

Congenital defect of craniovertebral junction in twins with Kniest dysplasia – case report

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Abstract

Kniest dysplasia is a rare disease from the group of osteochondrodysplasias, rated type II collagenopathies, and like other skeletal dysplasias may be associated with defects of cranio-vertebral junction with atlanto-axial instability. Objects: a case of spontaneous atlanto-axial subluxation with spinal canal narrowing at the level of craniovertebral junction in twins with Kniest dysplasia was presented. Methods: on the basis of clinical examination, radiographs and MRI study, children were qualified for operative treatment - posterior widening of the foramen magnum and occipito-cervical fusion with titanium anchors. During control examination 4 years after primary surgery, patients was very active, without any neurological deficits, x-ray pictures and MRI study revealed widening of the foramen magnum and full fusion in the place of surgery. Conclusion: authors suggestion is that in the case of foramen magnum and spinal canal narrowing, operative treatment with decompression of nervous structures and stabilization should be advised.

Key words: osteochondrodysplasia, congenital atlanto-axial subluxation, spinal stenosis, cervical spine.

INTRODUCTION

Dysplasia, described by Kniest [7] in 1952, is a rare disease from the group of osteochondrodysplasias, rated type II collagenopathies. It may be caused by over ten different mutations in COL2A1 gene [1,14,15]. Patients with Kniest dysplasia have the following clinical features: disproportionate dwarfism, with a big, round skull over a short neck, a flat face and midface hypoplasia. Clubfoot deformity and kyphoscoliosis may also occur. Hearing loss, cleft palate, myopia and retinal detachment are parts of typical phenotype [10].

Kniest dysplasia, like other skeletal dysplasias – achondroplasia, spondyloepiphyseal dysplasia and diastrophic dysplasia may be associated with defects of cranio-vertebral junction with atlanto-axial instability. This results by inborn in odontoid hypoplasia, ligament laxity, narrowing of the foramen magnum and malformation of the base of the skull (platybasia). Imaging techniques often also reveal a dense fibrotic epidural band, that compresses the spine and decreases the fluid reserve of the spinal canal.

The goal of the work is a case presentation of spontaneous atlanto-axial subluxation with spinal canal narrowing at the level of craniovertebral junction in twins with Kniest dysplasia and the description of applied treatment.

CASE REPORT

Patients M.K. and A.K who were admitted to our department for the first time at the age of 6, are female homozygotic twins from a fourth pregnancy. The first and second pregnancy ended with miscarriages, a third pregnancy ended with the delivery of a healthy girl.

Just after birth, due to visible disproportionate disturbances of body build, the girls underwent pediatric and genetic examinations which resulted in a diagnosis of Kniest dysplasia.

CASE 1 – PATIENT A.K.

Five months prior to admission to hospital she fell out of bed with flexion-like head injury in form. It was followed by total flaccid quadriplegia with quick spontaneous partial resolution and persistent quadriparesis. Occasional sleep apnea also was observed. On admission to hospital we noted typical phenotype of Kniest dysplasia, disproportionate dwarfism with short trunk, and a growth deficiency that equaled –7.1SD in comparison with normal age and sex, also slight overweight – weight to height ratio was +28%. Symptoms included: waddling gate with lumbar hyperlordosis, limited spine motion and 20° varus deformity of both tibia.

During neurological examination bilateral ankle clonus was noted, especially on the right leg with increased muscle tone. Periosteal reflexes were asymmetric, clonic on the right side. Somatosensory evoked potentials suggested partial damage of the ascending tracts of the spinal cord.

Imaging techniques (radiograms, CT and MRI) showed congenital defects of the craniovertebral junction and cervical spine (Fig.1, Fig.2, Fig.3). The odontoid was deformed and placed near the anterior arch of C1, the vertebral body of C2 was displaced posteriorly. The skull and atlas were transposed anteriorly (anterior unstable subluxation relative to the C1 vertebra). The foramen magnum was narrowed to a distance of 7-8mm in the sagittal plane. The dural sac was compressed and modeled on the posterior edge of the odontoid. The spinal cord was compressed, with areas of gliosis. In the upper cervical spine (C1-C4) was schisis of arches and bulging of intravertebral discs. On the basis of clinical examination, radiographs and MRI study, the child was qualified for operative treatment. A posterior widening of the foramen magnum was performed, releasing the spine compressed by the fibrotic band. Intraoperatively we noted a huge defect of C1

arch and schisis of C2-C7 arches. Posterior occipito-cervical fusion with titanium anchors was done as well (fig.4, fig.5). Operation time was 2h25min. After proper wound healing, the patient left hospital on the 13th day after surgery. She was advised to wear a latex Schanz collar. Thirteen months after the operation, after minor trauma, occipital anchors were displaced. The radiological and ultrasound signs of fusion allowed us to remove the implants. During a control examination, 4 years after primary surgery, the girl was very active, without any neurological deficits. She was only 5cm taller than before her treatment. MRI study revealed widening of the foramen magnum –in the sagittal plane distance between the bony edges of the foramen was 12mm (Fig.6, Fig.7).

CASE 2 – PATIENT M.K.

At the admission to hospital we noted typical phenotype of Kniest dysplasia, disproportionate dwarfism with a short trunk, and growth deficiency that equaled -7.1SD in comparison with normal age and sex, and the weight to height ratio was +21%. Symptoms included: waddling gate with lumbar hyperlordosis, limited spine



Fig. 1. Patient A.K. Radiogram before treatment (lateral view).

Fig. 2. Patient A.K. CT scan before treatment (3D reconstruction).

