The aetiology behind torticollis and variable spine defects in patients with Müllerian duct/renal aplasia-cervicothoracic somite dysplasia syndrome: 3D CT scan analysis

Ali Al Kaissi · Rudolf Ganger · Jochen G. Hofstaetter · Klaus Klaushofer · Franz Grill

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Abstract The aim of the article is fourfold; firstly, to detect the aetiology of torticollis in patients with Müllerian duct/renal aplasia-cervicothoracic somite dysplasia syndrome; secondly, spine pathology in Müllerian duct/renal aplasia-cervicothoracic somite dysplasia syndrome varies considerably from one patient to another and there are remarkable differences in severity and localization; thirdly, mismanagement of congenital spine pathology is a frequent cause of morbid/fatal outcome; and fourthly, the application of prophylactic surgical treatment to balance the growth of the spine at an early stage is mandatory. Reformatted CT scans helped in exploring the craniocervical and the entire spine in these patients. The reason behind torticollis ranged between aplasia of the posterior arch of the atlas, assimilation of the atlas and extensive fusion of the lower cervical vertebrae (bilateral failure of segmentation) in four patients; in one patient, in addition to the hypoplastic posterior arch of the atlas, we observed ossification of the anterior and the posterior longitudinal spinal ligaments giving rise to a block vertebrae-like suggestive of early senile ankylosing vertebral hyperostosis (Forestier disease). Scoliosis at different spine levels was

attributable to variable spine defects. Pelvic ultrasound showed the classical renal agenesis in four patients; whereas in one patient, the MRI showed pelvic cake kidney (renal fused ectopia) associated with ovarian, uterine and vaginal abnormalities. This is the first exploratory study on the craniocervical and the entire spine in a group of patients with MURCS association.

Keywords MURCS association · Torticollis · Failure of segmentation · Forestier disease · Posterior spine ankylosis · CT scan

Introduction

A number of syndromes are associated with spine pathology, one of the most notable being MURCS (Müllerian duct/renal aplasia-cervicothoracic somite dysplasia), VATER (Vertebral defects, Anal atresia or stenosis, Tracheo-Esophageal fistula, Radial defects and Renal anomalies, and VACTERL (Vertebral defects, Anal atresia or stenosis, Tracheo-Esophageal fistula, Renal anomalies, Cardiac defects and limb defects) associations [1–3].

To delineate the aetiology behind torticollis we explored the craniocervical and cervical spine regions via CT scanning. There were variable reasons behind torticollis ranging from hypoplasia of the posterior arch of the atlas, assimilation of the atlas and bilateral failure of segmentation of the lower cervical and upper thoracic vertebrae [4–9].

Scoliosis was found to be correlated with variable spine abnormalities such as extensive malsegmentation of the thoracic vertebrae, diffuse fusion of the posterior spine elements and a ‘pebble beach’ sign (smooth outlines of the vertebral bodies) [10].

A. Al Kaissi (✉) · J. G. Hofstaetter · K. Klaushofer
4th Medical Department, Ludwig Boltzmann
Institute of Osteology at the Hanusch Hospital of WGKK,
AUVA Trauma Centre Meidling, Hanusch Hospital,
Heinrich Collin Str. 30, 1140 Vienna, Austria
e-mail: ali.alkaissi@osteologie.at

A. Al Kaissi · R. Ganger · F. Grill
Paediatric Department, Orthopaedic Hospital of Speising,
Vienna, Austria

J. G. Hofstaetter
Department of Orthopaedic Surgery, Vienna General Hospital,
Medical University of Vienna, Vienna, Austria

All our MURCS patients demonstrated the classical renal agenesis associated with ovarian aplasia, uterine hypoplasia and a rudimentary vagina; however, the MRI imaging showed unusual fusion of both kidneys (pelvic cake kidney) in one patient.

Methods

The study protocol was approved by the Medical University of Vienna (Ethics Committee, EK Nr: 921/2009) and informed consents were obtained from the patients' guardians. Five patients of different ethnic origins were ascertained through the Osteogenetic Department (Orthopaedic Hospital, Speising, Vienna, Austria). We studied the natural history of these patients and found that variable spine deformities were present. All patients presented with torticollis; however, the degree of scoliosis varied. A clinical dysmorphologist experienced in phenotypic and radiographic characterisation, a paediatric orthopaedic surgeon experienced in diagnosing congenital orthopaedic abnormalities, as well as a paediatrician examined all the patients thoroughly. The patients who underwent the routine conventional radiographic documentation and pelvic ultrasound showed normal development of breast and external genitalia. Endocrine investigations showed normal serum levels of luteinizing hormone, follicle stimulating hormone, prolactin, estradiol and progesterone hormone. The karyotyping of the patients and their parents was normal. Metabolic parameters to assess blood glucose, uric acid, serum calcium, phosphorus and parathyroid hormone levels were normal. All patients underwent musculoskeletal and neurological evaluations, which included muscle strength, sensations, and the reflexes were normal. We subdivided the patients into two categories in accordance with the most prominent pathology.

