CASE REPORT

Surgical treatment of symptomatic vertebral canal stenosis in patients with achondroplasia – case report

Chirurgiczne leczenie objawowego zwężenia kanału kręgowego u pacjentów z achondroplazją – opis przypadku

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Abstract

Authors report the history of two patients with achondroplasia, treated for symptomatic lumbar spinal stenosis. Both patients underwent cross-limb lengthening. During control visits they reported lumbar spine pain with increasing neurogenic claudication. In magnetic resonance imaging of the lumbar spine, we measured dural sac cross-sectional area (DSCSA) and spinal canal cross-sectional area (SCCSA) and the patients were qualified to the group of patients with critical spinal stenosis. They underwent "split" type laminotomy in the lumbar spine. In both patients, based on the Oswestry questionnaire, we noted very good result of surgical treatment. The most important change was increase of the walking distance after the procedure. In patients suffering from achondroplasia, with critical lumbar spinal canal stenosis and neurological symptoms the procedure of choice is surgical treatment. Surgical decompression of the spinal canal using "split" type laminotomy is an effective technique, preventing the destabilization of the posterior column and sagittal imbalance of the spine.

Key words: achondroplasia, vertebral canal stenosis, surgical treatment

Streszczenie

Autorzy przedstawiają historię choroby dwóch pacjentów chorych na achondroplazję, leczonych z powodu objawowego zwężenia kanału kręgowego w odcinku lędźwiowym. Obydwie pacjentki przeszły operacje krzyżowego wydłużania kończyn dolnych. Podczas wizyt kontrolnych zgłosili ból kręgosłupa lędźwiowego z narastającym chromaniem neurogenym. W badaniach obrazowych rezonansu magnetycznego kręgosłupa lędźwiowego, na podstawie pomiaru pola przekroju worka oponowego (dural sac cross-sectional area=DSCSA) jak i pola kanału kręgowego (spinal canal cross-sectional area=SCCSA), chorzy zostali zakwalifikowani do grupy pacjentów z krytycznym zwężeniem. Wykonano u nich zabieg laminotomi typu „split” w odcinku lędźwiowym. U chorych, w oparciu o kwestionariusz Osowski, odnotowano bardzo dobry wynik leczenia operacyjnego, a najistotniejsza zmiana dotyczyła zwiększenia dystansu jaki potrafi pokonać po zabiegu. U chorych z achondroplazją, ze skrajnym zwężeniem kanału kręgowego w odcinku lędźwiowym i objawami neurologicznymi postępowaniem z wyboru jest leczenie operacyjne. Chirurgiczne odbarczenie kanału kręgowego poprzez laminotomię typu „split” jest skuteczną techniką operacyjną zapobiegającą destabilizacji tylnej kolumny kręgosłupa i dalszym zaburzeniem balansu strzałkowego kręgosłupa.

Słowa kluczowe: achondroplazja, stenoza kanału kręgowego, leczenie chirurgiczne
**Introduction**

Skeletal dysplasias, i.e. osteochondrodysplasias, is a group of diseases characterized by generalized growth disorder and disturbance of bone and cartilage development. Achondroplasia is the most common form of dwarfism among over 200 skeletal dysplasia described [1,2].

Achondroplasia is a rare genetic disease with a prevalence estimated at about 1 case per 30,000-50,000 births. It is inherited in an autosomal dominant way, although over 80% of cases are due to new spontaneous mutations [2]. This mutation refers to the gene encoding the type 3 fibroblast growth factor receptor (fibroblast growth factor-3 – FGFR-3) mapped on chromosome 4 [3]. As a result of the disorder FGFR-3 there is an impairment of cell division in the proliferative zone of growth cartilage and this creates the phenotypic features of achondroplasia.

Patients with achondroplasia are characterized by dwarfism with shortened proximal limb segments (arms, thighs), what is termed as rhizomelic dysplasia. The length of the torso is close to the normal values. The motor development in children is delayed, but proper motor coordination and gait mechanics are achieved around the age of 2. Intellectual development is normal unless there is a complication in the central nervous system. Face features certain characteristics like prominence of frontal bone tumors and underdevelopment of the middle part of the face. The hands of people affected by this disease are characterized by an additional gap between the third and fourth finger creating the “trident hands”. Generalized ligamentous laxity accompanying this disease causes the genu varum and whole lower limbs are set in external rotation [4]. In the spine, we observe excessive lumbar lordosis with horizontally positioned sacral bone and kyphosis of the thoraco-lumbar passage [5,6]. There is also a shortening of the pedicles of the vertebrae, widening the surface of intervertebral joints and overgrowth of the ligamentum flavum which ultimately leads to central and foraminal stenosis.

