

ATLANTOAXIAL INSTABILITY ASSOCIATED WITH C2 ODONTOID ABNORMALITY IN PATIENTS WITH DOWN SYNDROME: DESCRIPTION OF TWO CLINICAL CASES WITH THE AUTHOR'S MODIFICATION OF STABILIZATION

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Objective. To present two clinical cases of successful surgical treatment of atlantoaxial instability in Down syndrome patients with os odontoideum.

Material and Methods. The results of surgical treatment of atlantoaxial instability in two Down syndrome patients with os odontoideum were analyzed. The neuroorthopedic status of the craniovertebral junction was evaluated perioperatively according to CT and MRI (ADI, Swischuk test, Power's ratio). The assessment of the patients' quality of life was carried out using the Y.A. Orlov scales.

Results. The presented clinical cases demonstrate the results of reconstructive and stabilizing correction of C1-C2 instability using the Goel - Shah method combined with original patented technique.

Conclusion. The use of the Goel - Shah method in combination with remodeling of the C1-C2 articular facets (original patented technique) provides optimal correction of instability in C2 anomalies in patients with Down syndrome.

Key Words: Down syndrome; Os odontoideum; atlantoaxial instability; craniovertebral junction; atlantoaxial fixation; cervicomedullary decompression.

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One of the main reasons for atlanto-axial instability in patients with Down syndrome (DS) is os odontoideum, an anomaly that can be asymptomatic but often causes atlantoaxial dislocation and spinal cord compression [1–3].

The prevalence of os odontoideum in patients with DS is 1.3–3.0 % and is mostly found in childhood and adolescence, less frequently in adults [1, 4]. To date, the predisposing factors leading to the os odontoideum have not been identified. Nevertheless, some authors report individual anatomical and physiological features of patients with DS, focusing on low bone mineral density, weakness of transverse ligaments, poor muscle tone, and over-mobility of joints [1, 3].

The primary objective of surgical treatment of atlantoaxial instability, including that associated with the

os odontoideum, is to eliminate spinal cord compression to prevent the development of disabling neurological deficit [4, 5]. Failure of stabilization of the C1–C2 segment at the level of the C2 os odontoideum is manifested by varying degrees of marked instability of the atlantoaxial joints with combined anterior and posterior compression of the spinal cord. The posterior compression factor is the posterior arch of C1, which stenoses the spinal canal during anterior dislocation of the atlas. Anterior spinal cord compression originates at the level of the posterosuperior margin of the C2 body. It is associated with deformity of the spinal canal because of anterior translation and the C1 inclination relative to the C2 vertebra. Therefore, the objective of spinal cord decompression in atlantoaxial instability is closely related to the need to correct the existing deformity and provide stable fixation of the segment in a position close to the physiological standard.

Nowadays, a sufficient number of surgical techniques are used for decompression and fixation in instability of the C1-C2 segment, ranging in scope of surgery, injuries, and potential complications [4, 5]. The use of short monosegmental fixations is a universally recognized trend. The most common is the posterior screw fixation (Goel-Harm's technique). However, the variety of anatomical features involved in craniovertebral malformations, combined with the frequent occurrence of this pathology in children, does not allow us to ignore the numerous variants of craniocervical fixation, including in combination with prior halo-correction.

In this article, we present examples of surgical treatment of C1–C2 vertebral instability in patients with os odontoideum associated with DS using the modified Goel-Shah's technique [6].

The objective is to present two clinical cases of successful surgical treatment of atlantoaxial instability in Down syndrome patients with os odontoideum.

Material and Methods

The modified Goel-Shah's technique of surgical correction of atlantoaxial instability was used in both described clinical cases. Autogenous tricortical grafts from the iliac crest were used as spacers. The choice of the material was determined by the peculiarities of implant use in children because of the small size of the articular surfaces. Remodeling of the previously dehyalinized articular facets was performed by resection of the posterior third of the articular facets of the C2 vertebra and the anterior third of the lower articular facets of the C1 vertebra using a high-speed diamond bur and Kerrison rongeur [7].

