Persistent torticollis, facial asymmetry, grooved tongue, and dolicho-odontoid process in connection with atlas malformation complex in three family subjects

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Abstract Congenital clefts and other malformations of the atlas are incidental findings identified while investigating the cervical spine following trauma. A persistent bifid anterior and posterior arch of the atlas beyond the age of 3–4 years is observed in skeletal dysplasias, Goldenhar syndrome, Conradi syndrome, and Down's syndrome. There is a high incidence of both anterior and posterior spina bifida of the atlas in patients with metabolic disorders, such as Morquio's syndrome [Baraitser and Winter in *London dysmorphology database*, Oxford University Press, 2005; Torriani, Lourenco in *Rev Hosp Clin Fac Med Sao Paulo* 53: 73–76, 2002]. We report two siblings and their mother, with congenital, persistent torticollis, plagiocephaly, facial asymmetry, grooved tongues, and asymptomatic “dolicho-odontoid process”. All are of normal intelligence. No associated Neurological dysfunction, paresis, apnoea, or failures to thrive were

encountered. Radiographs of the cervical spine were non-contributory, but 3D CT scanning of this area allowed further visualisation of the cervico–cranial malformation complex in this family and might possibly explain the sudden early juvenile mortality. Agenesis of the posterior arch of the atlas and bifidity/clefting of anterior arch of the atlas associated with asymptomatic “dolicho-odontoid process” were the hallmark in the proband and his female sibling. Some of the features were present in the mother. All the family subjects were investigated. To the best of our knowledge the constellation of malformation complex in this family has not been previously reported.

Keywords Facial asymmetry · Plagiocephaly · Persistent torticollis · Hypoplastic atlas · Dolicho-odontoid process · CT scan

Introduction

Failure of development and failure of segmentation of the atlas might result in abnormal articulation between the clivus, the atlas, and the odontoid process. Variations may consist of partial absence of the posterior arch of the atlas and a bifid/clefting anterior arch. A congenital clefting of the anterior arch results in two halves of the atlas vertebrae acting like a complex Jefferson fracture with lateral displacement [5, 9, 13]. This condition should be considered grossly pathological if it persists beyond the age of 3 years. Therefore, in order to investigate the cause of congenital persistent torticollis and facial asymmetry, and to assume the most probable aetiology of the sudden death of a 1-year-old first male sibling, 3D CT scanning was resorted to. This

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demonstrated an unsuspected malformed atlas in three family subjects.

The purpose of this study is to describe and illustrate the connection between congenital malformation of the cervico–cranium, and a constellation of craniofacial features. We suggest that familial agenesis of the posterior arch of the atlas and bifidity/clefting of the anterior arch of the atlas, might constitute part of a malformation complex that has not been previously reported, which in addition, had contributed in the development of congenital torticollis, facial asymmetry, grooved tongue, “Dolicho-odontoid process”, and possibly, the sudden juvenile mortality. The diagnosis of a congenitally malformed atlas should be considered in the evaluation of patients presented with plagiocephaly, facial asymmetry and torticollis. Computerised tomography should be used to confirm the diagnosis. The mode of the transmission of the disorder in this family is compatible with an autosomal dominant pattern of inheritance.

Clinical report

Patient I

This boy was first observed at the age of 9 months for mild torticollis and facial asymmetry. He was the second child of a young mother of 28 years of age, gravida 4 (abortus 1, mortality 1), married to a 31-year-old unrelated man. Her first male child had died suddenly at the age of 1 year, and no reason was identified!

The boy was born at full term, the product of an uneventful gestation. At birth his weight was 2,950 g, length 50 cm, and OFC 36 cm, and by the age of 9 months his length was 62 cm, his weight was 8 kg and his OFC was 40 cm. There was mild left-sided torticollis, plagiocephaly and facial asymmetry. Neurological investigations were carried out to exclude any neurogenic disorders, and all proved to be normal. The ophthalmologic, oto-rhino-laryngologic and cardiologic examinations were also normal as were renal ultrasound scans. The MRI confirmed the normality of the facial nerve, and the inner ears and did not show any associated Chiari malformation. His intelligence was normal and his subsequent course of development was within the normal limits as well. Laboratory studies showed normal white and red blood cell and platelet counts, normal calcium, phosphorus, and alkaline phosphatase levels. Urine aminoacids and mucopolysaccharides were normal, and he had a normal karyotype.

