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ORIGINAL ARTICLE

Surgical outcome after treatment of thoracolumbar spinal stenosis in adults with achondroplasia

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Abstract

Purpose To describe the complications and the outcome of patients with achondroplasia undergoing thoracolumbar spinal surgery.

Methods Retrospective analysis of prospectively collected data of all patients with achondroplasia undergoing surgery within the years 1992–2021 at the thoracic and/or lumbar spine. The outcome was measured by analyzing the surgical complications and revisions. The patient-rated outcome was assessed with the COMI score from 2005 onwards.

Results A total of 15 patients were included in this study undergoing a total of 31 surgeries at 79 thoracolumbar levels. 12/31 surgeries had intraoperative complications consisting of 11 dural tears and one excessive intraoperative bleeding. 4/18 revision surgeries were conducted due to post-decompression hyperkyphosis. The COMI score decreased from 7.5 IQR 1.4 (range 7.1–9.8) preoperatively to 5.3 IQR 4.1 (2.5–7.5) after 2 years ($p = 0.046$).

Conclusion Patients with achondroplasia, the most common skeletal dysplasia condition with short-limb dwarfism, are burdened with a congenitally narrow spinal canal and are commonly in need of spinal surgery. However, surgery in these patients is often associated with complications, namely dural tears and post-decompression kyphosis. Despite these complications, patients benefit from surgical treatment at a follow-up of 2 years after surgery.

Keywords Achondroplasia · Thoracic · Lumbar · Surgery · Laminectomy

Introduction

Achondroplasia is considered the most frequent chondrodysplasia and cause of short-limb dwarfism [1]. Its incidence is reported to be 1 in 10,000 to 26,000 live births [2, 3]. Around 80% of the affected individuals have new mutations, the remaining 20% are of autosomal dominant inheritance [4]. Achondroplasia is caused by a mutation of the FGFR3 (fibroblast growth factor receptor 3) gene, which results in a defect in enchondral bone formation [5]. Individuals typically have a long narrow trunk, short limbs (rhizomelia), bowing of the lower legs, a relatively large head (macrocrania) with frontal bossing, and mid-face depression [6]. Medical complications such as sleep apnea, frequently obstructed upper airways, otitis media, and decreased pulmonary function can all be encountered.

Spinal manifestations, with typical pathologies including symptomatic spinal stenosis and/or thoracolumbar kyphosis, are common. Stenosis can occur along the whole length of the spinal canal (Fig. 1). Incidence increases at a later stage of life, typically in the thoracic and lumbar spine due to the superposition of degenerative changes. Anatomy of the spine is characterized by a steep sacral angle, high lumbar lordosis of the lumbar spine, thoracic hyperlordosis and scalloping of the posterior vertebral body. 10–15% of all patients with achondroplasia develop a fixed hyperkyphosis of the thoracolumbar junction with a prevalence of scoliosis in up to 60% [7]. Contrary to patients without achondroplasia, the inter-pedicular distance decreases from L1 to L5 [8]. Due to the premature closure of the physis, the spinal canal diameter and pedicle length are substantially smaller than in non-achondroplastic individuals, with the narrowest spinal canal cross-sectional area being at the L4 level [9]. Such narrowing allows less room for the neural structures as degenerative changes occur, such that patients present earlier

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Fig. 1 T2-weighted MRI image of a 53-year-old female patient suffering from achondroplasia. (**a**) Axial image between the 10th and the 11th thoracic vertebra featuring a narrow spinal canal. (**b**) Sagittal imaging of the thoracic spine demonstrating multiple segment spinal stenosis

with neurological disturbances and claudication symptoms [9]. As a consequence, this patient population is frequently in need of surgical treatment. However, the current evidence on the postoperative outcomes and surgical complications for these patients is very limited. Therefore, it was the goal of this study to analyze and describe the outcome after spine surgery of the thoracolumbar spine in individuals suffering from achondroplasia.

Methods

For this study, we retrospectively analyzed the prospectively collected data from our single-center patient database and medical records. All patients with achondroplasia who had undergone surgery between the years 1992–2021 at the thoracic and/or lumbar spine were included in this study.

Data were extracted from our clinic s information system through a full-text search for the keywords "achondroplasia" and "chondrodysplasia." Detailed patient information was obtained from the patient charts. Subsequently, outcome scores were retrieved from our local Spine Surgery Outcomes Database, which is part of the International Spine Tango Registry.

Pre- and postoperative complications were evaluated reviewing the operation reports and the discharge papers of our patient cohort. For patient-rated outcomes, the Core Outcome Measures Index (COMI) was utilized [10]. The score is routinely evaluated preoperatively and at 3, 12, and 24 months after surgery. A COMI score of 0 is considered the best and a score of 10 is considered the worst. However, as the COMI score was introduced to our clinic in 2005, several patients had no patient-rated outcome available (Table 1).

