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Case Report

An Adolescent Refugee with Congenital Torticollis

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Abstract

A Burmese adolescent of refugee-background was referred to the paediatric refugee health service following resettlement with congenital right torticollis not previously investigated or managed. She was symptomatic with facial asymmetry, intermittent right-sided neck pain, paraesthesias and reduced strength distally in her right upper limb. Investigations subsequently diagnosed a congenital complex cranio-cervical junction abnormality with atlantoaxial instability. Other skeletal anomalies were also detected, however no underlying genetic cause has been identified. Presumption of benign torticollis was perpetuated by communication challenges due to limited interpreter availability for primary language, interrupted education and low health literacy. The resultant delay in diagnosis and management has had negative health consequences including concerns around adequate informed consent for surgical intervention. Adolescence is a high-risk time for diagnosis and management during transition to adult hospitals and/or primary care, requiring coordination of care across jurisdictions. Risks in the transition period to adult services for this adolescent were amplified, requiring paediatric advocacy for safe linkage and resettlement assessment.

Keywords: Congenital torticollis; Adolescent; Refugee; Case report; Resettlement.

CASE PRESENTATION

A 17-year-old adolescent female of Burmese refugee-background was referred to the tertiary paediatric refugee health service regarding congenital right torticollis and right ear abnormalities identified on post-arrival screening. Preliminary screening assessments were undertaken with a Burmese in-person interpreter (second language). The family spoke a rarer dialect (primary language), accessible intermittently by telephone interpreter in Western Australia.

The patient was born overseas via uncomplicated term vaginal delivery. Right torticollis was noted at two months of age, with no formal investigation or treatment. The patient reported initially to be asymptomatic. When assessed in her primary dialect she articulated right sided neck pain, intermittent paraesthesia (right arm and leg) and reduced self-esteem. No other neurological, cognitive or developmental concerns were raised, nor relevant family history.

The patient was post-pubertal and proportionately small (height 3rd, weight 4th centile). Facial asymmetry was noted with a "cupped" appearance of her right ear, being anteriorly protuberant with a thickened helix and less defined antihelical folds. Her left jaw angle was superior to the right. Her neck was short without webbing or hairline abnormalities. There was significant right-sided neck range of motion restriction and shortening of the right sternocleidomastoid and trapezius muscles. Her spine was midline with no evidence of scoliosis or tenderness. A right sided duplicated thumb with fingernails facing outwards (Wassel-IV type) was evident. Power was reduced distally from the elbow on the right side (4+/5). No wasting or hyperreflexia was noted. Full neurological examination was otherwise normal.

Cervical spine radiographs demonstrated reduced disc space and partial fusion of the C5/C6 vertebral bodies, superior and inferior endplate wedging of C4, and dysplastic lateral atlantoaxial and right C2-3 facet joints. The atlantodental interval was widened (5mm). Neck ultrasound showed diffuse fusiform enlargement of the left sternocleidomastoid muscle (SCM). Magnetic resonance imaging and computed tomography further characterised a complex craniocervical junction abnormality with a dysplastic dens, dysplastic atlanto-axial and atlanto-occipital articulation on the right side, and a pars defect of the right C2 pedicle. Dynamic views demonstrated evidence of atlantoaxial instability, with cervical cord impingement upon the cervical medullary junction (Figure 1). There were no lumbosacral or cranial abnormalities. Screening for other anatomical abnormalities identified an additional pair of ribs; abdominal ultrasound was unremarkable. With the presence of multiple congenital anatomical abnormalities, an underlying genetic cause was suspected. The patient had a normal chromosomal microarray and was referred to the state Genetic Services. Whole-exome sequencing was normal, with sequencing of the EFTUD2 gene to assess for mandibulofacial dystosis with microcephaly pending.

The aetiology of the patient's right congenital torticollis was determined to be likely osseous with diagnosis of a congenital malformation of the C2 vertebra. Her jaw asymmetry was attributed to hemifacial hypoplasia from prolonged torticollis. The finding of enlarged left SCM was attributed to compensatory changes from longstanding untreated osseous torticollis, with concurrent congenital muscular torticollis a differential diagnosis. Due to age, the patient required referral to state adult tertiary Spinal and Neurosurgery Services. Management options were discussed initially in Burmese, with the patient waitlisted for spinal fusion surgery within 6 weeks. The family felt inadequately consented, with concerns around use of Burmese in the context of limited health literacy. Surgery has been delayed and further discussions

