First & Second Arch syndrome

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Abstract

Development of the craniofacial structures is a complex process that proceeds in an orderly fashion throughout embryonic and foetal stages of formation. Craniofacial growth occurs due to a relatively rapid and orderly composition of mesodermal and cranial neural crest cells via a complex signaling network. Syndromes of the first and second branchial arches manifest along a spectrum of hypoplasia and aplasia of the structures derived from these arches. The first arches produce mainly the lower jaw – mandible, two bones in the middle ear malleus and incus, muscles for chewing that is muscles of mastication and a dedicated nerve mandibular nerve which is responsible for their supply. The second arch give rise to all of the muscles of facial expression, facial nerve, stapes bone in the middle ear, most of the outer ear, and parts of the bone above the larynx (voice box) along with palatine tonsils and middle ear cavity. The third arch produce the nerves for swallowing and the rest of the bone above the larynx that is glossopharyngeal nerve. The remaining arches give rise to the nerves for the vocal cords and other nerves and cartilage in the neck.

Some differences between abnormalities of the first and second branchial arch derivatives may reflect differences in the embryologic age at the time of the insult with respect to neural crest cell migration. Other changes are related to deregulation of cell- signalling pathways triggered by a combination of genetic and environmental factors. The manifestation and severity of the congenital abnormality depend on the alteration of gene-expression profiles.

Key words: *Branchial arch, Treacher Collins syndrome, First arch syndrome, Facial dysmorphism*

Introduction

It is a generic term that includes syndromes of malformations and congenital abnormalities involving the derivatives of the first branchial arch, with or without associated malformations. This includes mandibulofacial dysostosis, micrognathia with peromelia, otomandibular dysostosis, acrofacial dysostosis, and others.

The first and second branchial arch syndrome results in a wide spectrum of anomalies that encompass diverse, superimposed, and heterogeneous phenotypes within the so-called oculoauriculovertebral spectrum.[1,2]

In the first two months of pregnancy, tissues from each side of a fetal head and neck grow toward one another and fuse at the center line. These tissues are known as the pharyngeal, branchial or visceral arches; each arch is responsible to form the skeletal elements and crucial bones, muscles adjoining the bones as well as covering mucosa or skin. Each arch has a dedicated blood vessel and mixed nerve to supply the derivatives during this period and also to other features of the head and neck. These pairs of tissue are composed of six separate branchial/ pharyngeal arches.

The first arches produce mainly the lower jaw – mandible, two bones in the middle ear malleus and incus, muscles for chewing that is muscles of mastication and a dedicated nerve mandibular nerve which is responsible for their supply. The second arch give rise to all of the muscles of facial expression, facial nerve, stapes bone in the middle ear, most of the outer ear, and parts of the bone above the larynx (voice box) along with palatine tonsils and middle ear cavity. The third arch produce the nerves for swallowing and the rest of the bone above the larynx that is glossopharyngeal nerve. The remaining arches give rise to the nerves for the vocal cords and other nerves and cartilage in the neck.

Any factor during this period which may or disrupts/arrests this process - whether in the production, growth, or movement of key cells in the arches - will cause parts of the face to develop abnormally. The problem may occur in both sides of the face, in which case it is considered a 'symmetrical' condition, or predominantly on one

side, when it is considered asymmetrical, or uneven. Children with these disorders often have ear abnormalities and mandibular hypoplasia, an underdeveloped lower jaw.

Embryology of the First and Second Branchial arches

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Multiple craniofacial syndromes have been shown to result from an abnormality in the quantity or quality of neural crest cell migration (i.e., TCS and VCFS).[5,6]

Synchronously with the formation of the nasofrontal prominence, there is formation of 6 mesodermal arches that are separated from each other externally by ectodermal lined branchial clefts (grooves) and internally by endodermal lined pharyngeal pouches.[7]

At the end of the fourth week of gestation, four well-defined pairs of branchial arches contribute to the characteristic external appearance of the human embryo.[8] The mandibular prominence of the first arch lies caudal to the stomodeum. The maxillary prominence represents the dorsal portion of the first branchial arch and is located lateral to the stomodeum and the frontonasal prominence. The mesenchyme of the maxillary process gives rise to the maxilla, zygomatic bone, and a part of the temporal bone through the process of membranous ossification. The mandible is also formed by membranous ossification of mesenchymal tissue surrounding the Meckel cartilage, the cartilaginous mesenchymal component of the first branchial arch. The first branchial arch additionally gives rise to the muscles of mastication, the short crus and body of the incus and the head of the malleus, parts of the auricle, the anterior two-thirds of the tongue, and the mandibular branch of the trigeminal nerve. The second or hyoid arch enlarges and grows so that by the sixth week, it will overlap and cover the third, fourth, and sixth arches. The Reichert cartilage is the mesenchymal contribution to the second arch that forms the styloid ligament; the manubrium of the malleus; the long process of the incus; the head, neck, and the crura of the stapes; and portions of the body and the lesser horn of the hyoid bone. The second arch also contributes the muscles of facial expression, the stapedius, the stylohyoid, and the posterior belly of the digastric muscle. These muscles are innervated by the facial nerve, though they migrate into the territory of the first branchial arch.[9,10]

