

Case Report

# Congenital defects of C1 arches and odontoid process in a child with Down's syndrome: A case presentation

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## Abstract

We present the case of a 2-year-old child with Down's syndrome who presented to our unit with torticollis. Imaging studies revealed the rare occurrence of anterior and posterior C1 arch defects, absent odontoid process, and atlantoaxial subluxation. We managed her conservatively for 3 years without neurological deficits or worsening of atlantoaxial subluxation. We discuss the rare occurrences of anterior and posterior arch defects of the atlas, the radiological presentations of axis defects in patients, and the occurrence of atlantoaxial instability in patients with Down's syndrome. Management options with consideration to surgery in asymptomatic and symptomatic patients are also discussed.

**Key words:** C1, congenital anomalies, Down's syndrome

## INTRODUCTION

Congenital abnormalities of the atlas and odontoid process are rare occurrences, but arch defects in children with Down's Syndrome predisposes them to an increased risk of atlantoaxial instability. In this article we present a three year-follow up of a child who initially presented at the age of two with symptoms of torticollis and neck pain.

## CASE REPORT

A 2-year-old girl with Down's syndrome presented to our clinic with symptoms of acute torticollis. Clinical examination demonstrated no neurological deficit. Initial investigation included x-rays with flexion and extension views and these

revealed a 2.5 mm atlantoaxial subluxation and absent odontoid process [Figure 1]. Further imaging demonstrated a partially ossified odontoid process and absent ossification centers in the anterior and posterior arches of C1 with midline defects [Figures 2 and 3]. A further cleft was also noted within the left C1 lamina. The patient was conservatively managed for the past 3 years with regular follow-up. The child's parents were also advised that the child possesses an unstable cervical spine and therefore, restriction of certain physical activities is necessary.

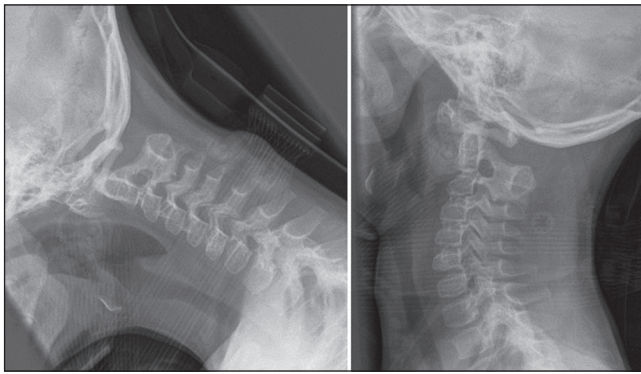
At the most recent review, the child had no other symptoms other than a mild torticollis. She continues to be managed conservatively. Imaging at the age of 5 years shows no further

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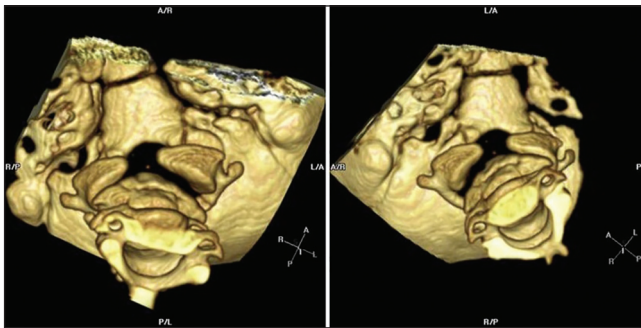
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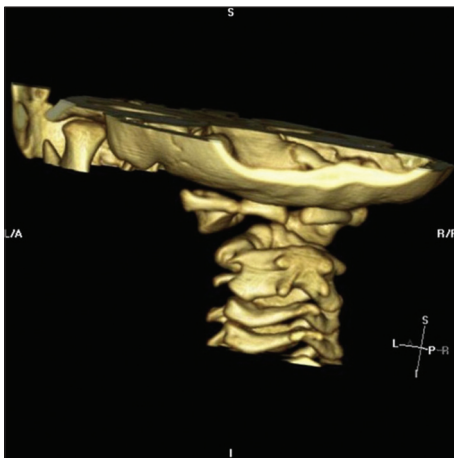
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**Figure 1: Composite of cervical spine lateral x-rays in flexion (left) and extension (right). This patient presented with torticollis. Noted is the atlantoaxial subluxation and absent odontoid process**



**Figure 2: 3D reconstruction of CT imaging of patient in rotational position. Most prominent in the imaging is the absent ossification center in the anterior arch of C1 with midline defects**



**Figure 3: 3D reconstruction of CT imaging of patient from posterior view. Noted is the absent ossification center in the posterior arch of C1 and largely absent odontoid process of C2**

ossification or bone growth to have taken place in the C1 arches or of the odontoid process. If instability increases, an occiput to C3 posterior fixation would be an alternative treatment option.

