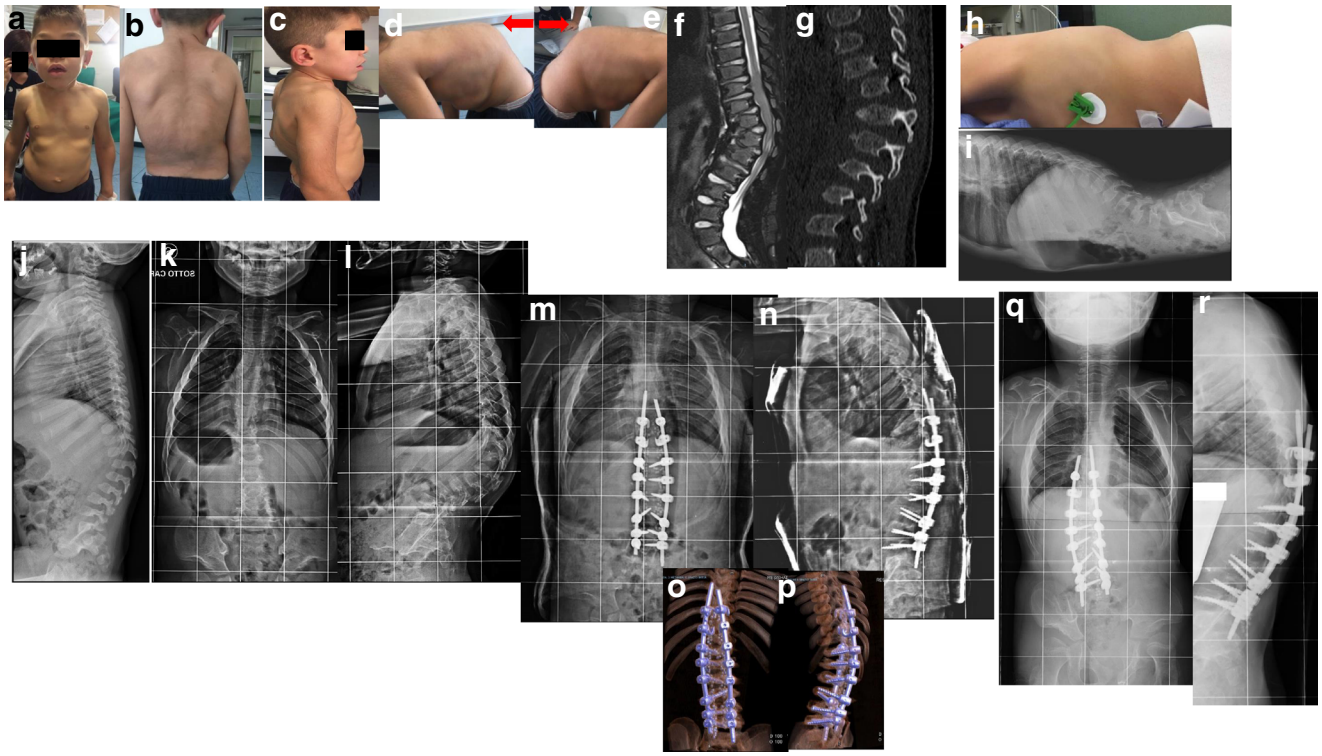

Fig. 5 M.E. A 7-year-and-7-month-old boy affected by MPS I with worsening thoracic lumbar kyphosis, unsuccessfully treated by brace since 4 years of age; **a**, **b**, **c**, **d**, and **e**: clinical appearance with thoracic lumbar "gibbus"; **f** MRI of thoracic lumbar kyphosis showing vertebral canal narrowing with compression on nervous structures: patient had nervous symptoms complaining paresthesia in both legs and showing limbs somatosensory evoked potentials slowed down ; **g** CT scan of thoracic lumbar kyphosis showing vertebral canal narrowing. **h** and **i** kyphosis was partially reducible in extension. **j**: M.E. at 4 years of age, lateral standing radiograph shows 30° Cobb T10-L4 kyphosis, treated by brace. **k** and **l** M.E. 7-years-7-months-old standind anterior posterior and lateral radiographs: he had 70° Cobb T10-L4 kyphosis. **m** and **n** Anterior posterior and lateral standing radiographs after surgery. Patient has been treated by right lateral costal transverse approach, T11-T12, T12-L1, and L1-L2 discectomy; T11-T12, T12-L1, and L1-L2 interbody fusion with autologous bone graft from rib; and posterior instrumented arthrodesis T10-L4; T10-L4 thoracic lumbar kyphosis was reduced to 20° Cobb. **o**, **p** 5 months follow-up CT scan showing complete interbody fusion **q** and **r**: 1 year follow-up anterior posterior and lateral view radiograph; lateral view shows slight proximal junctional kyphosis

Bracing is a viable treatment strategy in flexible kyphosis and can obtain good clinical results at medium terms follow-up even if kyphosis deformity remains in radiographs: our follow-up is still too limited to provide data on deformity evolution in long terms, particularly at time of growth spur.