Contributory Factors

Disorders of the first and second branchial arches are generally thought to result from a combination of inadequate migration and formation of facial mesenchyma.[11] Because many structures of the head and neck migrate during fetal development, an understanding of embryologic development helps to determine the origin and nature of congenital lesions. Familiarity with craniofacial embryology and its associated effects on resultant anatomy also leads to a better understanding of the pathophysiologic basis of craniofacial syndromes. Additionally, it helps to establish a search pattern for characteristic radiologic features of many of these anomalies.

Asymmetrical pharyngeal arch syndromes

Microtia and atresia: Children with microtia and atresia do not hear well and have ears that look abnormal and less developed. In children with microtia, the outer, visible portion of the ear (the pinna or auricle) is underdeveloped. The result can be an ear that looks otherwise normal, but is too small. There can be a miniature ear that is folded and joined to the side of the head, with bits of skin attached where the ears would normally be, or skin tags on the cheek[12]. In atresia, by contrast, the ear canal, which carries sounds from the outside to the inner ear, is either closed off or absent. Microtia and atresia usually occur together. Typically they are part of hemi-facial syndrome.

Hemi facial microsomia (HFM): This is the second most common type of facial anomaly, after cleft lip and cleft palate. Hemifacial microsomia is known by a variety of other names, including craniofacial microsomia, first and second pharyngeal arch syndrome, Goldenhar syndrome, and lateral facial dysplasia. It occurs when soft tissue and bone from the pharyngeal arches on one side of a child's face fail to develop fully. The result is that facial and jaw bones in the area are smaller than normal. Although it generally occurs on just one side of the face, it can also appear on both sides. Children with HFM have several common facial features. They may have one-sided (unilateral) or two-sided (bilateral) underdevelopment of the eye, cheekbone, lower jaw, facial nerve and muscles. Bits of skin and cartilage may occur in front of the ears. There may be hearing loss caused by the underdevelopment of the middle ear. The soft palate (the fold of tissue at the back of the mouth that separates the mouth from the nasal cavity) may move to the unaffected side because of muscle weakness on the affected side[12]. The tongue may be small with nerve weakness on the affected side. In about 40 percent of children with HFM, the nerve responsible for moving the facial muscles is weak on the affected side or, rarely, on both sides. About a third of children with the disorder have macrostomia, literally 'large mouth,' because of an opening at the corner of the mouth. Typically, microtia and atresia are part of Hemifacial syndrome.

Symmetrical pharyngeal arch syndromes

Treacher Collins ("mandibulofacial dysostosis, or MFD") syndrome: Unlike the asymmetrical conditions described above, children with Treacher Collins syndrome have underdeveloped structures on each side of the face, usually to the same degree. And, unlike some children with asymmetrical conditions, children with Treacher Collins syndrome do not have weakness of the facial muscles[3]. The features of Treacher Collins syndrome include some or all of the following: downward-slanting eyelids; notching (Colobomas) of the lower eyelids; underdevelopment of the bones of the face, including the cheekbones (zygomas), lower jaw (mandible), and upper jaw (maxilla); an abnormal bite (malocclusion); a smaller than average face; underdeveloped and/or malformed ears; hearing loss to a variable extent, which is often severe, due to a narrowing or absence of the ear canal. A very rare condition that is similar to Treacher Collins syndrome is known as Miller or Wildervanck-Smith syndrome. Its characteristics include underdeveloped cheekbones, an abnormally small jaw, a cleft palate, small, protruding "cup-shaped" ears, and drooping of the lower eyelids. In addition, some children with the syndrome have incomplete development of one or more of the limbs, webbing of the fingers or toes, absence of the little fingers or toes, and underdevelopment of bones in the forearm. The condition does not, however, affect a child's intelligence.

Nager syndrome: Children with this condition share many of the problems of those with Treacher Collins syndrome. It is distinguished by flat cheeks, down-slanting eyes, almost total absence of eyelashes, low-set, cup-shaped ears, and a very small lower jaw. Children with the condition also have underdeveloped thumbs and forearms and may have genitourinary abnormalities or heart problems.

Summary

Craniofacial abnormalities especially congenital along with dysmorphism and with some functional impairment are not very uncommon and one should always keep craniofacial syndromes in mind while dealing such cases. One of the most important causes is branchial arch syndromes and their knowledge remains the keystone for their better understanding about the causation, treatment planning and rehabilitation for future.

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