### C1 and C2 odontoid peg development

The C1 vertebra is commonly formed from three centers of ossification. One ossification center was found in the anterior arch and two were found in the posterior arch (forming the

lateral masses and the posterior arch).<sup>[1,2]</sup> The anterior arch is fused between the ages of 5 years and 8 years and the posterior arch is fused between the ages of 3 years and 5 years.<sup>[2]</sup>

In contrast to the C1 vertebrae, the C2 odontoid peg has four ossification centers present at birth: One in each neural arch, one in the body, and one in the odontoid. Between the ages of 3 years and 6 years, a secondary ossification center appears and fuses with the remaining dens by the age of 12 years.<sup>[3]</sup> Congenital anomalies of the odontoid process are rare and can be identified as os odontoideum, ossiculum terminale, aplasia-hypoplasia, and duplication of the dens.<sup>[3]</sup>

### Anomalies of atlas

Cadaveric and imaging studies have verified that the anomalies in the posterior arch range between 4% and 5%, whereas congenital anomalies of the anterior arch are rare with 0.1% prevalence in the population.<sup>[4-6]</sup>

Currarino *et al.* described five types of posterior arch of atlas defects based on the extent of the absence of the posterior arch. These types include type A: Failure of posterior midline fusion of the two hemiarches; type B: Unilateral cleft; type C: Bilateral cleft, type D: Absence of the posterior arch with a posterior tubercle present; and type E: Absence of the entire arch including the posterior tubercle.<sup>[7]</sup> Of these posterior defects, type A was the most common, with 90% of the defects attributing to this type.<sup>[1]</sup>

Anterior arch defects, although rare, are also associated with posterior arch abnormalities. In a study by Guenkel *et al.*, only two of 1,069 patients examined with computed tomography (CT) scans of the cervical spine were found to possess anterior with posterior arch defects.<sup>[4]</sup> In contrast, a study by Menezes, 2008 identified 12 out of 54 patients with anterior and posterior arch defects.<sup>[8]</sup> One study by Goel *et al.*, identified 70 out of 1,200 patients with bifid anterior and posterior arches of atlas.<sup>[9]</sup> A case study by Choi *et al.* and Petre *et al.* reported anterior and posterior arch defects in patients presenting with traumatic injury, leading to neurological injury.<sup>[10,11]</sup> A case by Thavarajah and McKenna reported congenital absence solely of the anterior arch of the atlas in a patient with no neurological deficit.<sup>[12]</sup>

In the study by Goel *et al.*, out of 70 total patients with anterior and posterior arch defects, 57 patients had bifid (two segment) anterior arch defects, 11 patients had trifold (three segment) defects, and two patients had quadrifold (four segment) defects.<sup>[9]</sup>

Atlantoaxial subluxation is a common occurrence in 15–20% of the patients with Down's syndrome, with 1% of subluxation being symptomatic.<sup>[13]</sup> In a study by Elliott *et al.* (1988), atlantoaxial instability was present in no children out of 11 under the age of 5 years, in five children out of 30 (17%) between the ages of 5 years and 9 years, and in two children out of 26 (8%) between the ages of 10 years and 14 years. In addition to atlantoaxial instability, odontoid hypoplasia was found in 15 out of 90 (17%) patients under the age of 19 years with Down's syndrome and in 14 out of 71 (20%) patients over the age of 19 years with Down's syndrome.<sup>[14]</sup>

## Management

Patients presenting with asymptomatic atlantoaxial subluxation are managed conservatively in clinics with follow-up radiographs of the cervical spine in flexion and neutral position. Children with increased risk of atlantoaxial instability are restricted from contact sports and activities such as diving, trampolining, and gymnastics. Dimar *et al.* (2012) discussed asymptomatic patients with an atlantoaxial interval between 4.5 mm and 10 mm to be managed conservatively with restriction from high-risk activity.<sup>[15]</sup> Menezes (2008) suggests that atlantoaxial instability with sagittal plane excursion of more than 8 mm requires surgical attention.<sup>[8]</sup> In contrast, Goel *et al.* (2015) managed all patients with partially and/or complete atlantoaxial instability with surgical intervention.<sup>[9]</sup> In children with symptomatic instability, younger children are opted to have autologous bone graft fusion while instrumented fusion is preferred in older children. Incorporation of the occiput is advised in conditions of atlantoaxial instability, congenital abnormality of the atlas, and/or after transoral odontoidectomy.<sup>[11]</sup>

## CONCLUSION

Congenital abnormalities of the atlas are uncommon but posterior arch defects have a higher prevalence than anterior arch defects. Children with Down's syndrome are at an increased risk of atlantoaxial instability. Imaging using radiographs, CT scans, and magnetic resonance imaging (MRI) images are essential in the diagnosis and monitoring of complications. Children with asymptomatic atlantoaxial instability are conservatively managed with close follow-up in clinics and parent education of avoidance of vigorous sports activities.

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## Conflicts of interest

The authors declare that they have no conflict of interest

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