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Case Report

Cervical spine surgery in patients with diastrophic dysplasia: Case report with long-term follow-up

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Abstract

Cervical kyphosis in diastrophic dysplasia (DTD) is a very dangerous deformity which may lead to compression of neural structures resulting in tetraplegia or even. Treatment of this deformity is usually surgical, but no long-term follow-up studies are presented in the literature. Authors present a case of two children with DTD who underwent anterior corpectomy due to severe cervical kyphosis. The kyphotic deformity was corrected and the normal spinal canal width was restored. The effects of the correction remained stable for respectively 6 and 10 years of the follow-up period. The unique follow-up confirms that this type of intervention leads to an effective and long lasting results. Significant cervical kyphosis in patients suffering from DTD may be treated surgically using anterior approach even in young children with a favorable and lasting results.

Key words: Cervical spine, diastrophic dysplasia, surgery

INTRODUCTION

Diastrophic dysplasia (DTD) previously defined as atypical achondroplasia is a rare genetic condition with the highest incidence in Finland.[1] DTD results from mutations localised at the distal long arm of chromosome 5. Diagnosis is possible before birth using sonograms and DNA testing.[2] Clinical picture includes short stature with short limbs, hip joint dislocations, unstable knee joints, and clubfeet. The thumb usually is wide, hypermobile, with radial deviation and is referred as "hitchhiker's thumb." The nasal bridge is narrow, with broad nostrils and cleft palate.[1,3] Scoliosis constitutes the most common spinal deformity, together with altered sagittal balance.[1,4] Cervical kyphosis is less common, but may a cause of severe neurological complications.[1,4,5] Surgery is rarely advocated. Hence no long-term outcomes are present in the literature.[5] Therefore, purpose of this study is to present the result of treatment of two patients with DTD who underwent surgery for cervical kyphosis with postoperative follow-up of 6 and 10 years, respectively.

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CASE REPORTS

Case 1

The patient, Caucasian male, with genetically confirmed DTD, presented at the age of six due to progressing spinal deformity

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and increasing walking problems. Medical history included previous hip and foot surgeries. The clinical picture included Hitchhiker's thumb, facial dimorphism, knee joint contractures, hip dislocations, clubfeet, and scoliosis. Spinal films showed a 70° right-sided thoracic scoliosis with 26° kyphosis. Hypoplasia of C5 vertebral body with a 10 mm anterior dislocation of C4 vertebra was present [Figure 1]. Segmental C4/C5 kyphosis equalled 40° and the entire cervical spine exhibited 20° of kyphosis. The width of the spinal canal at this level was 5.7 mm in the sagittal plane with spinal cord modelling, but with no intramedullary changes. Due to difficulties with walking associated with critical stenosis at the level of the defect and confirmed progression of cervical kyphosis surgery was recommended. Partial corpectomy of C5 was performed with reposition of C4/C5 dislocation and anterior C4-C6 fixation with mesh cage and plate. No intraoperative complications were noted. The soft collar was recommended for 6 weeks. Surgery restored the correct sagittal cervical spine profile to 34° of lordosis and segmental C4/C5 angulation to 10° lordosis. In the following years, the patient underwent surgery for scoliosis, hip osteotomy and forefoot reconstruction. Ten years after cervical spine surgery C4/C5 segmental lordosis was 10°, C1-C7 lordosis 51°. Smooth outlines of the vertebral canal walls in the cervical spine were present in a follow-up computer tomography scan [Figure 2]. The patient remained an independent, self-sufficient walker.

Case 2

The patient, Caucasian female, was admitted to the hospital at the age of five due to balance disturbances, dizziness, and episodes of fainting resulting from neck motion. The clinical picture was typical for DTD. Cervical radiograms showed an anterior C4/C5 dislocation. Cervical lordosis was 32°, but segmental C4/C5 kyphosis was 52°. Magnetic resonance imaging revealed other anomalies: Deformation of the dens, cleft in the C3 and C4 vertebrae. Doppler ultrasound of the carotid arteries presented an impaired blood flow during neck motion. Flexion/extension cervical films showed hypermobility at the level of dislocation. The patient underwent C5 corpectomy with reposition of the dislocation, reconstruction with iliac autograft and anterior fixation with plate. No intraoperative or postoperative complications were noted. The soft collar was utilised for 6 weeks. Postoperatively, the C4/C5 angle equalled 7° of lordosis, and the entire cervical lordosis angle was 37°. Clinical symptoms resolved. Six years postoperation the deformity was stable, with 9° of C4/C5 lordosis and 45° of cervical lordosis. A solid fusion at C4-C6 was present although the lower adjacent segment showed signs of degeneration [Figure 3]. Further surgery was performed for scoliosis and foot deformity.

