Expert's Comment concerning Grand Rounds Case entitled “Congenital C1 arch deficiency: Grand Round Presentation” (by M. Elmalky, S. Elsayed, G. Arealis, H. Mehdian doi:10.1007/s00586-013-2682-z)

Dominic N. P. Thompson

Published online: 6 March 2013
© Springer-Verlag Berlin Heidelberg 2013

Keywords Atlas · Craniovertebral junction · Cervical spine · Atlantoaxial instability

As the authors point out in this thought provoking case report [1], absence of the posterior atlantal arch, whilst rare is by no means a unique entity and will present from time to time to spinal surgeons particularly those with a paediatric interest. The radiological appearances are striking yet the clinical presentation may range from no symptoms at all to overt myelopathy. The literature on the subject is sparse and largely restricted to individual case reports and a few small series, however, guided by what literature there is and with reference to the anatomy and embryology of the

atlas as well as from experience of other craniovertebral anomalies in childhood it is possible to shed some light on the possible aetiology and implications of this anomaly.

Anatomy

Stability at the C1–C2 joint is primarily maintained by the atlanto axial joints and by the ligamentous complex comprising the alar, apical and especially the transverse ligament. None of these are likely to be compromised by absence of the posterior arch of C1, indeed the only muscle attachment to the posterior arch of C1 is the relatively small rectus capitis posterior minor muscle, and the ligamentous attachments to the posterior arch, the posterior atlantoaxial membrane and ligamentum flavum at C1/C2 contribute little to stability. Thus, on purely anatomical grounds it is perhaps unsurprising that this anomaly is not inherently unstable.

Embryology

The posterior arch of the atlas is derived from the lateral zone of the first cervical sclerotome and so its origins are embryologically distinct from the anterior arch which is derived from the hypochordal bow (which also forms the anterior tubercle of the clivus) [2]. Interestingly, the posterior arch and the lateral mass of C1 share an origin from the first sclerotome yet in this case and in the majority of atlantal arch defects the lateral masses remain anatomically normal. It is thus unlikely that this anomaly is a true aplasia dating back to the stage of somitic differentiation of development but rather a deficiency in the transformation of the chondroid precursor of the posterior arch to its fully

D. N. P. Thompson (✉)
Department of Paediatric Neurosurgery,
Great Ormond Street Hospital for Children,
Great Ormond Street, London WC1N 3JH, UK
e-mail: Dominic.Thompson@gosh.nhs.uk

ossified mature form. Such a “deficiency” may occur for example following a fracture of this brittle chondroid ring that fails to unite, the site of the fracture would then determine which variant of atlantal arch anomaly occurred, Currarino B, C or D.

As stated in this article, the posterior arch of the atlas ossifies from lateral to medial from two ossification centres, meeting in the midline thus completing the posterior arch from about 4 years of age. Thus, prior to this an incompletely ossified C1 ring may be a normal finding, indistinguishable from Currarino type A and without sinister clinical implications. By contrast, abnormalities of the C1 ring (either bifid anterior or posterior arch) that persist beyond an age where ossification should have completed may warrant a more cautious approach. Such abnormalities are not infrequently seen in the context of other conditions for example Down’s syndrome [3] and some of the bone dysplasias (such as spondyloepiphyseal dysplasia and Morquio disease). In these instances, there is commonly underlying atlantoaxial instability due to ligamentous laxity, and, rather than being the cause of instability the posterior arch defect is a consequence of that instability, the excessive movement has impeded the normal ossification of the C1 ring. This underscores the importance of flexion–extension cervical spine X-rays to exclude this possibility.