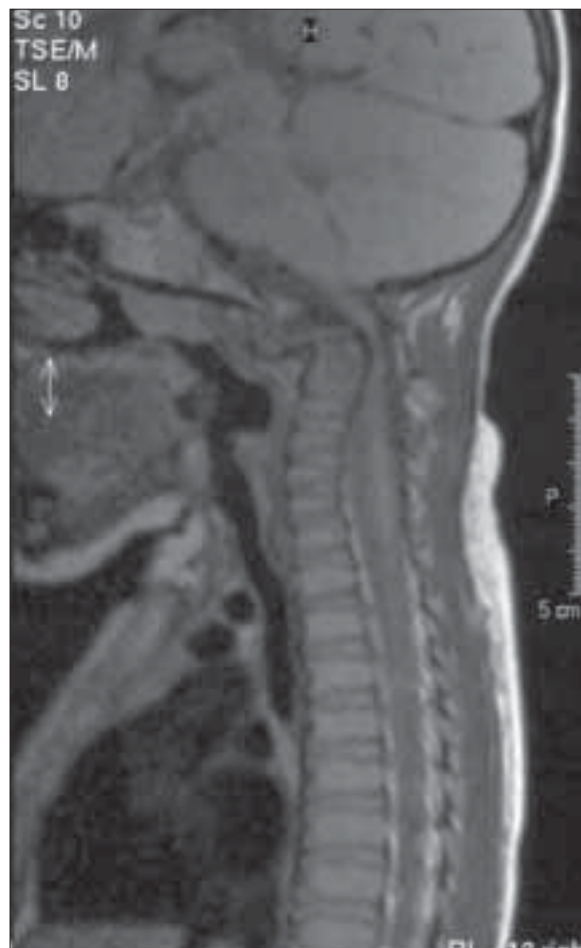
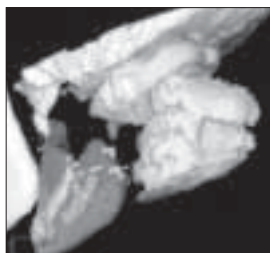


Fig. 3. Patient A.K. MRI image before treatment.

motion and 20° varus deformity of both tibia. In the neurological examination we did not state neurological deficits. Somatosensory evoked potentials suggested partial damage of spinal cord ascending tracts. Imaging techniques (radiograms, MRI) showed congenital defects of the craniovertebral junction and upper cervical spine. The odontoid was deformed and placed near the anterior arch of C1, the vertebral body of C2 was displaced posteriorly. The skull and atlas were transposed anteriorly (anterior unstable subluxation relative to the C1 vertebra). The foramen magnum was narrowed to a distance of 8mm in the sagittal plane. The

dural sac was compressed and remodelled on the posterior edge of the odontoid. The anterior reserve of cerebro-spinal fluid was decreased. In the upper cervical spine (C1-C4) was schisis of arches and bulging of intravertebral discs. On the basis of clinical examination, radiographs and MRI examination, taking into account the risk of probable future neurological deficits, the child was qualified for operative treatment. A posterior widening of the foramen magnum was performed, with occipito-cervical fusion with titanium anchors. Intraoperatively we noted a huge defect of C1 arch and schisis of C2-C7 arches. Operation time was



Fig. 4. Patient A.K. Radiogram after surgery (lateral view).



Fig. 6. Patient A.K. Radiogram 2 years after surgery (lateral view).



Fig. 5. Patients A.K – titanium implants in operation site.



Fig. 7. Patient A.K. MRI image after follow-up.

2h15min. After proper wound healing the patient left hospital on the 13th day after surgery. She was advised to wear a latex Schanz collar. Thirteen months after the operation, during the control examination, we found radiological and ultrasound signs of fusion that allowed us to remove the implants. During a control examination 4 years after primary surgery, the girl was very active, without any neurological deficits. She was only 5cm taller than before her treatment. The MRI study revealed widening of the foramen magnum –in the sagittal plane the distance between the bony edges of foramen was 18mm.

DISCUSSION

In Kniest dysplasia, defects of craniovertebral junction are the second most frequent spine deformities after kyphoscoliosis. Their morphology is similar to those in achondroplasia. Initially, such anomalies may be symptomless, but they gradually lead to quadriplegia and sleep apnea, like at our first patient. Extremely important is then early recognition in children with different osteochondrodysplasias, even in the cases where there is a lack of clinical symptoms [6,9].

The most frequent anomalies in the region of craniovertebral junction are: narrowing of the foramen magnum with thickening of its posterior rim, the presence of dense fibrotic epidural band [6], and/or narrowing of the spinal canal in the upper cervical area [4] and atlanto-axial instability. Gordon [5] describes risk signs

that point out the necessity of operative treatment: brisk reflexes, small foramen magnum, central hypopnea. In these cases, the following operative techniques are used: removal of dens with anterior or posterior fusion, posterior cord decompression with fusion. However, most of the authors state, that the best, required treatment is an operative decompression of cranio-vertebral junction through widening of the foramen magnum and removal of the C1 posterior arch [4, 11]. Sometimes removal of fibrotic band is necessary [11]. We think, that when primary instability exists, and cord decompression is then performed, additional internal stabilization with implants is crucial [4]. Craniobvertebral stabilization is not a simple procedure, due to stable occipital elements – that is why numerous solutions of this problem exist: from wires to hooks and screws [2, 3,12]. Due to the great stability of implants and small diameters, we applied stabilization with occipital anchors, similar to hooks described by Sandhu [8,12,13]. Despite implant displacement out of bone, this stabilization plays its role until fusion occurs.

Additionally, in the case of such dysplasia, minimal increase of height does not cause the risk of cervical hiperlordosis. In summary, we think that in children with Kniest dysplasia early recognition of cranio-vertebral disorders and myelomalacia is justified. In the case of foramen magnum and spinal canal narrowing, operative treatment is necessary: decompression of nervous structures with occipito-vertebral stabilization and fusion.

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