There are very rare congenital anomalies of the posterior arch of C-1. These may appear as clefts in the arch, and their location and size vary. According to the classification of Currarino et al., there are five types of anomalies: Type A defects are defined as failure of the two hemiarches to fuse at the posterior midline (failure of the two lateral ossification centers to unite posteriorly in the midline); Type B, unilateral clefts, ranging from a small defect to a complete absence of one hemiarch; Type C, bilateral clefts of the lateral aspects associated with preservation of the most dorsal portion of the arch; Type D, the complete absence of the posterior arch with a persistent posterior tubercle; and Type E, the complete absence of the posterior arch and posterior tubercle. According to the classification of Currarino et al., there are five types of anomalies: Type A defects are defined as failure of the two hemiarches to fuse at the posterior midline (failure of the two lateral ossification centers to unite posteriorly in the midline); Type B, unilateral clefts, ranging from a small defect to a complete absence of one hemiarch; Type C, bilateral clefts of the lateral aspects associated with preservation of the most dorsal portion of the arch; Type D, the complete absence of the posterior arch with a persistent posterior tubercle; and Type E, the complete absence of the posterior arch and posterior tubercle. Few cadaveric imaging studies have been reported on the variations of such C-1 arch defects, and their clinical significance for neurosurgical practice is unclear. We therefore investigated the incidence of these congenital defects in the cervical spine on CT studies, in fresh human cadaveric cervical spines, and in dried C-1 specimens. Anatomical and imaging features of these defects and their clinical significance for neurosurgical practice were the focus of this study.

**Clinical Material and Methods**

To evaluate the annual incidence of congenital C-1 arch defects, we reviewed the institutional database and retrospectively evaluated consecutive CT scans obtained in 1104 patients (664 males and 440 females; mean age 41 years, range 10–106 years) who presented to our institution between January 2006 and December 2006. The patients presented with various medical problems, including head and neck trauma, cervical radiculopathy, posterior inferior auricular aneurysms, and head and neck pain. When a congenital defect of the C-1 posterior arch was identified on a CT scan, the patient’s medical record was also reviewed to determine his or her neurological status. Dried C-1 specimens from 166 adults that had previously been used in another study were also reviewed, but the sex and age of the specimens were unavailable. Fresh human cadaveric cervical spines from 84 adults (52 males and 32 females; mean...
age at death 56.8 years, range 32–71 years) were dissected at the level of C-1, and congenital defects of the posterior and anterior arch of the atlas were recorded. Altogether, 1354 cases were evaluated. Defects were grouped according to the aforementioned classification of Currarino et al.2

Results

Overall, in the 1354 evaluated cases (1104 patients, 166 dried specimens, and 84 fresh cadaveric specimens), 40 anomalies (2.95%) were found. On cervical spine CT scans obtained in 1104 patients, congenital defects of the C-1 posterior arch were observed in 37 (3.35%) (Table 1). Of these 37 patients, 29 (2.6% of total and 78.4% of congenital defects; 14 female and 15 male patients; mean age 36.6 years, range 15–61 years) had a Type A defect (Fig. 1A). Six (0.54%; two female and four male patients; mean age 45.3 years, range 11–60 years) had a Type B defects. Two (0.18%; one male and one female; mean age 35 years, range 34–36 years) had a Type E defect. There were no Type C or D defects. An anterior arch cleft was observed in only one patient (0.09%).

Based on their medical records, all 37 patients were neurologically intact at the time of their examination. Seven patients had undergone CT scanning for transient neck pain related to head and neck trauma. In the other 30 patients, the anomaly was discovered as an incidental asymptomatic finding. Two patients had rare multiple cleft anomalies. An 11-year-old boy had a Type B posterior cleft anomaly, an anterior cleft anomaly, and fusion of the C2–3 vertebrae (Klippel–Feil syndrome, Fig. 1B). A 36-year-old woman, who had a Type E posterior cleft anomaly, also had anomalies involving the occiput and C-2 (Fig. 1C and D).

