

Medial Displacement of the Internal Carotid Arteries in Velocardiofacial Syndrome

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IMAJ 2005;7:749-750

A 19 month old baby presented with gradually developing respiratory distress and stridor. Episodes of cyanosis during feeding started immediately after birth, accompanied by a weak cry. Fiberoptic pharyngolaryngoscopic examination revealed prominent pulsations along the posterior pharyngeal wall, and severe subglottic stenosis. Computed tomography-angiography of the neck demonstrated severe subglottic stenosis, bilateral enlarged, aberrant, and medially deviated internal carotid arteries, seen in the submucosa of the posterior pharyngeal wall [Figure 1].

A three-dimensional reconstruction of the CT-angiography showed medially positioned internal carotid arteries [Figure 2]. The imaging findings in this infant suggest a diagnosis of velocardiofacial

syndrome, also known as DiGeorge sequence or Shprintzen syndrome.

Velocardiofacial syndrome is a relatively common, underdiagnosed disorder that presents in approximately 1:3,000 individuals [1]. It is the most common multiple anomaly syndrome associated with cleft palate. It constitutes 8% of patients with cleft palate, including overt, submucous, and occult submucous cleft palate [2]. Approximately 5% of all patients at large interdisciplinary centers dealing with cleft palate-craniofacial entities have this syndrome [2,3]. The syndrome is caused by a deletion in the region of chromosome 22q11.2. The most common features of this syndrome are clefting of the secondary palate, cardiac defects, facial dysmorphism, learning disabilities, as well as speech and

feeding problems. Absence or hypoplasia of the adenoids is a common manifestation of the syndrome in addition to velopharyngeal insufficiency with hypernasal speech that requires surgical treatment.

Abnormal medial displacement and ectopia of the internal carotid arteries in velocardiofacial syndrome were initially reported by McKenzie-Stepner et al. [4] and D'Antonio and Marsh [5] in 1987. Embryologically, the carotid arteries originate in the third aortic arch and the dorsal aorta. Normally, the dorsal aortic root descends into the chest by the eighth week of development, thereby straightening the course of the internal carotid artery [6]. Incomplete straightening of the carotid vessels enables the embryonic angulation to persist, resulting

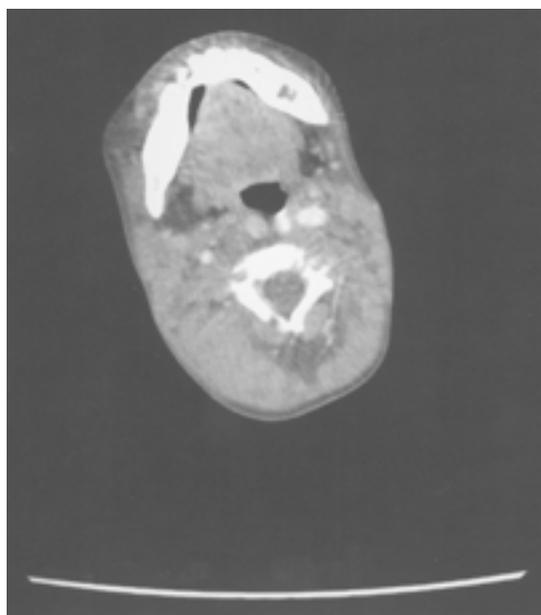


Figure 1. Axial CT-angiography of the neck showing enlarged and aberrant internal carotid arteries at the posterior pharyngeal wall.

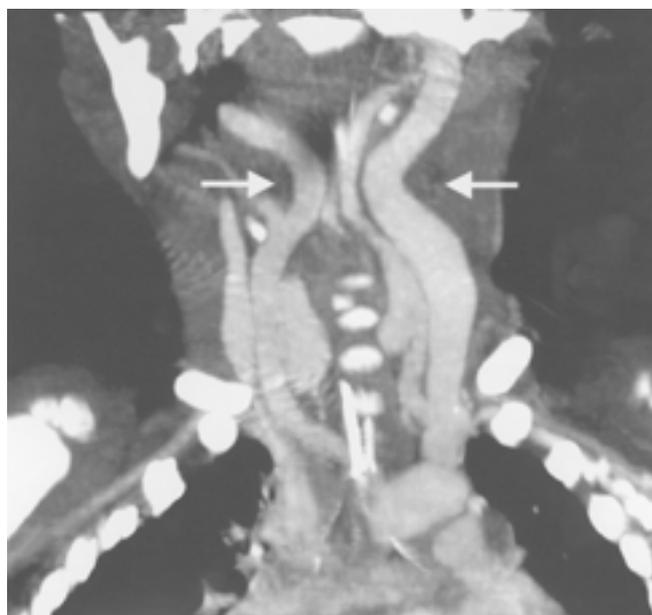


Figure 2. A three-dimensional reconstruction of the CT-angiography of the neck demonstrating medial displacement of the internal carotid artery bilaterally (arrows).

in congenitally tortuous or aberrant internal carotid arteries in the retropharyngeal space, as in velocardiofacial syndrome. Since this anomaly lies in close proximity to the midline of the posterior pharynx, it poses a significant risk for hemorrhage during both minor and major pharyngeal surgery such as tonsillectomy, adenoidectomy, and velopharyngeal narrowing procedures.

There has been some controversy over the need for preoperative imaging procedures when planning pharyngeal surgery principally for correcting velopharyngeal insufficiency. Most of the reports suggest that modern imaging techniques such as CT or magnetic resonance imaging, including contrast-enhanced magnetic resonance angiography, should be used for the detection of suspected variants in the course of cervical arteries preoperatively and for determination of the relationship to the pharyngeal flaps in case of pharyngoplasty operations.

Recognizing this syndrome as early as possible is critical to prevent delayed treatment of the hypernasal speech due to velopharyngeal insufficiency. Awareness of this syndrome is important to pediatricians, radiologists, otolaryngologists, plastic surgeons, and oral and maxillofacial surgeons because failure to recognize it prior to any head and neck surgery can result in serious iatrogenic injury including damage to the anomalous carotid arteries.

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