Results

Clinical case 1

A girl with DS, 3 years old, the disease onset started with pain in the cervicooccipital area and tilting of the head to the left. The clinical manifestations progressed within one year, worsening in the form of increasing muscle weakness in the extremities to severe spastic quadriparesis. According to MRI data during examination, atlantoaxial dislocation, instability at the level of C1-C2 vertebrae, spinal canal stenosis up to 2 mm with spinal cord compression, and formation of a myelopathic MR-signal focus at the level of the C1 vertebra were detected. The clinical picture of the child upon admission to the hospital includes pain in the cervico-occipital area. The neurological status shows spastic quadriparesis (right: up to 4 points; left: up to 3 points), a pathological plantar reflexes: Babinski's and Puusepp's signs on both sides. Developmental features:

hypertelorism, epicanthus, macroglossia, and foot clonus on both sides. The patient's quality of life was 10 points according to the adapted Yu.A. Orlov scale, which corresponded to an unsatisfactory level [8].

Cervical MRI scan dated November 09, 2021: atlantodental interval was more than 4 mm; a myelomalacia focus was verified in the center of the spinal cord at the level of the C1 vertebra.

Cervical CT scan dated November 10, 2021: atlantoaxial dislocation associated with the C2 os odontoideum. The dislocation of the C1 vertebra to the front from the anterior margin of the C2 body was 6 mm, the Swischuk test was 2.73 mm, and the Power index was <1.

It was decided to perform reconstructive and stabilization surgery for correction of atlantoaxial instability, decompression of the spinal cord at the upper cervical level, corrective facetectomy of C1–C2, artificial ankylosis of the atlantoaxial joints with autogenous bone grafts, and posterior screw fixation of C1–C2.

Course of surgery. Pronate position of the patient, head fixed in the Mayfield skull clamp in a neutral position. A posterior midsection of the skin and subcutaneous tissue was made from the external occipital protuberance to the C5 spinous process. The nuchal ligament was dissected, and the squama occipitalis, posterior C1 arch, spinous process, and C2 vertebral arches were skeletonized. There was an anterior translation of the C1 vertebra relative to C2. The venous plexus in the C1-C2 interval was mobilized subcutaneously, gradually coagulated, and transected together with the C2 nerves. The C1-C2 joint capsules were opened, and anterior tilt of the articular facets with dislocation of the lateral masses of C1 to the front was noted. The articular surfaces were dehyalinized. The posterior margin of the upper articular facets of the C2 vertebra were resected using a high-speed bur. The anterior margin of the C1 lower articular facets were resected in a similar way. The correction of the anterior translation of C1 was performed under neurophysiological control by repositioning the patient's head in the Mayfield skull clamp. Autogenous tricortical grafts obtained from the crest of the patient's iliac bone with the dimensions of $10 \times 10 \times 7$ mm were placed in the intra-articular spaces of C1-C2 under moderate distraction. Polyaxial screws of 20.0×3.5 mm were placed into the lateral masses of the C1 and transpedicular to the C2 vertebral body under image intensifier tube control. The structure was mounted on 3.5 mm beams. The adjacent surfaces of the C1 and C2 arches were decorticated, and autogenous cortical powder was placed between them at the level of the structure. Hemostasis was competent at an arterial pressure of 120/80 mmHg. The layered closure of the wound without drainage.

The early postoperative period was unremarkable. The patient was verticalized on the day 2 after surgery in a rigid cervical collar. Neurological status was without increasing focal symptoms and long-tract signs. The patient was discharged on the day 10 with regression of pain syndrome (Fig. 1).