Of the 166 adult dried C-1 specimens, two (1.2%) had a Type A congenital defect of the C-1 posterior arch (Table 1). Of the 84 human cadaveric cervical spine specimens, one (1.19%) had a Type A defect (Table 1). No anterior defects were found in the cadaveric dissections.

Discussion

Development of Atlantal Defects

During the embryological period, the atlas has three primary ossification centers,4,5,12,13,15–17,20,21 an anterior center for formation of the anterior tubercle and two lateral centers from which the lateral masses and posterior arch form. Rarely, a fourth center is present and forms the posterior tubercle. During the 7th week of gestation, the lateral centers extend dorsally to form the posterior arch. With chondrogenesis, the posterior arches are almost fused at birth. Between 3 and 5 years of age, the arches fuse completely. However, incomplete fusion may persist in 3 to 5% of patients. The anterior center usually fuses with the two lateral centers between 5 and 9 years of age.4,5,12,13,17,19

![Fig. 1. Axial (A, B, and D) and sagittal (C) reconstructed CT scans. A: Image showing a Type A defect. B: Image showing a combined anterior and Type B cleft. C: Image showing a combined Type E and occipital cleft. D: Image obtained in the same patient as shown in C demonstrating a Type E cleft.](image-url)
Congenital defects of the C-1 posterior arch

Different theories have been proposed to explain congenital defects of the C-1 posterior arch. However, the exact mechanism remains elusive. Arnold–Chiari malformations, gonadal dysgenesis, Klippel–Feil syndrome, and Turner and Down syndromes have been associated with congenital defects of the posterior arch of C-1. Martich et al.\(^9\) found hypoplasia of the posterior arch in 26% of 38 children with Down syndrome. If hypoplasia of the atlas is found in a young patient, some authors recommend ruling out Down syndrome, gonadal dysgenesis, achondroplasia, or Turner syndrome. Both of our patients with Klippel–Feil anomalies (Fig. 1) were asymptomatic and had sought treatment for other medical issues.

Incidence of Atlantal Defects

There are two main types of these anomalies: median clefts (Type A) and various degrees of posterior arch dysplasia (Types B–E). In a previous study, clefts of the posterior arch were found in 4% of 1613 cadaveric dissections, 97% of which were median clefts. This finding is consistent with our findings from the cervical spine CT scans obtained in 1104 patients: 37 patients had posterior arch defects, and of these, 29 (2.6%) had median clefts. Therefore, Type A defects represented 78.4% of all posterior arch defects. Furthermore, two (1.2%) of the 166 dried C-1 specimens and one (1.19%) of the 84 fresh human cervical segments also had such defects (Table 1). We did not investigate the association of C-1 anomalies with others such as occipital and C-2 anomalies. Therefore, whether the dried C-1 specimens were associated with other bone anomalies at different levels of the spinal column is unknown.

Total or partial aplasia of the posterior C-1 arch (Types B–E) is very rare. Its true incidence is unknown, and few articles on these particular malformations have been reported. In 1994 Currarino et al.\(^2\) evaluated 1440 lateral cervical radiographs and found one such defect (0.69%). Similarly, we found total or partial aplasia of the posterior C-1 arch (Types B–E) in only eight cases (0.72%, Table 1), and there were no Type C or D defects in either the CT cases or cadaveric specimens. Thus, the overall incidence of anomalies of the posterior atlantal arch in the 1354 evaluated cases was 2.95% (40 cases).

Anterior arch defects are even less common than defects of the posterior arch. There was one (0.09%) anterior arch cleft in the 1104 CT-documented cases, which is comparable to its reported incidence (0.1%) in other studies.\(^5,6,9,21\) No anterior arch defects were found in our cadaveric dissections.