In this study, three different techniques were used for decompression surgery. Unilateral decompression consisted of a midline incision with unliteral sharp dissection along the spinous process with preservation of the interspinous and supraspinous ligament. The midline decompression was performed with bilateral sharp muscle dissection, sacrificing the interspinous and supraspinous ligament with the removal of the ligamentum flavum. The laminectomy consisted of full resection of the affected lamina and therefore also dissection of the adjacent segment inter- and supraspinous ligament.

According to the distribution according to the Kolmogorov–Smirnov test, the results are reported as the median and interquartile range (range) or mean and standard deviation. To assess the effect of surgery, the Wilcoxon test was used to compare preoperative scores with those at the 2-year follow-up. For the comparison of dichotomous data, Chi^2-test was employed. p Values less than 0.05 were considered statistically significant. Statistical analyses were carried out using IBM SPSS statistics version 23.

This study was approved by the local ethics committee before its start and all patients included gave informed consent for the use of their data.

Results

A total of 15 patients matched our inclusion criteria and were included in this study. The study cohort consisted of 8 male and 7 female patients. The mean age at the last spine surgery performed in these patients was 52 years (SD 10.4). Between February 1992 and July 2021, a total of 31 surgeries were performed on patients with achondroplasia with decompression surgeries at 79 thoracolumbar levels. Unilateral decompression was done in 1 surgery, midline decompression in 25 and laminectomy in 5 surgeries. As 5

surgeries did not include decompression, the mean number of decompressed segments was 2.3 (SD 1.2; range 1–9). The median follow-up after the last surgical procedure was 24 IQR 34 (range 4–85) months.

Surgeries consisted of 12 primary surgeries at our center and 19 surgeries at levels that had been previously operated on or were adjacent to previously operated segments. The indication for primary surgery was myelopathy in 5/12, spinal claudication in 4/12, paraparesis in 2/12 and acute foot drop in one of these cases. All primary surgeries were decompression surgeries alone.

In those cases of revision surgery, 3/19 surgeries were conducted due to early postoperative complications during the same hospital stay or within 30 days after discharge. 2/19 were revisions due to a dural tear and wound healing disorder and one due to early adjacent segment collapse after fusion which was treated with an extension of the fusion (Fig. 2).

In the late (> 30 days) revision group, which consisted of 16/19 revision surgeries, surgeries were indicated due to same or adjacent segment spinal stenosis with claudication, myelopathy or paraparesis in 7/16 cases. 4/16 surgeries were conducted due to post-decompression hyperkyphosis with significant neural compression in 2 instances. 2/16 surgeries were conducted due to adjacent segment disease and 2/16 had pseudoarthrosis. A single patient underwent surgery due to a posttraumatic deformity. The late revision surgeries consisted of decompression of adjacent segments in 5/16 surgeries, decompression and fusion in 8/16 surgeries and alignment correction with an osteotomy in 3/16 surgeries.

Prior to operative treatment, a neurologic motor deficit or bladder dysfunction was present 18 times. In 5 cases, the neurologic recovery was full, while 11 cases only exhibited a partial neurologic recovery and 2 patients did not improve their neurological status postoperatively.

In total, 12/31 surgeries had intraoperative complications consisting of 11 dural tears and one patient with excessive intraoperative bleeding and therefore, an early surgery termination. The complications were distributed equally between primary and revision cases, with 5 dural tears and the excessive bleeding complication in the primary surgery group and 6 dural tears during revision surgeries. Eight out of 31 surgeries had postoperative complications consisting of 2 cases of implant failure, 2 cases of wound healing disorder, 1 patient suffering from ileus postoperatively, 1 patient having urosepsis due to a cystofix with bladder dysfunction,