Clinical case 2

A 9-year-old girl with DS was followed for a long time by an orthopedist at her place of residence for deformities of the lower extremities; MSCT scan revealed the C2 os odontoideum and C1-C2 instability. MRI scan of the cervical spine showed stenosis of the spinal canal at the level of the apex of the C2 os odontoideum up to 5 mm and spinal cord compression by the apex of the C2 os odontoideum. The clinical picture on admission to the hospital was presented by a defense attitude of the head with a tilt to the right, limitation of movements in the cervical spine, and pain in the cervical-occipital area, intensified by head movement. Neurological status: reflex quadriparesis, constant stereotyped compulsive athetoid movements, macroglossia, and genu valgum are noted. No strength paresis or sensation disorders were detected. The patient's quality of life was 40 points according to the Yu.A. Orlov scale, which corresponds to a poor level [9].

Cervical MRI scan dated November 15, 2022: deformity of the spinal canal at the C1–C2 level associated with anterior dislocation of the C1 vertebra with spi-

nal cord compression at the level of the upper margin of the C2 vertebral body.

Cervical CT scan dated November 15, 2022: atlantoaxial dislocation associated with the C2 os odontoideum. Dislocation of the C1 vertebra to the front from the anterior margin of the C2 vertebral body was 10.6 mm, Swischuk test was 4.76 mm, Power index <1.

It was decided to perform reconstructive and stabilization surgery aimed at correction of atlantoaxial instability, decompression of the spinal cord at the upper cervical level, corrective facetectomy of C1–C2, artificial ankylosis of the atlantoaxial joints with autogenous bone grafts, and posterior screw fixation of C1–C2.

The course of surgery is similar to that outlined in clinical case 1.

The early postoperative period was unremarkable. The patient was verticalized on the day 2 after surgery in a rigid cervical collar. Neurological status was without increasing focal symptoms and long-tract signs. The patient was discharged on the day 7 with regression of pain syndrome (Fig. 2).

Discussion

Musculoskeletal symptoms are quite frequent in patients with DS. Some authors believe that the presence of os odontoideum in this group of patients is a key reason for the development of instability at the craniovertebral level. Atlantoaxial instability combined with atlantoaxial dislocation is a particularly hazardous clinical complication in patients with DS, since it results in the formation of cervical medullary syndrome [3, 5, 10].

From the clinical point of view, the anomaly may be asymptomatic, have local symptoms, or cause cervical myelopathy or symptoms associated with vertebrobasilar ischemia [11]. Local symptoms include pain, torticollis, and restricted mobility of the cervical spine, considered to be associated with impaired congruence of the articular surfaces of the C1–C2 joints [4, 5, 12]. Symptoms of cervical myelopathy can range from temporary paresis and sensation disorders to disabling quadriparesis with

difficult swallowing and breathing disorders [3–5]. Vertebrobasilar ischemia can result from compression of vertebral arteries, manifesting with dizziness, syncope, visual impairment, and other symptoms [4, 5, 13].

There is a range of concepts in determining indications for surgical treatment. Goel et al. [14] believe that surgical treatment is indicated for all patients with atlantoaxial instability of any origin. The authors suggest that even in cases of asymptomatic os odontoideum, sur-

