

*Tuesday, February 24, 8:00AM-12:00PM
Plenary Session*

Here's Looking at You: Defects of the Head and Neck

Moderator: Angela Lin, Massachusetts General Hospital, Boston, MA

Embryology of the Head and Neck and Associated Birth Defects

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Prior to completion of neural tube closure in the 3rd week (after fertilization) of development, neural crest cells migrate from the neural folds into the craniofacial region. These cells will form all of the bones of the face and are extremely important for normal development of this area. Insults to these cells are responsible for many birth defects, including oculoauriculovertebral syndrome, Treacher Collin syndrome, Robin sequence, 22q deletion syndrome (DiGeorge anomaly, etc.) and many facial clefts. Neural crest cells also participate in septation of the heart and this contribution together with their role in craniofacial development explains why many children with facial abnormalities also have cardiac malformations. While crest cells form the bones, paraxial mesoderm (from somites along the neural tube) forms part of the skull and all of the voluntary muscles.

By the end of the 4th week, the neural tube has closed and a number of swellings appear laterally on both sides of the pharynx and cranially in the prospective facial region. Those that are positioned laterally resemble the gill bars of fish with clefts between each pair of bars. The bars are called arches and the clefts, which are only indentations and do not communicate directly with the pharyngeal space like they would in fish, are called clefts. Opposite the clefts inside the pharynx are similar indentations called pouches. Each of these structures will contribute specific derivatives to the head and neck region. For example, the 1st arch gives rise to the maxilla and mandible, the 2nd arch to part of the hyoid bone, etc. The first cleft forms the external auditory meatus, while the remainder disappear. The first pouch forms the auditory tube and middle ear cavity, while the remaining pouches give rise to endoderm derived glands in the region, including the tonsils, parathyroids, and thymus (The thyroid gland is derived from the floor of the pharynx).

Swellings in the facial region include, the maxillary, mandibular, and frontonasal prominences. In the 5th week 2 other prominences appear on each side of the frontonasal prominence: These are the medial and lateral nasal prominences. Derivatives from all of these prominences form the nose, upper lip and maxilla, and the mandible. Many of these prominences must grow together by merging or fusing and, if these phenomena are inhibited, then facial clefts occur. For example, fusion between the medial nasal prominences and the maxillary prominences forms the lateral portion of the upper lip and maxilla, whereas merging between the 2 medial nasal prominences forms the medial portion of the upper lip and philtrum. Inhibition of the former results in a lateral cleft lip; whereas inhibition of the latter causes a medial cleft lip. These and other embryologically related defects will be discussed.