Torticollis

Torticollis was present in all our patients; physical examination showed fixed torticollis with rotation of the head to the right associated with flexion of the neck. When they attempted to turn their heads, the patients were unable to pass the midline. There was no associated muscle spasm; gentle passive movements failed to correct the torticollis. During sleep, the torticollis remained unchanged. There was no associated radiculopathy or myelopathy on clinical examination. Initial radiographic evaluation showed an ill-defined occipito-atlanto-axial malformation complex. Torticollis was present from birth in three patients, in one patient before the age of nine, and one adult patient was referred because of amenorrhea. In the older patients, headache and suboccipital pain were the main complaints.

Developmental anatomy of the atlanto-axial joints

All typical vertebrae, except for the atypical atlas and axis, ossify from three ossification centres. There is a gradual union from these three ossification centres. There is a gradual union of the primary ossification centres of the anterior centrum and posterior arches. Most of the anterior portions of the neural arch contribute to vertebral body formation. The atlas is unique; it appears that the anterior primary ossification centre is present in fewer than 20% of all neonates. The anterior ossification is termed as the intercetraxium-1. The posterior synchondrosis and the anterior neurocentral synchondroses are fused by the time a child is 4 or 5 years old, and the diameter of the atlas canal reaches adult proportions at approximately age 5 or 6. Most of the congenital variants encountered in the cervical spine involve the first two cervical vertebrae. Synchondrosis fractures or dislocations are difficult to appreciate radiographically and contribute to the high diagnostic error rate for cervical spine injury in young children [4–8].

The abnormal cervical vertebral fusion (bilateral failure of segmentation or Klippel–Feil anomaly) causing torticollis and cervical scoliosis was seen in one adult patient. The “hallmark” of this condition is the improper segmentation of cervical motion segments, discernible as congenitally fused vertebrae with or without associated manifestations [9, 10]. In this patient, we searched for adverse outcome of this diffuse bony ankylosis such as fracture above or through a fused segment, cervical spondylotic development, subluxation, bulging of the disc and/or ligamentum flavum, and certainly the loss of cervical alignment. A comprehensive reformatted CT scanning of the craniocervical junction (axial, sagittal, coronal and reconstruction) was performed.

Scoliosis

We encountered variable spine curvatures in our patients. Some presented with vertebral anomalies at birth; in others, the clinical deformity was evident in later childhood. In order to understand the natural history of the variable spine curvatures, it was necessary to correlate the principles of normal growth of the spine with the pathologic anatomy of the various types of congenital vertebral anomalies [8].

Developmental anatomy of the spine

During early embryonic development, somites, which are transient precursors of the axial skeleton, are formed in a process called somitogenesis. Disruptions in somitogenesis cause vertebral malformations, including the formation of uneven segments (hemivertebrae and wedge vertebrae), fused segments (block vertebrae), and vertebrae with

failure of midline fusion (butterfly vertebrae). The normal longitudinal growth of the spine is the sum total growth occurring at the end plates on the upper and lower surfaces of the vertebral bodies. This growth occurs symmetrically; as a result, the spine grows in a balanced manner without a pathologic curvature. However, in the presence of a congenital vertebral anomaly, there is an asymmetric deficiency in either the number of growth plates or their rate of growth on one side of the spine, resulting in a longitudinal growth imbalance and an increasing spinal deformity as the child grows. Scoliosis develops if the defective growth occurs laterally, and kyphosis occurs with defective anterior growth [8, 9]. The radiographic appearance of the vertebral curvatures was inconsistent. We referred to the reformatted CT scan.

Results

Data were obtained from all five patients in this series. The study consisted of five female patients. The mean age of presentation to our clinic was 9 years (range 2–16 years). Refer Table 1.

In this group of MURCS patients, our plan of correction should occur at the atlanto-occipital joint and the rest, in

the lower segments of the cervical spine. When the craniocervical junction is fused, most of the required correction is obtained. The latter should be performed in correlation with careful checking of neurological and vascular status.