DISCUSSION

Scoliosis and cervical kyphosis constitute most common spinal deformities associated with DTD.[4,5] Of these two, cervical kyphosis, despite a mostly favorable natural course, may deem serious neurological complications.[5-7] Incidence of this deformity is up to 30%.[5] The etiology of deformity results from hypoplasia of the vertebral bodies in the mid cervical spine. This furtherly leads to an anterior dislocation of the

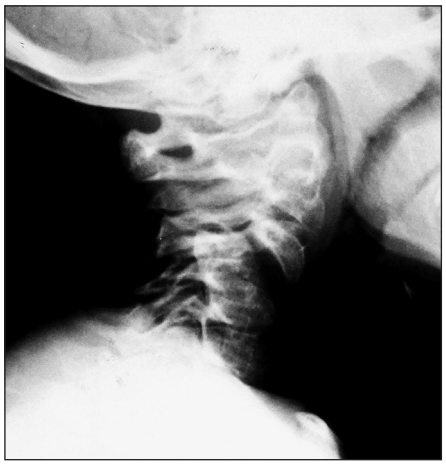

Figure 1: Case 1, male. Lateral radiograph of the cervical spine before surgery, hypoplasia of C5 vertebral body and anterior luxation of C4

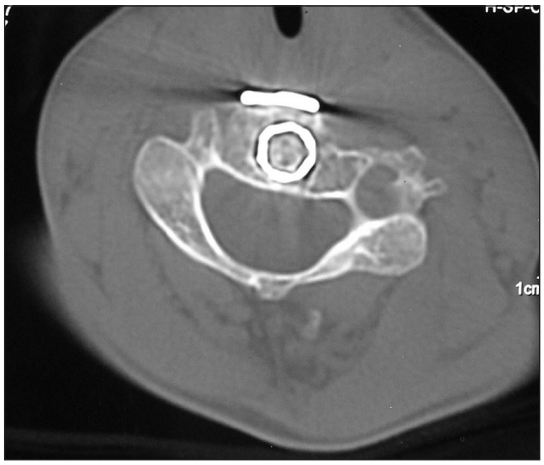

Figure 2: Case 1, male. Follow-up computed tomography scan of the cervical spine with visible smooth shape of the vertebral canal

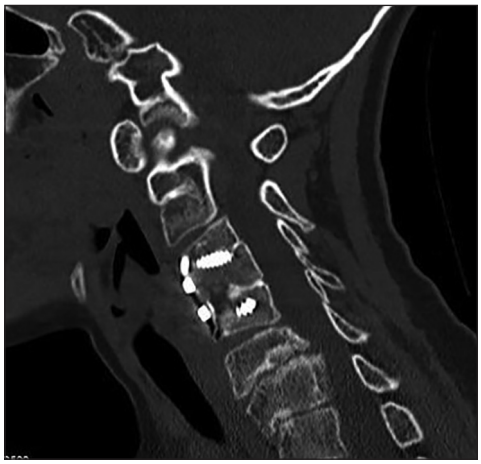

Figure 3: Case 2, female. Solid fusion at C4-C6 is visible. The lower adjacent segment shows signs of degeneration

superior adjacent vertebra with potential stenosis and spinal cords compression. In less severe deformities, a spontaneous regression may be observed. Remes suggested that 60° or more of segmental kyphosis is not likely to regress without surgical intervention.[5] Another important factor contributing to surgical treatment is the presence of clinical symptoms clearly related to the affected level.[5] Neither clear surgical guidelines are present in the literature nor studies evaluating long-term outcomes of surgery. In general, combined anterior and posterior fusion has been the most favored technique.[7] No data on the restoration of segmental or cervical lordosis are present. In our series, anterior approach with the reconstruction of the defect utilizing either autograft or mesh cage was sufficient to permanently restore the cervical sagittal profile and resolve the clinical symptoms. In the female patient, 6 years after cervical spine surgery signs of degeneration were present at the lower adjacent level, this finding might be related to relative hypermobility, due to long fusion (three levels), or might be a typical situation in DTD patients. It was shown, that cervical spine shows signs of degeneration already in the second decade of life.[8,9] Due to the rare incidence of the need of cervical kyphosis surgery the presented course of treatment may be helpful in the decision-making process.

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Conflicts of interest
There are no conflicts of interest.

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