If deformity worsens despite bracing, and in presence of nervous symptoms, surgery should be considered. In our experience, bracing is less effective for kyphosis over 40° Cobb.

Many surgical technique have been used in the treatment of progressive thoracolumbar kyphosis [37]: in situ fusion [15, 38], anterior stabilization [20, 35, 36], combined anterior, and posterior fusion [17, 20, 37], and recently fusionless VEPTR (vertical expandable prosthetic titanium rib). Bekmez et al. described a complex posterior approach with vertebral osteotomies treating six cases of MPS kyphosis [40].

In situ fusion has been proposed to treat kyphosis under 50°Cobb [39], to create a posterior spinal tether which with anterior vertebral growth causes gradual correction of the gibbus [37]. In MPS patients, anterior vertebral growth is impaired after the first year of life [38, 40], so the grade of correction obtainable with this relatively simple technique is questionable [37]; moreover, the possibility of a solid arthrodesis is impaired by the intrinsic instability due to ligamentous laxity and vertebral dysplasia [37].

Anterior fusion with rib graft [35] without or with associated posterior instrumentation [36] has the disadvantage of anterior approach in patient with comorbidities and, in the cases associated with posterior instrumentation, the procedure is split into two surgical access. In their series, Yasin et al. [20] had five patients treated by anterior fusion without posterior instrumentation; all patients developed deformity adjacent to the operated segment. Yasin et al. [20] treated a single patient by Vertical expandable prosthetic rib (VEPTR) with a rib-pelvis construct. These authors report that VEPTR was effective in correcting sagittal plane deformity distracting the concavity of deformity: patient needed two distraction interventions in a year to adequate instrumentation to trunk development, and the problem of definitive fusion remains. To reduce the morbility related to combined approach, anterior-posterior, as in treatment of congenital deformities, we applied our previous experience on the treatment of

progressive congenital scoliosis, kyphoscoliosis, and kyphosis in young children by posterior approach-only vertebra resection to the problem of thoracolumbar kyphosis in Hurler syndrome. We have used posterior approach-only hemivertebra resection from 2006 [41]. The advantage of posterior approach technique vertebral resection and pedicle instrumentation in young child is obtaining an early correction, before kyphosis progression is so severe to cause possible nervous impairment and extended spine deformity with the necessity of an extended instrumentation.

Our indication is treating thoracolumbar kyphosis over 40° Cobb, previously braced without results.

Even if severe thoracic lumbar kyphosis cannot always be completely corrected, the cosmetic and functional results at follow-up are good, and we believe that incomplete kyphosis correction may be useful in avoiding proximal junction kyphosis phenomena at follow-up. Critical point is the diminutive vertebra dimensions in our patients that cause difficulties in correct positioning of even the smaller cage available; nevertheless, in our patient, we obtained complete fusion at follow-up, due to good osteointegration capability of young patients and to prolonged bracing after surgery.

A different surgical strategy by lateral costal transverse approach avoids the problem of cage positioning and the eventual blood loss from vertebrectomy/vertebral osteotomies.

Conclusions

Spinal involvement is a common feature of some types of MPS; it leads to chronic and progressive diseases that are often major cause of morbidity and mortality for MPS patients. Occipital cervical stenosis and instability can cause severe nervous symptoms requiring surgical decompression and stabilization. Thoracic lumbar kyphosis is a main issue in MPS spine pathology. Bracing is a viable treatment strategy in thoracic lumbar kyphosis and can obtain good clinical results at medium terms follow-up. Surgical treatment is effective in severe evolving cases, but still there are no clear indication on when propose surgical treatment, and which type of surgical intervention is effective. In our experience, main difficulties arose from unavailability of dedicated instrumentation in very young patient (rods and cages), as even smallest devices available are often too big. The choice of posterior only approach surgery, not without technical difficulties, avoid possible complications and comorbidity of combined anterior posterior approach; when correction is possible avoiding osteotomies, this choice avoids the problem of positioning cages that are often too big, even in the smallest dimensions available, and blood loss is diminished too.

Compliance with ethical standards

Competing interests The authors declare that they have no competing interests.

Informed consent Informed consent was obtained from all individual participants included in the study.

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