Clinical Implications

Overall, these anomalies have been considered to be benign anatomical variations. Indeed, in all of our cases, the anomalies were found incidentally in asymptomatic patients. Although rare, acute neurological symptoms or deficits and atlantoaxial instability have been associated with these defects.\(^5,21\) A small number of patients with instability

<table>
<thead>
<tr>
<th>Author &amp; Year</th>
<th>No. of Patients</th>
<th>Type of Anomaly*</th>
<th>Neurological Deficits</th>
</tr>
</thead>
<tbody>
<tr>
<td>Richardson et al., 1975</td>
<td>1</td>
<td>D</td>
<td>transient quadriparesis</td>
</tr>
<tr>
<td>Schulze &amp; Buurman, 1980</td>
<td>2</td>
<td>C, E</td>
<td>none</td>
</tr>
<tr>
<td>Galindo &amp; Francis, 1983</td>
<td>1</td>
<td>A &amp; anterior cleft</td>
<td>none</td>
</tr>
<tr>
<td>Currario et al., 1994</td>
<td>7</td>
<td>A, C, D, E</td>
<td>none</td>
</tr>
<tr>
<td>Torram et al., 1996</td>
<td>1</td>
<td>D</td>
<td>transient quadriparesis</td>
</tr>
<tr>
<td>Sharma et al., 2000</td>
<td>3</td>
<td>2 C, D</td>
<td>transient quadriparesis, weakness in both upper limbs, intermittent tingling in both hands</td>
</tr>
<tr>
<td>Hosalkar et al., 2001</td>
<td>1</td>
<td>C &amp; anterior cleft</td>
<td>none</td>
</tr>
<tr>
<td>Urasaki et al., 2001</td>
<td>1</td>
<td>A</td>
<td>transient quadriparesis</td>
</tr>
<tr>
<td>Torriani &amp; Lourenco, 2002</td>
<td>1</td>
<td>E</td>
<td>none</td>
</tr>
<tr>
<td>Gangopadhay &amp; Aslam, 2003</td>
<td>1</td>
<td>B</td>
<td>none</td>
</tr>
<tr>
<td>Klimo et al., 2003</td>
<td>1</td>
<td>C</td>
<td>transient quadriparesis</td>
</tr>
<tr>
<td>Garg et al., 2004</td>
<td>1</td>
<td>A</td>
<td>transient quadriparesis</td>
</tr>
<tr>
<td>O’Sullivan &amp; McManus, 2004</td>
<td>1</td>
<td>D</td>
<td>none</td>
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<tr>
<td>Schrödel et al., 2005</td>
<td>1</td>
<td>E</td>
<td>none</td>
</tr>
<tr>
<td>Sagiuchi et al., 2006</td>
<td>1</td>
<td>D</td>
<td>transient quadriparesis</td>
</tr>
</tbody>
</table>

* According to Currarino et al.
may require surgical stabilization. When spinal cord compression is attributed to an osseous tubercle, surgical decompression with excision of the tubercle may be curative. The clinical presentation of reported cases has varied (Table 2). Motor and sensory deficits such as quadriparesis and paresthesia have followed minor cervical or head trauma. However, the medical records of our CT-documented cases indicated that none of these patients had chronic symptoms or neurological deficits related to their anomaly. All patients were neurologically intact, and none required surgical intervention. Only seven patients had transient neck pain after head and neck trauma probably unrelated to the C-1 anomaly. This finding is consistent with the literature. Most reported cases have either been asymptomatic or have experienced only transient neurological symptoms (see Table 2).

The discovery of a craniovertebral junction malformation that requires management includes dynamic flexion/extension imaging to assess stability or instability. Assimilations of the atlas are often hardly reducible; they evolve toward progressive instability and may require posterior occipitoatlantoaxial imaging to assess stability or instability. Assimilations are incidental findings (that is, the patients did not have symptoms related to the anomaly and thus further imaging was not performed).

Another limitation of this study is that the population estimation is based on CT scans obtained in 1104 patients. This is not a prospective population-based series, from which the true frequency can be more reasonably estimated. Nevertheless, surgeons should realize that in a very small percentage of these uncommon anomalies, the posterior arch may be incompetent and surgical planning should be tailored appropriately.

Conclusions

Most congenital anomalies of the atlantal arch appear to be found incidentally in asymptomatic patients. Congenital defects of the posterior arch are more common than defects of the anterior arch. These patients seldom need surgical treatment.

References


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