Discussion

The MURCS association first described by Duncan et al. [1] is rare and includes non-random association of Müllerian duct aplasia or hypoplasia (MU), renal agenesis or ectopy (R), and cervicothoracic somite dysplasia. Rarely, the MURCS association may be designated for a real malformation because of multiple malformations. The differential diagnosis is with VATER and VACTERL association; however, MURCS association is characterised by Müllerian duct aplasia, which is not a feature in VATER or VACTERL association.

Moreover, in the MURCS association, the vertebral malformations occur consistently in the cervicothoracic region as compared with the anomalous vertebrae of VATER patients, which are more frequently found in the lower thoracic, lumbar and sacrococcygeal spine. Upper limb abnormalities are found in 24% of VATER

Table 1 Radiographic/ scanning and imaging results

Imagings	Patient	Craniocervical/cervical spine	Spine	Extra-spinal
Radiographs	All patients	Ill-defined abnormalities (Fig. 1a, b)	Scoliosis	Unilateral renal aplasia
Sagittal CT scan	9-year-old	Hypoplasia of the atlas, fusion of C2/4 (Forestier disease) (Fig. 2)	–	
Axial reformatted CT scan	5-year-old	Hypoplastic anterior arch of atlas and assimilation of the posterior arch (Fig. 3)	–	
Sagittal reformatted CT scan	5-year-old	Anterior displacement of odontoid process in relation to the posterior aspect of the anterior arch of C1 (Fig. 4)	–	
3D reconstruction CT scan	14-year-old	Bilateral failure of segmentation (Fig. 5)	–	
3D coronal CT	2-year-old	–	‘Pebble beach’-like sign (Fig. 6)	
Sagittal reformatted CT	16-year-old		Diffuse ankylosis of the posterior spine elements (Fig. 7)	
Anteroposterior spine radiograph	2-year-old		VEPTRL operation (Fig. 8)	
Pelvic MRI	2-year-old			Pelvic cake kidney, ovarian agenesis, uterine hypoplasia and a rudimentary vagina (Fig. 9)

Fig. 1 **a** Anteroposterior upper cervical radiograph shows torticollis (*arrow*) and ill-defined craniocervical abnormality; **b** lateral skull radiograph shows hypoplasia of the atlas and assimilation of the posterior arch (*arrow*)

Fig. 2 Sagittal reformatted CT scan of the upper cervical spine shows severe hypoplasia of the posterior arch of the atlas (*arrowhead*) associated with subsequent fusion of the C2/4 because of ossification of the anterior and posterior longitudinal ligaments, respectively (this is not a true block vertebrae, note the preservation of the intervertebral disc space), this is suggestive of early senile ankylosing vertebral hyperostosis (Forestier disease, *arrow*)

association patients, but are unusual in the MURCS association [1–3].

Torticollis is often a clinical manifestation of a congenital osseous cervical spine anomaly. Tumours of the posterior fossa, infection and cervicothoracic scoliosis also may result in torticollis. The deformity is characterised by lateral tilt of the head with rotation of the face. Often, the deformity is mild and is unconsciously compensated for by the patient. Facial asymmetry, which may be permanent, can occur in longstanding untreated patients. Hemiatlas may cause a rather severe and progressive torticollis when the child is

Fig. 3 Axial reformatted CT scan shows a combination of anomalies of remnants of a hypoplastic anterior arch (*arrowhead*) associated with aplasia and assimilation (fusion with the occiput) of the posterior arch of the atlas (*arrows*)

young; the neck, despite the deformity, is flexible and the torticollis can be passively corrected in some patients. However, in some patients, it becomes increasingly severe and fixed [9–11]. The underlying cause of torticollis is variable, and often uncertain. In a study of congenital muscular torticollis by Yu et al. [12], the aetiology of torticollis was caused by idiopathic fibrosis of the sternocleidomastoid muscle that restricted movement and pulled the head towards the involved side. On 14 patients (aged 1 month to 24 years), tomographic studies, using skull images that were rotated into a standard orientation and then reconfigured for evaluation of the cranium, endocranial base and facial skeletal structures, showed that the cranium and cranial base deformation took place early in infancy, with the most prominent change occurring in the posterior cranial fossa. Apart from the idiopathic fibrosis of the sternocleidomastoid, there was no other underlying cause.