At the age of 13 years (Fig. 1a), his height was 119 cm (−3SD), weight 32 kg (50th percentile), and OFC 51 cm (50th percentile). The clinical features at this age were a prominent forehead, a beaked nose, low set ears, facial asymmetry, and persistent torticollis with no pain. Clinical evaluation showed no associated spasm of the sternomastoid muscle on the opposite side of the torticollis, and there was no shortening of the sternomastoid muscles. Skeletal survey did not reveal any features of significance.

At this stage, we referred him for 3D CT sagittal scan and reconstruction of the cervico–cranial junction. We noted total agenesis of the posterior arch of the atlas, a remarkable clefting deformity of anterior arch of the atlas and unexpected “dolicho-odontoid process”. The tip of the odontoid was 6.6 mm above Chamberlain’s line (Figs. 2, 3, 4); however, there was no associated symptomatology of basilar invagination.

Patient II

The sister of patient I (Fig. 1b) was the product of an uneventful gestation and was born by normal vaginal delivery at 40 weeks. She had a birth weight of 3,150 gm, a length of 48 cm, and a head circumference of 32 cm. Facial asymmetry and congenital torticollis were noted and registered in her birth notebook.

At the age of 2 years she was examined and found to have the same cranio-facial contour as her older sibling. In addition, torticollis and facial scoliosis were noted. An MRI was done and asymmetry of the skull (plagiocephaly) and a shallow posterior cranial fossa, were noted. The appearance of the facial nerve and inner ears were within normal limits, and she was of normal intelligence.

Fig. 1 Patient I (a) and his younger female sibling (patient II, b), note the facial scoliosis (asymmetry) and the torticollis

Fig. 2 3D reconstruction CT scans of patient I showed clefting/bifidity of the anterior arch of the atlas

Fig. 3 Sagittal CT scan of patient I showed congenital absence of the posterior arch of the atlas resulted in progressive upward bulging of the odontoid (6.6 mm above Chamberlain's line)

Fig. 4 T1-weighted MRI image of patient I showing no intramedullary signal abnormality

Skeletal survey was done at the age of 6 years, and nothing remarkable was noted. The torticollis and the facial asymmetry persisted.

We then performed 3D CT sagittal scans of the crano–vertebral junction, and observed agenesis of the posterior arch of the atlas and typical clefting of anterior arch like that of her brother were noted (Figs. 5, 6).

Patient III

Examination of the mother revealed a healthy woman with similar hallmarks of facial asymmetry and torticollis, and the imaging studies showed hypoplastic posterior arch of the atlas (Fig. 7).

Discussion

Embryologically, the atlas is different from the other cervical vertebrae. It is the only vertebra to develop from only two lateral ossification centres—one in each lateral mass. These nuclei fuse in the midline, posteriorly, to form the posterior arch of the atlas, whereas the anterior arch results from the anterior fusion of the nuclei with a dense band of tissue known as the hypochordal bow [20]. Haakonsen et al. [11] described the formation of a third ossification centre, and its absence in 20% of the newborns can lead to the development of a split atlas. The two lateral centres form the posterior arches with chondrogenesis, extending dorsally at 3–5 years of age, and the anterior centre unites these at 5–9 years of age. It is speculated that approximately

Fig. 5 Sagittal CT scan of the female sibling (patient II): **a** the odontoid is on the upper normal limit of Chamberlain's line; **b** agenesis of the posterior arc, whereas the anterior is preserved

72% of spinal injuries in normal children under 8 years old occur in the cervical spine [12].

Currarino et al. [6] classified the anomalous C1 arch into five types on the basis of CT and radiographs. In this classification, types C and D manifest as absence of the bilateral posterior arches and an isolated posterior tubercle. They reported six children and one adult, the mother of one of the children, with an anomalous posterior arch of the atlas. The anomalies in the seven patients were an absence of the posterior arch of the atlas in four, bilateral clefts in two, and a unilateral cleft in one. They concluded that congenital defects of

Fig. 7 Axial CT scan of patient III (mother) showed hypoplastic posterior arch

the posterior arch of the atlas may be discovered as incidental, asymptomatic findings, but symptoms might occur after trauma to the head and neck, or spontaneously. None of the patients was noted to have additional features.

