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UNILATERAL AGENESIS OF THE STERNOCLEIDOMASTOID MUSCLE*

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An eight-year-old girl with an asymmetrical appearance of the neck is presented. Physical examination and ultrasonographic (USG) findings revealed the absence of the patient's right sternocleidomastoid muscle. *Key words: sternocleidomastoid muscle, congenital absence, ultrasonography.*

Congenital agenesis of some muscles has been described, but the descriptions have been based primarily on anatomical dissections. Of individual muscle defects, the pectoralis is the most common and best known, followed by the trapezius, serratus anterior and quadratus femoris, but many other muscles may be affected either singly or in combination[1]. Here we describe a patient with congenital agenesis of the right sternocleidomastoid muscle. We used ultrasonography (USG) in this case to demonstrate the agenesis.

Case Report

An eight-year-old girl was admitted to the Department of Pediatric Neurology, Hacettepe University, because of the asymmetrical appearance of her neck. The girl's parents had noticed a depression in front of her right lower neck. She was born after a normal gestation and delivery. Her developmental milestones, both motor and cognitive, appeared normal. She had one older sibling, alive, in good health, and without any known musculoskeletal defect. The family history was unremarkable. During infancy the patient had suffered from congenital stridor. She had been admitted to the same hospital at that time, but there had been no abnormal findings in her physical examination and laboratory tests. The direct laryngoscopy was normal. On examination, nothing was noted in relation to the asymmetrical appearance of her neck at that time. The stridor dissapeared spontaneously when she was 1.5 years old.

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The latest physical examination of the patient revealed a healthy child with an asymmetrical appearance in her neck (Fig. 1). There was no facial asymmetry. The right supraclavicular area appeared to be deeper than normal and bulged when she breathed. The overlying skin was normal in appearance. She was found to have full motion of her neck. The results of the neurologic examination were within normal limits. The right sternocleidomastoid muscle could not be palpated either at rest or when isometrically contracted.

Laboratory investigations revealed normal muscle enzymes, liver and kidney functions. The roentgenography of the neck showed no abnormality, while USG of the neck revealed that there was no sternocleidomastoid muscle on the right side. A representative section is illustrated in Figure 2.

Fig. 1: Photograph showing right supraclavicular depression.

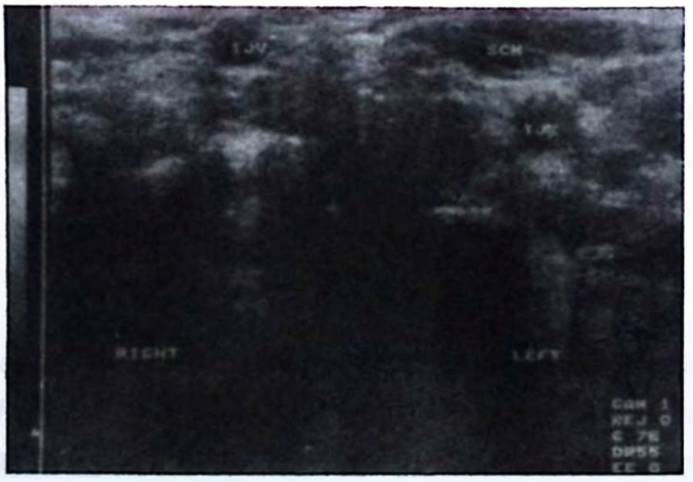

Fig. 2: Ultrasound of the neck shows absence of the right sternocleidomastoid muscle.

Discussion

The sternocleidomastoid muscle is one of the muscles of the neck. It originates from the upper part of the manibrium sterni and the medial third of the upper surface of the clavicle. The heads join and are inserted into the mastoid process and the superior nuchal line of the occipital bone. The contraction of one muscle pulls the ear down to the shoulder on the same side and rotates the head to the opposite side. In addition, the muscle can act as an accessory inspiration muscle[2].

The congenital absence of muscles is extremely rare, with an incidence of approximately one in 11,000 births[1]. Of these, the absence of the sternocleidomastoid muscle is particularly rare, representing three of the 185 cases described in the literature[3]. Congenital abnormalities of muscles tend to be unilateral and are often limited to a single muscle or a related group of muscles[1].

Although the diagnosis is not based on anatomical dissection, we believe the findings obtained by USG to be quite convincing. In some reports, magnetic resonance imaging and computerized tomography techniques are used to demonstrate muscle agenesis or anomalies without dissection[4-6].

The etiology is unknown. Some studies have revealed abnormalities and variations of muscles, vessels and peripheral nerves in trisomies, especially in trisomies 13, 18 and 21[7]. A few familial cases have been reported[8-11]. There is nothing in the history of this patient or in the clinical and roentgenographic findings to suggest a possible cause for the anomaly. Nothing can be said about the functional disability that this patient may suffer in the future.

We believe that our patient's stridor during infancy may have resulted from agenesis of the sternocleidomastoid muscle, which is an accessory muscle in respiration. During infancy the anomaly could not be found because of the shortness and increased fat tissue of the neck. The defect must be compensated by adaptive hypertrophy of the local muscular structures, which is why the stridor disappeared and we could not find any deficit on neuromuscular examination. We suggest than in cases of infantile stridor, investigations be performed to show whether any muscular anomaly is present, if routine investigations show no etiologic factor.

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