Fig. 4 Sagittal reformatted CT image reveals anterior displacement of the odontoid process (O) in relation to the posterior aspect of the anterior arch of C1. Note the Wackenheim clivus line* (a), which is drawn along the posterior aspect of the clivus toward the odontoid process; in our patient, the line does not intersect and is not tangential to the odontoid process, which confirms the abnormality and indicates the existence of occipito-atlas subluxation. The Chamberlain's line** (b) shows no associated basilar invagination

Fig. 5 3D reformatted CT scan shows torticollis (white arrow). Black arrow shows significant failure of segmentation extending from C3 and involved the rest of the cervical spine (bilateral failure of segmentation). A bilateral failure of segmentation in a number of adjacent cervical vertebrae resulted in the development in block vertebrae. The vertebrae appear as narrow and fused. As a result, longitudinal growth is impaired on both sides of the spine, producing a shortened segment. This is known as Klippel–Feil anomaly

Fig. 6 3D reformatted CT scanning showed massive segmentation defects throughout the lower cervical and the upper thoracic vertebral column (black arrow), this was accompanied with smooth outlines to the vertebral bodies, resembling 'pebble beach' sign (white arrows). 'Pebble beach' sign is a radiographic feature usually encountered in patients with spondylocostal dysostosis (SCD). However, in SCD, the malsegmentation involves the entire spine and not a portion as seen in patients with MURCS

Assimilation of the atlas, also known as occipitalisation of the atlas, is partial or total fusion of the atlas with the occiput. Anomalies at the occipitocervical junction are commonly seen in patients with achondroplasia, diastrophic dysplasia, spondyloepiphyseal dysplasia, Larsen syndrome and Morquio syndrome [13].

Occipitalisation of the atlas is also seen in conjunction with fusion of the second and third cervical vertebrae, and this pattern may predispose the atlanto-axial joint to abnormal stress, premature degeneration and potential instability. McRae and Barnum [14] described 25 patients with this condition and found that the fusion of the anterior

Fig. 7 Sagittal reformatted CT scan of the thoracolumbar spine shows marked malsegmentation of the vertebral bodies anteriorly (*arrowhead*) associated with diffuse ankylosis of the posterior spine elements (*arrows*)

Fig. 8 Surgical intervention has been performed via (vertical expandable prosthetic titanium rib (VEPTR) operation) on the left side (thorax- 2-rib-rib- implantation). The fusion has been parted between third and fourth rib, then one cradle end half on the first and the other on the tenth rib was performed (as far medial as possible). Strict monitoring of the spinal cord with careful distraction and implantation of the vertical part has been done

Fig. 9 Coronal pelvic magnetic resonance imaging showed pelvic cake kidney (ectopic fused kidney *white arrow*); ovarian agenesis, uterine hypoplasia and a rudimentary vagina

aspect of the complex was more common than that of the posterior elements. The posterior elements of the atlas may be hypoplastic and anomalous, causing posterior compression. Of the 25 patients in the report of McRae and Barnum, 18 (72%) reported neurological problems, such as weakness, numbness, or pain in the upper extremities or dull headaches. Of these 18 patients, 16 had long-tract signs indicative of pyramidal compression. Wackenheim [15] reported that instability at this level might be caused by a deficiency of the transverse atlantal ligament. Respiratory and autonomic dysfunction has been reported in patients with this condition. All patients were identified as having isolated non-syndromic occipitalisation of the atlas [16]. Hemiplasia or hypoplasia of the atlas can cause a marked torticollis, which if untreated, constitutes an unacceptable deformity. Dubousset [17] described three forms of the anomaly.

Type I is characterised by an isolated hemiatlas; type II, by partial or complete aplasia of one hemiatlas with congenital fusions in the cervical spine; and type III, by partial or complete occipito-atlantal fusion and symmetrical or asymmetrical hemiatlas aplasia with or without anomalies of the odontoid or other cervical vertebrae. Syndromic associations were not part of the Dubousset study.

Congenital aplasia of the posterior arch of the atlas has been described as a primary cause of sensory deficit. Klimo et al. [18] described a male patient with a solitary presentation of congenital partial aplasia of the posterior arch of the atlas who developed acute sensory deficits in his distal lower extremities owing to a mobile isolated bone

fragment caused by repeated trauma by the bone fragment to the dorsal spinal cord with neck extension. Forestier and Rotes-Querol in 1950 [19] described senile ankylosing vertebral hyperostosis in patients over 40 years of age. It is characterised by an ossifying, non-inflammatory, non-erosive enthesopathy favouring the dorsal spine but sparing the sacroiliac joints. Forestier disease leads to acquired narrowing of the spinal canal due to the presence of osteophytes, which sometimes create bony hooks within the spinal canal. Some authors have noted the occasional familial incidence of vertebral hyperostosis, leading to a suspicion of genetic predisposition in the Japanese elderly population. It is empirical to distinguish congenital vertebral blockage from other causations of spinal fusion [20–22].