Clinical implications

It is quite clear from published reports that anomalies of the posterior atlantal arch can lead to symptoms of myelopathy, however, it is equally clear that many cases are entirely asymptomatic. It would be of obvious benefit if one could identify which types of arch deficiency were at particular risk for compression and indeed on reviewing the literature cited in this article it appears that there are two particular configurations that are common to those cases presenting with myelopathy. First, there are those cases where the posterior tubercle is separate (Currarino types C and D). This fragment may move anteriorly on neck extension resulting in compression of the thecal sac. This “posterior arch remnant” is the predominant atlantal anomaly cited in three of the references [4–6]. The second type of anomaly at particular risk of causing symptoms is what might be termed “true hypoplastic posterior arch”. This variety is not included in the Currarino classification though would appear to be the mechanism responsible for symptoms described in two of the cited references [7, 8]. In these instances, the ring might be functionally intact, however, the AP diameter of the C1 ring is reduced (the atlas becoming more ovoid than circular) thus compromising the neural canal width. Each hemilaminar may

actually invaginate into the C1 ring. Phan et al. suggests that this type of posterior arch anomaly is due to failure of dorsal expansion of the two lateral ossification centres. We have seen this not uncommonly in some of the bone dysplasias, e.g. mucopolysaccharidoses and this may go on to cause myelopathy.

The literature is insufficient to provide a clear evidence base for the management of these unusual anomalies of the C1 and so it would seem prudent to take a pragmatic approach based on first principles in conjunction with what Class III evidence there is. With that in mind the following points might be worthy of consideration:

1. The clinical status of the patient is the principle arbiter of the need for intervention.
2. In a symptomatic patient, once instability has been excluded resection of the posterior arch remnant appears effective and is unlikely to compromise stability.
3. In a symptomatic patient with instability, reduction, decompression (C1 laminectomy) and stabilisation by posterior arthrodesis should be considered. In a child, the C1 lateral masses are often inadequate to accept lateral mass screws and so it may be necessary to incorporate the occiput into the fusion construct.
4. For asymptomatic patients those at particular risk include Currarino types C and D and those with a true hypoplastic C1 arch. These should be evaluated for instability and may warrant MRI. It is this group where activities might have to be modified and where on going clinical and radiological surveillance are indicated.
5. Asymptomatic patients deemed to be of low risk include Currarino types A, B and E. In these cases, there is no evidence to support restricting activities and follow-up monitoring is probably unnecessary.

Conflict of interest None of the authors has any potential conflict of interest.

References

1. Elmalky MM, Elsayed S, Arealis G, Mehdian H (2012) Congenital C1 arch deficiency: Grand Round Presentation. *Eur Spine J*. doi: [10.1007/s00586-013-2682-z](https://doi.org/10.1007/s00586-013-2682-z)
2. Pang D, Thompson DNP (2010) Embryology and bony malformations of the craniovertebral junction. *Childs Nerv Syst* Published Online First: 31 December 2010. doi: [10.1007/s00381-010-1358-9](https://doi.org/10.1007/s00381-010-1358-9)
3. Nader-Sepahi A, Casey ATH, Hayward RD, Crockard HA, Thompson D (2005) Symptomatic atlantoaxial instability in Down syndrome. *J Neurosurg* 103:231–237
4. Sharma A, Gaikwad SB, Deol PS, Mishra NK, Kale SS (2000) Partial aplasia of the posterior arch of the atlas with an isolated posterior arch remnant: findings in three cases. *AJNR Am J Neuroradiol* 21:1167–1171

5. Klimo P, Blumenthal DT, Couldwell WT (2003) Congenital partial aplasia of the posterior arch of the atlas causing myelopathy: case report and review of the literature. *Spine* 28:E224–E228
6. Richardson EG, Boone SC, Reid RL (1975) Intermittent quadriplegia associated with a congenital anomaly of the posterior arch of the atlas. Case report. *J Bone Joint Surg Am* 57:853–854
7. Nishikawa K, Ludwig SC, Colón RJ, Fujimoto Y, Heller JG (2001) Cervical myelopathy and congenital stenosis from hypoplasia of the atlas: report of three cases and literature review. *Spine* 26:E80–E86
8. Phan N, Marras C, Midha R, Rowed D (1998) Cervical myelopathy caused by hypoplasia of the atlas: two case reports and review of the literature. *Neurosurgery* 43:629–633