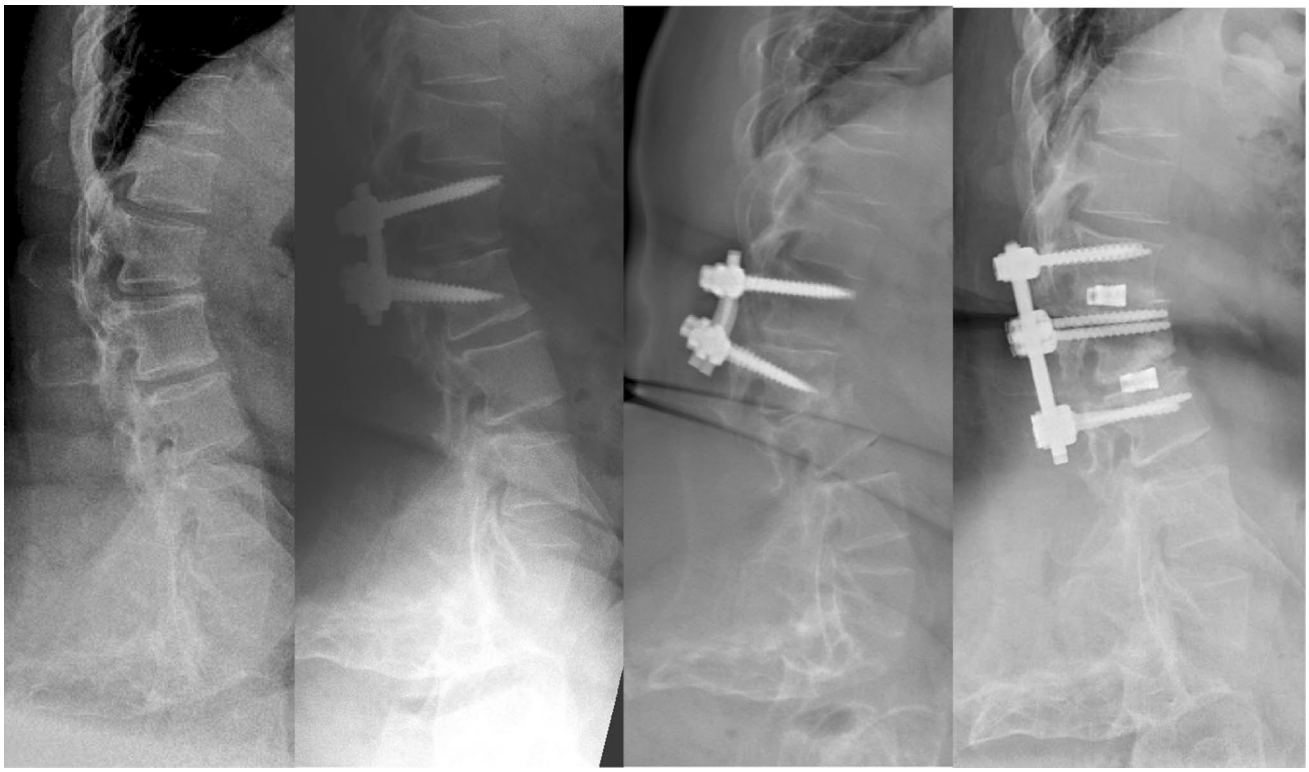

Fig. 2 Conventional lateral radiographs of the lumbar spine of a 54-year-old female. (**a**) Preoperative imaging. (**b**) Postoperative imaging after posterior Fusion L1-L2. (**c**) 2 years postoperatively she developed adjacent segment kyphosis with neurologic symptoms due to central spinal stenosis. (**d**) After the revision, three days postoperatively, the sagittal spinal alignment of the collapsed level was partially restored. However, the most cranial level showed early screw loosening and cage subsidence, which led to subsequent revision surgery

1 superficial wound infection and 1 case of postoperative anemia. In the surgical cases following a laminectomy, there was a significant association with the postoperative occurrence of segmental kyphosis ($p = 0.048$). We found no difference between patients with and without instrumentation regarding neither intraoperative ($p = 0.739$) nor postoperative complications ($p = 0.592$).

Outcome scores were available for 6 patients. The overall outcome score decreased from 7.5 IQR 1.4 (range 7.1–9.8) preoperatively to 5.3 IQR 4.1 (2.5–7.5) after 2 years, which showed a significant decrease in the score at the last follow-up ($p = 0.046$). Patient characteristics are provided in Table 1.

Discussion

Achondroplasia is associated with a developmental narrow spinal canal diameter and therefore the common need for spinal decompression surgery [11]. Due to these anatomical differences, patients are usually younger during index surgery when compared to the mean age in decompression in patients without achondroplasia [11, 12]. Unlike in adults without Achondroplasia, the rate of intraoperative complications in these patients is exceedingly high, which is also well displayed in this study, as 11 out of 31 surgeries had intraoperative dural tears. In patients with achondroplasia, the dura mater is often found to be extraordinarily thin and therefore susceptible to injuries. Also, the dura can be directly adherent to the bone and the joint capsule, even in cases without prior surgery, making it difficult to be separated from osseus structures without causing an injury. The incidence of dural tears for lumbar decompression surgery in patients without skeletal dysplasia conditions is between 1.6% and 10% [13]. The dura is usually very thin and pseudo-meningocele can develop even in the absence of evidence of a dural tear [14]. In case of incidental durotomy, special attention should be paid to obtaining a watertight closure, including coverage of the exposed dura with a muscle flap so that no dead space is left. In our experience, this helps to prevent the formation of a pseudo-meningocele. Another challenge in these patients arises from the small stature, pronounced lumbar lordosis, configuration of the pelvis and horizontal sacrum [11]. These anatomical features may lead to difficulties in patient positioning. Also, the narrow pelvis further hampers the approach. In particular, it is crucial that the lowest possible abdominal pressure is created, in order to reduce intraoperative excessive blood loss, which may have been the case in one patient in this study and could have been prevented.

