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CLINICAL BRIEF

Congenital Torticollis Due to Sternomastoid Aplasia with Unilateral Cerebellar Hypoplasia: A Rare Association

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Abstract Congenital torticollis is most commonly caused by sternomastoid contracture. Aplasia of sternomastoid muscle causing congenital torticollis, though rare, has been reported. However the association of cerebellar hypoplasia with sternomastoid aplasia is extremely rare. The authors describe a case of congenital torticollis due to absence of the left sternomastoid with ipsilateral cerebellar hypoplasia, confirmed by MRI.

Keywords Congenital torticollis · Sternomastoid aplasia · Sternomastoid agenesis · Cerebellar hypoplasia

Case Report

A 4 –y- old boy presented with torticollis on the right side from birth. There was no history of prolonged labour or sternomastoid tumor at birth. The growth and development of the child both physically and neurologically have been normal.

On examination, the child's head was tilted to the right and the chin rotated toward the left (Fig. 1). The right sternomastoid was tight and the left sternomastoid could not be palpated (Fig. 2). There were no obvious cervical spine abnormalities. Neurologically the boy was normal. An MRI showed that the whole sternomastoid on the left was absent (Fig. 3). Incidentally the brain scan showed that the left cerebellar hemisphere was hypoplastic (Fig. 4). Though the logical treatment would be to divide the right sternomastoid, parents preferred physiotherapy as the mode of treatment.

Discussion

The most common cause of torticollis in children is sternomastoid contracture following a sternomastoid tumor in infancy. Other causes of torticollis are anomalies of cervical spine, weakness of extra ocular muscles, intermittent gastro oesophageal reflux called Sandifer syndrome and rarely due to posterior fossa tumours.

Congenital unilateral absence of sternomastoid resulting in torticollis, though reported, is rare [1]. In sternomastoid aplasia, the torticollis occurs due to uninhibited action of the normal sternomastoid muscle, the head is tilted away from the affected side and the chin points towards the shoulder of the affected side. Lung herniation into the neck associated with congenital absence of the sternomastoid muscle has been reported [2]. Unilateral congenital agenesis of sternocleidomastoid and trapezius muscles in the same child is a rare occurrence [3].

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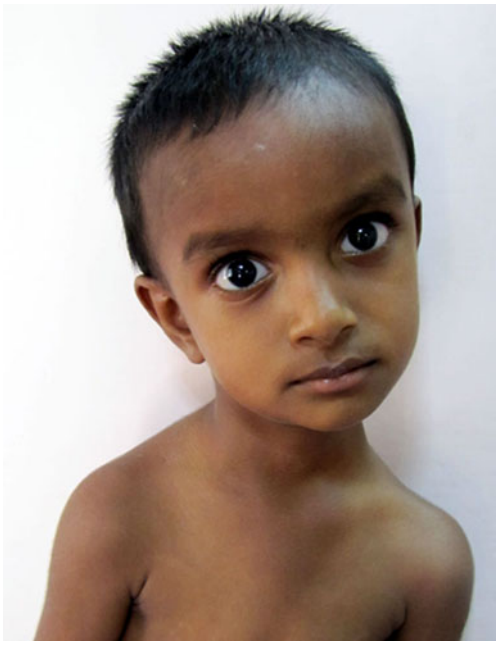

Fig. 1 Photograph of the child showing torticollis to the right side

A child with severe torticollis caused by a hereditary unilateral absence of the sternocleidomastoid and trapezius muscles has been reported. Both his father and paternal grandfather had a forme fruste, with similar but milder findings [4].

In the above case, the agenesis of sternomastoid was associated with ipsilateral cerebellar hypoplasia. The child did not manifest any neurological signs. Such association is extremely rare. The MRI scan which confirmed the aplasia of the sternomastoid, also showed the cerebellar hypoplasia.

Unilateral cerebellar hypoplasia (UCH) is a rare pathological condition characterized by the loss of volume in cerebellar hemispheres ranging from mild asymptomatic to severe symptomatic cases. Severe symptomatic cases exhibit developmental delay, speech delay, seizures, microcephaly, hypotonia, ataxia and impaired coordination, abnormal movements (tremor or titubation); hypertonia, autistic features, ocular signs such as nystagmus, strabismus, and abnormal ocular movements [5].