Total agenesis of the posterior arch of the atlas is rare, and almost always reported incidentally, or as a consequence of trauma, or in patients with neurological deficits secondary to atlanto–axial instability [7, 14, 16, 19].

Saltzman et al. [20] reported an inherited form of cervical vertebral dysplasia, affecting 9 of 12 family

Fig. 6 Reformatted CT scan of patient II showed the clefting/bifidity of the anterior arch of atlas, and the impacted os terminale of the odontoid between the cleft of the anterior arch of the atlas and C2—this usually fuses at the age of 12 years and can be confused with fracture

members from three generations. All of the affected subjects had an abnormality of the first cervical vertebra. Some also had defects of the axis and caudal to it. The mode of transmission of the disorder was autosomal dominant, with apparently complete penetrance, but variable expressivity. None of the reported patients manifested similar features to our family. Devi et al. [8] described five children, all of whom presented with myelopathic features due to inturned two halves of the arch, but none of the reported patients manifested a syndromic association.

Some children presenting with facial asymmetry have the asymmetric crying face syndrome, which can be explained on the bases of facial paralysis affecting the elevator of the angle of the mouth. This condition has a frequency of 1 in 160 at birth, and it is known to be associated with cardiac lesions—especially a VSD—and with genito-urinary anomalies [1, 15]. Congenital facial diplegia is another clinical entity, which is characterised by unilateral or bilateral facial and abducens palsies. Möbius [17] described a syndrome of congenital bilateral facial palsy and abducens nerve paralysis, but the criteria are not compatible with those of our patients. Goldenhar [10] described a syndrome of hemifacial microsomia, a condition that affects the face bilaterally, but asymmetrically. The overall clinical and the vertebral features in this clinical entity are totally different from our reported family.

Atkin et al. [2] described patients with midline-grooved tongue, with additional findings of severe retardation, short stature and obesity. They reported 11 moderately-to-severely retarded males and 3 mildly retarded females. Our patients were of normal intelligence and had different abnormalities.

Tomographic studies on 14 patients, aged from 1 month to 24 years, presented with torticollis, using skull images for the evaluation of the cranium, endocranial base, and facial skeletal structures by Yu et al. [24] showed that the cranium and the cranial base deformation took place as early as in infancy, with the most prominent change occurring in the posterior cranial fossa. Torticollis was assumed to develop because of idiopathic fibrosis of the sternocleidomastoid; no further craniocervical malformations were described.

Asymptomatic basilar impression “dolicho-odontoid process” was found in patient I, and upper limits of normal dens projection, in the female sibling. Basilar impression is defined as being present when the tip of the dens projects more than 5 mm above a line joining the hard palate to the posterior lip of the foramen magnum (Chamberlain’s line) [4]. Upward displacement of basilar and condylar portions of the occipital bone cause an infolding of the foramen magnum,

leading to a translocation of the upper cervical spine into the brainstem. This has the potential for severe neurological consequences. It can occur secondarily to a generalised bone disease such as osteomalacia, Paget’s disease, hyperparathyroidism, rheumatoid arthritis, and osteogenesis imperfecta [18, 21–23].

We suggest that in our patients the development of the dolicho-odontoid process occurred as a result to the coexistence of three malformations: first, the presence of an aplastic posterior arch of the atlas; second, the fusion of the residual part of the posterior arch to the posterior lip of the foramen magnum; and third, the clefting of the anterior arch of the atlas. Together, these malformations were responsible for a progressive shortening of the anatomical distance between the axis and the foramen magnum, i.e. congenital depressed basiocciput.

We conclude that children with cranio-facial asymmetry and persistent torticollis should be thoroughly investigated. The hypoplastic atlas as demonstrated in three family subjects may increase the risk of atlanto-axial subluxation, rotation, or a development of Jefferson and/or odontoid fracture and possibly sudden death. Also, the presence of the dolicho-odontoid process might increase the tendency to fracture in those already susceptible, and this could result in neurological complications. This study highlights the association of plagiocephaly, persistent torticollis, facial asymmetry, and a grooved tongue, with a malformation of the atlas. The transmission of this association in the family reported here was compatible with autosomal dominant inheritance, and from our extensive search of the literatures we did not find any similar description. We wish finally, to stress the value of extensive investigations in these patients and specifically the role of the CT scans in interpreting these abnormalities.

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