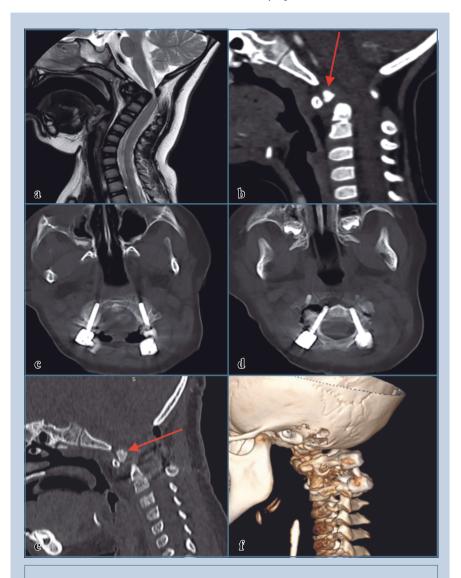


Fig. 1Data of examination of a three-year-old female patient: **a** − MRI scan of the cervical spine in T2-weighted mode, sagittal plane in the flexion position; **b** − MSCT scan of the cervical spine in the sagittal plane before surgery, the C2 os odontoideum is marked with an arrow; **c** − MSCT scan of the position of the fixation instrumentation elements at the level of the C1 vertebra; **d** − MSCT scan of the position of the fixation instrumentation elements at the level of the C2 vertebra; **e** − MSCT scan of the cervical spine in the sagittal plane in the flexion position after surgery, the C2 os odontoideum is marked with an arrow; **f** − MSCT scan with 3D reconstruction of the cervical spine after surgery

gery is indicated since the current level of technical equipment provides a reasonably safe way to perform the correction. At the same time, the individual features of the patient and the surgeon's professional experience should be kept in mind [14]. According to Wang et al. [15], on the contrary, conservative management with further clinical and radiological follow-up may be sufficient for patients with asymptomatic os odontoideum, and surgical treatment is indicated only in case of signs of atlantoaxial instability or its progression [3]. Salunke et al. [16] point to the correlation between the terms of clinical manifestation of atlantoaxial instability and the severity of articular facet tilt.

We are interested in studying this issue in the context of improving the algorithm of follow-up and timing of surgical treatment of neurologically compensated patients with atlantoaxial instability. Nowadays, we use the algorithm described by N.O. Khusainov et al. [17]. According to this algorithm, indications for surgical correction of C1-C2 segment instability are neurological disorders associated with spinal cord compression, obvious radiological signs of atlantoaxial instability (increased atlantodental interval more than 10 mm), decreased reserve space for the spinal cord less than 13 mm (violation of the rule of three Still), some anomalies in the development of bone structures of the craniocervical zone, in particular, the os odontoideum, basilar impression, and others [17].

The range of surgical techniques for the treatment of atlantoaxial instability includes various decompression options combined with craniocervical or C1-C2 fixation. If sufficient decompression by repositioning the atlas is not possible, posterior decompression in the form of laminectomy of the C1 vertebra is used in addition to stabilization [1]. In case of persisting prominent compression at the level of the craniovertebral junction, it is justified to supplement laminectomy of the C1 vertebra with resection of the posterior margin of the foramen magnum. The use of transoral odontoidectomy has also been described as an optional decompression. This technique requires appropriate experience on the part of the surgeon and is associated with the risk of developing specific severe, primarily infectious, complications [18–21]. Given these circumstances, the use of transoral odontoidectomy is reasonable only when there is a possibility of regression of conductive and bul-