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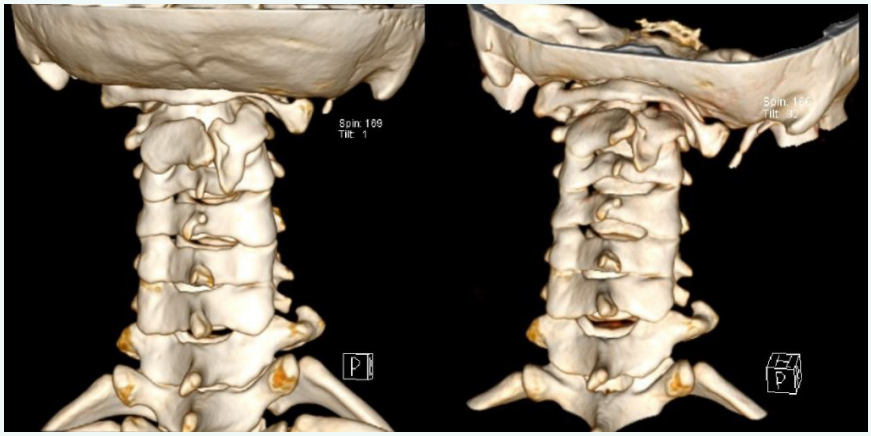

Figure: 3D reconstruction of CT cervical spine with flexion and extension views

undertaken with both neurosurgery and paediatrics in primary dialect. This enabled discussion to address the patient's concerns, appropriate safety-netting and iterative informed consent.

DISCUSSION

Congenital torticollis is a postural deformity of the neck characterised by limited neck rotation and lateral flexion [1]. In populations with adequate access to healthcare, congenital torticollis is usually identified and addressed in the first months of life [2,3]. Most cases are muscular, successfully managed with early intervention including positioning and passive stretching [2,3]. Rarer non-muscular causes include vertebral anomalies, clavicle fractures, plagiocephaly, craniosynostosis, ocular pathology or lesions of the central nervous system [1,2]. A thorough history and clinical examination including spine and neurological assessment should identify concerns for a non-muscular cause where further investigation is indicated [2,4]. The atlantoaxial joint has key roles of neck mobility and head stability and is in close proximity to several crucial neurovascular structures. Atlantoaxial instability consequently has risks of upper cervical myelopathy and other neurologic compromise, respiratory failure, vertebral artery dissection, and rarely quadriplegia or fatality [5]. Management varies with aetiology and severity of clinical and radiographic features. Definitive spinal stabilisation with fusion surgery is indicated if any neurologic deficits are present, or after failure of non-operative management [5].

Adolescence is a high-risk time for diagnosis and management during transition to adult hospitals and/or primary care, requiring coordination of care across jurisdictions. This is compounded in the post-resettlement period for adolescent refugees [6]. In this case, investigation and management of congenital torticollis was prevented due to limited overseas health access secondary to refugee status, protracted transit, interrupted education and reduced health literacy. Untreated congenital torticollis is associated with compensatory scoliosis and progression of facial asymmetry due to hemihypoplasia [7]. Permanent rotational and bending deformities of the cervical spine, most pronounced in the axis and atlas, have also been found in older children and adults with untreated congenital torticollis [8,9]. As our patient had no investigations for her congenital torticollis in early life, determination if all or any of her vertebral abnormalities can be attributed to dysplasia from untreated muscular torticollis is not possible. Muscular and non-muscular causes of congenital torticollis can present concurrently [10].

Communication post-resettlement was also impacted due to rarity of primary dialect interpretation and reduced patient/family health literacy, leading to delays in reporting neurological symptoms consistent with intermittent cord compression. If locally born, the patient's chronic neck pain and intermitted right-sided paraesthesias would have been minimised with access to adequate healthcare and intervention in early life. There were challenges of communicating diagnoses and management options in secondary language, with subsequent risk of inadequate informed consent for surgery. Appropriate use of interpreter services is therefore critical for patients of refugee background, with longer and repeat consultations often required. Receiving health professionals need awareness of barriers to health care overseas, particularly for transitioning adolescents who may have complex and often undiagnosed health concerns and need coordination of care across health jurisdictions.

IMPLICATIONS AND CONTRIBUTIONS STATEMENT

In accordance with Child and Adolescent Health Service (CAHS) ethical requirements, a CAHS patient case report consent form was completed through an interpreter. All authors contributed to manuscript preparation and revision.

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