Al Kaissi et al. [23] described a 17-year-old girl who presented with MURCS association associated with significant thoracic vertebral hyperostosis closely resembling senile ankylosing vertebral hyperostosis (Forestier disease).

Patients with Klippel–Feil syndrome may have pterygium colli, a condition characterised by various degrees of webbing of the skin from the sides of the neck to the shoulders. When pterygium colli is bilateral, it gives the appearance of a short neck; when unilateral, it can give the appearance of a torticollis. Klippel–Feil anomaly occurring elsewhere in the spine rather than the cervical spine is diagnosed less frequently [9].

Hensinger et al. [24] noted such a high prevalence of Klippel–Feil syndrome in patients with congenital cervicothoracic scoliosis that they recommended making radiographs of the cervical spine for such patients to screen for the syndrome. Trauma to an individual with Klippel–Feil anomaly can be catastrophic [25].

‘Pebble beach’ sign is a radiological feature usually seen in patients with spondylocostal dysostosis syndrome (SCD). SCD syndrome, however, is characterised by failure of segmentation of the *entire spine* and not associated with extra-spinal malformation complex such as Müllerian duct aplasia as seen in MURCS syndrome [10].

Congenital anomalies of the kidneys include a group of so-called fusion anomalies, in which both kidneys are fused together in early embryonic life. Fusion anomalies of the kidneys can generally be placed into two categories [1], horseshoe kidney and its variants and [2] pelvic cake kidney (fused ectopia). Horseshoe kidney is probably the most common fusion anomaly. The term horseshoe kidney refers to the appearance of the fused kidney, which results from fusion at one pole. The total incidence of lump kidneys is estimated to be between 1/13,000 and 1/7,500. The development of the normal kidney depends on the union of ureteric buds from the mesonephric ducts with the nephrogenic cords in the embryo. The union is believed to occur around the fourth gestational week, normally occurring at

the level of the first or second sacral vertebral segment. Subsequent straightening of the hind end of the embryo, along with differential growth of the developing pelvic structures, leads to the ascent of both kidneys to their normal dorsolumbar regions between the fourth and ninth weeks of gestation. The abnormal fusion probably occurs at the 5- to 12-mm embryonic stage, when the kidneys are in the true pelvis and the renal capsule has not yet matured. One embryologic explanation regarding midline fusion is that, at that stage, abnormal variation in growth, ventral flexion of the hind end of the embryo, or other variations in the growth of pelvic structures may bring the metanephric blastemas (developing kidneys) abnormally close together for a longer period; this proximity can lead to fusion. If the kidney fails to ascend, it remains unrotated (pelvic kidney or a pancake kidney because of its flattened shape). Such kidneys often maintain their foetal blood supply from the iliac vessels or the distal aorta [26].

It is not unusual to see the combinations of spinal anomalies which could result from a failure of orderly resegmentation of the sclerotomes and the frequent association of kidney anomalies with congenital scoliosis can be explained by the fact that the lateral portion of the same paraxial mesodermal cells that from the somites forms the mesonephros [24].

MacEwen et al. [27] revealed a 20% incidence of urinary tract anomalies detected on routine intravenous pyelography, whereas a study by Hensinger et al. on cervical spine anomalies found a higher rate of 33%. Many of these interesting anomalies, such as the presence of a single kidney, duplicate ureters or crossed renal ectopia, were not potentially dangerous. The life expectancy in patients with a pancake kidney is not different from the normal population without associated complications [28, 29]. None of the above-mentioned reports of renal agenesis and/or pelvic cake kidney is part of a syndromic complex such as MURCS.

Conclusion

For a patient presenting with spine pathology, the initial procedure is to be aware of the specific diagnosis of a suspected syndromic association. It is quite common that orthopaedic patients with unusual phenotypic features will be followed for years with an “undiagnosed syndrome”. When these patients are to undergo major surgical interventions, it is important to know what sort of medical problems may arise both during and after surgery as well as, and for any unique pitfalls to be encountered such as undiagnosed aortic and or cerebral aneurysm, bleeding problems, immunity deficits, cervical spine stenosis, cervical spine instability and or difficulties in intubation

because of blocked cervical vertebrae. Without a definite clinical diagnosis of a suspected syndrome, it is impossible to plan a preoperative strategy for a major spine surgery for minimising the risk of increased morbidity and mortality due to malformation syndromic association.

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