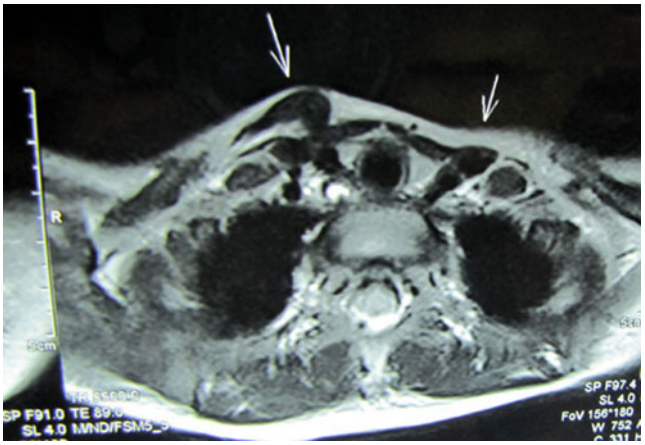

Fig. 3 MRI of the neck showing complete absence of left sternomastoid

The clinical outcome of patients varies from healthy subjects to marked developmental impairment which is related to ischemic or vascular injuries, implying a prenatally acquired disruption [6]. UCH is considered as residual change after a disruptive prenatal cerebellar insult, most likely hemorrhagic [7]. UCH has been associated with demonstration of vascular anomalies in the cerebellar and or vertebral arteries. Other types of malformations of the posterior fossa were also observed, like retrocerebellar cyst associated with obstruction of the aqueduct, cutaneous hemangioma or vascular malformation [8]. This child with cerebellar hypoplasia was neurologically asymptomatic

The case is presented for the rarity of the association of congenital torticollis with sternomastoid aplasia, and ipsilateral cerebellar hypoplasia. Such association has not been reported so far.

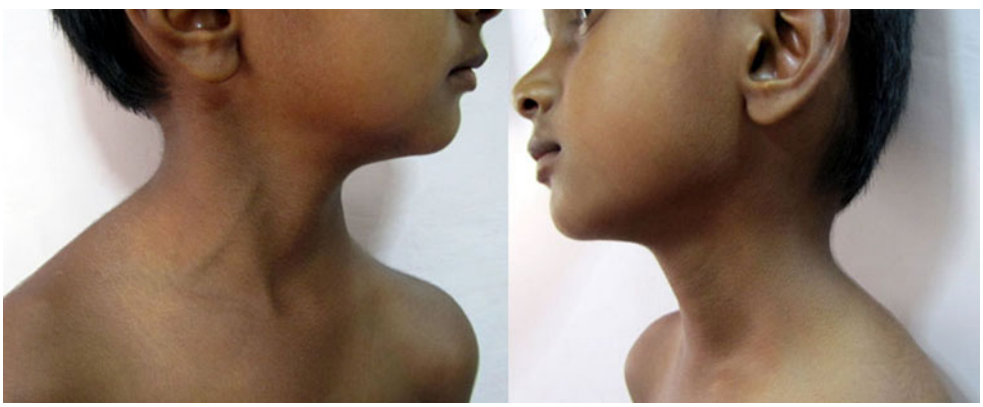

Fig. 2 Photograph of the child showing prominent right sternomastoid and aplasia of left sternomastoid

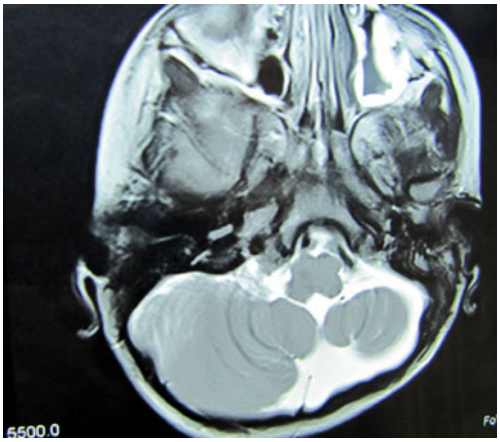

Fig. 4 MRI of brain showing hyoplastic cerebellum on the left side

Conflict of Interest None.

Role of Funding Source None.

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