Another complication encountered in this population was adjacent segment kyphosis which was associated with prior laminectomy ($p = 0.048$). A collapse of the adjacent segments may have been caused by the destabilization of posterior structures [15]. In contrast, Vleggeert-Lankamp et al. reported no such complication, even in kyphotic deformities over 40° in a cohort of 20 patients [16]. If the patient wants to refrain from fusion, decompression only, even in cases of more severe thoracic kyphosis, may be feasible; however, these patients should be followed radiographically. However, the results of this study are

Table 1 Patient characteristics sorted by follow-up after the last surgery

Patient Number	Levels decompressed	Levels fused	Age at last surgery	Gender	Number of surgeries	Complications	Follow-up/ months	COMI-Baseline	COMI 2 year follow-up
1	2	0	48	F	1	None	4	1.9	
2	2	0	53	M	1	None	12		
3	12	11	56	F	5	Dural tear, Urosepsis, Junctional kyphosis	12	4.95	
4	11	0	43	F	3	Delayed wound healing	15	2.85	
5	1	0	64	M	1	Infection	24	7.1	2.5
6	10	8	44	F	2	Dural tear	24	9.8	6.35
7	3	0	54	F	5	Ileus	24	8.1	4.2
8	4	0	46	F	1	Dural tear	24		3.55
9	5	0	49	M	2	Dural tear	24		0.2
10	10	4	65	M	3	Dural tear, Implant failure	26	7.2	6.45
11	7	8	50	M	6	Implant failure with junctional kyphosis	36	7.55	7.7
12	6	5	35	F	2	Dural tear, Excessive bleeding	46	2.85	
13	4	0	56	M	1	Dural tear	58	7.4	2.7
14	1	0	38	F	1	None	69	3.6	
15	1	1	74	M	1	None	85	3.2	

limited as 3 cases of adjacent segment kyphosis, of which in 2 cases were post-laminectomy, occurred in 1 patient.

In the literature, several other authors reported similar high complication rates. Carlisle et al. reported rates as high as 40.8% of the patients in her study [17]. Also, Shafi et al. found an overall complication rate of 48% despite multidisciplinary patient management, with dural tears being the most common complication at 36% [18]. Vleggert-Lankamp et al. only reported 1 dural tear in a cohort of 20 patients with wrong level surgery being the most common complication in 4 cases [19]. Carlisle et al. as well reported similar complication rates with an overall complication rate of 40.8%, with dural tears in 30.6% being the most common complication [17].

In previous studies, patient-related outcomes have rarely been reported as they were only provided by 3 authors [20]. Ain et al. reported an improvement in the Rankin score and an improvement in the walking distance in patients with achondroplasia after laminectomy in 49 patients [21]. Vleggert-Lankamp et al. provided outcome measures according to the Cooper myelopathy grading and Odom criteria [19]. As such, the Cooper score showed postoperative improvement of no more than only 1 point in only 8 out of 20 patients and poor outcome in poor or fair outcomes in 14 patients [19], despite the low complication rate reported. Carlisle et al. demonstrated an improvement in walking distance of patients undergoing surgery and an improvement of the Rankin level in patients with symptoms less than 6 months prior to surgery. As demonstrated, all outcomes provided in the literature so far are highly heterogenous and so far, no patient-related outcome are available. This is the first study to provide patient-related outcome measures according to the COMI outcome scores for a 2-year follow-up which provided proof that these patients profit from surgery in the long term.

This study's greatest limitations were the low availability of outcome scores, the low number of patients and the retrospective study design. Only 6 patients had an adequate outcome score available and the study cohort only consisted of 15 patients. There are several reasons for the low number of scores available. The COMI score was introduced at a later time point which means that it was not available for patients operated on before 2005, which was the case in 2 patients. One patient declined participation, and 3 patients had no outcome score due to an administrative error. Two patients had a follow-up of under 2 years. One patient came from abroad and was therefore lost to follow-up.

The spine surgeon must be aware of the difficulties that may arise in patients with achondroplasia. Thorough patient positioning and altered awareness for intraoperative complications, especially dural tears, are important in this challenging patient cohort. However, despite the present high complication rate, patients do benefit from surgical treatment at a long-term follow-up.

Conclusion

Achondroplasia is a rare genetic syndrome and the most common form of short-limb dwarfism. These patients are burdened with a congenitally narrow spinal canal and are commonly in need of spinal surgery. However, surgery in these patients is often associated with high overall complication rates, especially due to dural tears and post-decompression kyphosis. Despite these complications, patients benefit from surgical treatment at a follow-up of 2 years after surgery.

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Declarations

Conflict of interest The authors declare that they have no conflict of interest.

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