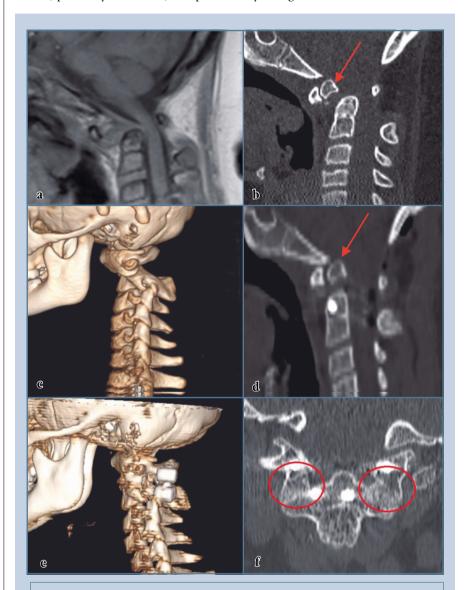


Рис. 2

Data of examination of a nine-year-old female patient: $\bf a-MRI$ scan of the cervical spine in T1-weighted mode, sagittal plane in the flexion position, severe stenosis of the spinal canal at the level of C1–C2 vertebrae; $\bf b-MSCT$ scan of the cervical spine in the sagittal plane before surgery, the C2 os odontoideum is marked with an arrow; $\bf c-MSCT$ scan with 3D reconstruction of the cervical spine before surgery; $\bf d-MSCT$ scan of the cervical spine in the sagittal plane after surgery, the C2 os odontoideum is marked with an arrow; $\bf e-MSCT$ scan with 3D reconstruction of the cervical spine after surgery; $\bf f-control MSCT$ scan 12 months after surgery, ellipses mark remodeling of bone implants placed between the lateral masses of the C1–C2 vertebrae

bar disorders associated with persistent anterior compression of the spinal cord and brainstem. However, in all cases, this decompression technique should be considered as an option to compensate for anterior spinal cord compression that remains after unsuccessful posterior correction, but not as a routine treatment option.

The development of microsurgical techniques in combination with neurophysiological control has significantly enhanced the possibilities of C1–C2 segment mobilization and intraoperative repositioning of the displaced atlas. Nevertheless, in some cases, preoperative halo correction remains appropriate to reduce the risk of disabling neurological complications during surgical treatment of gross upper cervical deformities [22].

Being undoubtedly a less risky procedure than C1-C2 fixation, craniocervical fixation has a number of serious disadvantages, minimizing its value as a routine treatment option. The restricted range of motion of the head relative to the neck has a significant impact on the life quality of patients, impairing spatial orientation and swallowing function. Furthermore, the use of extended craniocervical instrumentation in children in a number of cases requires repeated revision surgeries associated with both excessive load on the distal fixation elements and continued growth of the child's body [23, 24]. Meanwhile, atlantoaxial dislocations and instability in congenital malfor-

mations, such as DS, may be combined with gross anatomical anomalies, excluding the possibility of monosegmental screw fixation. Similarly, when stabilization of atlantoaxial instability is required in young patients or patients with stunted growth, C1-C2 screw fixation may not be possible because of insufficient development of the bony elements of the vertebrae [23, 24]. Frequently, bony anomalies are associated with anomalies of the vascular arterial bed of the craniovertebral junction. Diameter anomalies and aberrant vertebral artery at the V3 segment in combination with variants of the circle of Willis formation are associated with the risk of severe complications and may be a contraindication to Goel-Harms fixation [25]. Considering the above-mentioned circumstances, craniocervical fixation in its variants remains relevant in the treatment of this pathology.

C1–C2 fixation is the most common surgical treatment option for atlanto-axial instability. The use of various modifications of instrumental fixation of the C1–C2 segment in addition to deformity correction contributes to bone block formation and is associated with regression of the underlying neurological deficit. The Goel-Harms technique is safer than Magerl transarticular screw fixation and has greater stability than hook and wire stabilization options [25, 26]. Another fixation technique for atlantoaxial instability is the Goel-Shah technique, where-

by, in addition to posterior C1-C2 screw fixation, stabilization is performed with inter-articular spacers, which provide greater stability of the segment [6].

In the clinical cases mentioned above, the author's patented method of eliminating the anterior tilt of the articular facets of the atlantoaxial joints to compensate for the dislocating force caused by them, directed anteriorly and downwards, was applied, which provided increased stability of the bone block of the C1–C2 joints and reduced the load on the fixation instrumentation.

Conclusion

The Goel-Shah reconstruction and stabilization technique in combination with atlantoaxial joint facet remodeling is most relevant in cases of chronic atlantoaxial dislocations in patients with DS. This technique combines spinal cord decompression with monosegmental fixation to reduce the load on the instrumentation by eliminating the anterior tilt of the articular facets of the C1–C2 vertebrae.

The study had no sponsors. The authors declare that they have no conflict of interest.

The study was approved by the local ethics committee of the institution.

All authors contributed significantly to the research and preparation of the article, read and approved the final version before publication.

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