**Aim**

Aim of the work – to present the medical history of two patients with achondroplasia, treated for symptomatic spinal canal stenosis in the lumbar region with associated neurological symptoms.

**Materials and methods**

In recent years, in one orthopaedic centre two patients with achondroplasia have been treated for symptomatic spinal canal stenosis. The evaluation of the results of their treatment was carried out retrospectively based on the history of the disease and imaging studies available in the hospital’s archive.

**Results**

Patient No. 1, now a 27-year-old man, with achondroplasia, started his treatment due to dwarfism at the age of 12. The aim of the surgery was to lengthen the lower limbs using the Ilizarov apparatus. A satisfactory effect was obtained. From that time, the patient has been under constant observation of the orthopedic clinic.

In 2011, during another follow-up for the first time he reported pain in the lumbar spine. Physical examination revealed a limitation of lumbar spine mobility, and in lateral X-ray examination of the spine showed a typical disorder of the curvature of the spine in the form of kyphosis of the thoraco-lumbar junction, increased lumbar lordosis and horizontal positioning of the sacrum (Fig. 1). Diagnostic imaging was deepened and included magnetic resonance imaging (11.05.2011) which showed the spinal canal stenosis in the lumbar region. Due to the mediocre back pain conservative treatment was applied. A year later, for the first time the patient reported periodic pain in the lower limbs with longer...
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walking. Symptoms of neurogenic claudication did not significantly limit his daily activity, therefore the patient, after consultation with the doctor, did not decide to undergo surgical treatment. It was not until six years later that the patient reported intensifying pain, muscle weakness in the lower limbs and, as a result, disturbed gait mechanics. The distance of the claudication was reduced to 100 meters. Laseque’s symptoms were negative on both sides. In the magnetic resonance imaging of the lumbar spine (04.05.2017) there was a critical stenosis of the spinal canal in the lumbar area. In connection with the above, the patient was referred for surgical treatment. A “split laminotomy” L2 - L5 was performed. The course of the operation was uncomplicated. Seven weeks after the surgical intervention, pain and neurological symptoms completely subsided. Twelve weeks after the surgery, an episode of periodic numbness occurred in the area of the left buttock and dorsal surface of the left foot, occurring mainly in bending back of the spine. After pharmacological treatment these symptoms have withdrawn.

Split laminotomy is a procedure involving the intersection of supra and interspinous ligaments as well as spinous processes in the midline. Next, decompression of the spinal canal is performed by removing the posterior arches and the ligamentum flavum (Fig. 3). From this access it is also possi-

Fig. 2. X-ray pictures in two projections after surgery, patient No. 1.

Fig. 3. “Split laminotomy” – own drawings.
ble to decompress the roots of the spinal nerves by increasing the space in the lateral recesses. After dilation of the spinal canal, the nonabsorbable sutures are used to reapproximate previously split spinous processes together with the accompanying ligaments. This surgical technique reduces the traumatization of the paraspinal muscles and restores the ligaments stabilizing the posterior column of the spine [7].

Patient No. 2 is a 28-year-old man with achondroplasia who, like the previous patient, has undergone a number of surgeries, including lengthening of lower limbs. At the age of 25, the patient reported pain in the lumbar spine with numbness of the lower limbs. Magnetic resonance imaging of the lumbar spine (27.10.2015), revealed spinal canal stenosis. Pain complaints were moderate, therefore the patient, after consultation with the doctor, did not decide on surgical intervention. However, the symptoms associated with the spinal canal stenosis gradually began to increase, there was back pain with radiation to the lower limbs and deteriorating gait mechanics (Fig. 4). Therefore, more than two years later, during the follow-up visit, the patient underwent magnetic resonance of the lumbar spine and he was referred for surgical treatment, which took place six months later. Surgery that consisted of L4-S1 split laminotomy was complicated by dural damage and cerebrospinal fluid leakage (Fig. 5). During the procedure, the damaged dural sac was surgically sutured and sealed and a lumbar catheter was implanted at the L3
Both patients were routinely evaluated for the effects of lumbar spine pain on the ability to perform daily activities based on the Oswestry questionnaire [8]. The survey was completed before and after surgery. The most significant change after surgical treatment was observed in increasing the patient’s ability to cover more distance (Table 1). In patients, magnetic resonance images, in cross sections, both the dural sac cross-sectional area (DSCSA) and the spinal canal cross-sectional area (SCCSA) were measured (Fig. 6 and 7). Both patients were allocated to the group of patients with DSCSA critical stenosis [9] (Table 2).

### Table 1. Walking abilities before and after surgery

<table>
<thead>
<tr>
<th>Walking</th>
<th>Patient before op.</th>
<th>Nr 1 before op.</th>
<th>Nr 1 after op.</th>
<th>Patient before op.</th>
<th>Nr 2 before op.</th>
<th>Nr 2 after op.</th>
</tr>
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<tbody>
<tr>
<td>A Pain does not prevent me walking any distance</td>
<td></td>
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<tr>
<td>b Pain prevents me from walking more than 1500 m.</td>
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<tr>
<td>c Pain prevents me from walking more than 800 m.</td>
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<tr>
<td>d Pain prevents me from walking more than 400 m.</td>
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<td>e I can only walk using a stick or crutches</td>
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<tr>
<td>f I am in bed most of the time</td>
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### Table 2. MRI measurements

<table>
<thead>
<tr>
<th></th>
<th>DSCSA [mm²]</th>
<th>SCCSA [mm²]</th>
</tr>
</thead>
<tbody>
<tr>
<td>Patient nr 1</td>
<td>48,5</td>
<td>77,8</td>
</tr>
<tr>
<td>Patient nr 2</td>
<td>57,5</td>
<td>70,7</td>
</tr>
</tbody>
</table>
Discussion

Achondroplasia is a disease which, by shortening pedicles, widening the articular surfaces and hypertrophy of the ligamentum flavum, predisposes to stenosis of the spinal canal [10,11]. Aging is typically accompanied by degenerative changes such as: reduction of hydration and the height of the intervertebral discs, hernia of the nucleus pulposus, hypertrophy of the ligaments and joint surfaces, and finally spinal canal stenosis. At the time when the compensation capacities of the space in the spinal canal are terminated, there is pressure on the dural sac and nervous structures located within the canal. As a result, patients begin to present neurological symptoms in the form of neurogenic claudication [12]. The most frequently used diagnostic examination in the assessment of spinal canal stenosis is magnetic resonance imaging. On cross-sectional scans we can measure the dural sac cross-sectional area (DSCSA) and spinal canal cross-sectional area (SCCSA). The more sensitive parameter is the dural sac cross-sectional area (DSCSA), which is measured at L4 / L5 and if it is below 70 mm² it is responsible for the critical stenosis. The measurements used to assess the spinal canal stenosis are associated with clinical symptoms. Typical symptoms of spinal canal stenosis in the lumbar region include: uni- or bilateral buttock pain, neurogenic claudication and pain, numbness, weakness of the lower limbs leading to paresis or paralysis. In order to better understand the impact of lumbar spine pain on the limitations of everyday activities, the Oswestry Questionnaire was introduced, which also helps in assessing the results of surgical treatment. Decompression of the spinal canal in the lumbar region by laminotomy does not destabilize the spine, and due to the increased spinal canal cross-sectional area the compression on the dural sac and nerve structures decreases [13, 14]. As a result, the neurological symptoms disappear, and the patient quickly returns to the normal everyday activity. Carlisle emphasizes that the improvement is usually measured in the walking distance – similarly in our patients there was a significant reduction in neurogenic claudication [15]. It seems that the sooner we decide on surgery, the better result can be obtained, although in our patients the decision about surgical treatment was not easy. Such an operation does not change the sagittal profile of the spine but reduces the risk of potential complications associated with the changes of its primary sagittal and frontal balance [16]. Although Baca et al recommend using instrumentation to stabilize the spine, it seems to us that this depends on the extent of the laminotomy performed and is not always necessary [17].

Conclusion

1. In patients with achondroplasia, with extreme spinal stenosis, surgery is the treatment of choice.
2. “Split-laminotomy” without instrumentation seems to be an effective surgical technique in these patients.

References

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