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Commentary

Screening of Craniofacial Malformations and its Genetics

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DESCRIPTION

Craniofacial malformations include a variety of abnormalities, including cleft lip with or without cleft palate, craniosynostosis, microtia, and hemifacial microsomal. All of these abnormalities may be isolated or part of a defined genetic syndrome.

Craniofacial microsomal is a term used to describe a variety of abnormalities that primarily affect the development of the skull (cranium) and face before birth. Microsomal means an abnormally small body structure. Most people with craniofacial microsomia have a difference in the size and shape of facial structures between the right and left sides of the face (facial asymmetry). About two-thirds of cases have abnormalities on both sides of the face; these manifestations are often different on each side. Others have craniofacial microsomal that affects only one side of the face. The facial features of craniofacial microsomal often include underdevelopment on one side of the upper or lower jaw (maxillary or mandibular hypoplasia), which can lead to dental problems and difficulty in eating and drinking, speaking. In severe cases of mandibular hypoplasia, breathing may also be affected. The craniofacial complex is a type of bulletin board containing information about gender, health, ancestry, genes, and environment. A thorough knowledge of the genes underlying craniofacial morphology is fundamental to understanding craniofacial biology and evolution. These genes may also provide an important basis for practical efforts such as DNA-based face prediction and phenotype-based face diagnosis. We focus on different sources of knowledge regarding genes that influence craniofacial development patterns. Although great success has been achieved using these sources in both methodology and biology, many challenges remain. Among these, the main ones are precise phenotyping techniques and efficient modeling methods.

- Mouse embryology, evolution, and genetics shape our understanding of head development the most anatomically complex part of the body-and shed light on craniofacial disorders in People.
- Human craniofacial disorders arise due to changes in specific embryonic processes, such as brain pattern, cell migration, tissue fusion, and skeletal differentiation.
- Major craniofacial disorders are divided into several categories, depending on their type of malformation. Holoprosencephaly, cleft lip and palate, malformations of the cranial vault (such as craniosynostosis), and malformations of the first and second branch arches, are the basis of Treacher Collins and Treacher Collins syndromes.
- Loss of function mutant mice identify the genes responsible for these malformations and provide functional information on genes required for normal craniofacial development.
- The types of genes that cause these disorders range from transcription factors and signaling molecules, the loss of which causes distinct pattern defects, to genes required for heredity, cell proliferation and migration.

The most common mechanism for craniofacial abnormalities in mice and humans is haplo insufficiency, although gain-of-function mutations in the fibroblast growth factor receptor genes are the basis for most cranial fusion syndromes in humans, such as Apert and Pfeiffer syndromes. Studies of craniofacial anomalies have shown that they are often the result of subtle changes in cell division in the cranial mesenchyme rather than underlying structural defects. This discovery could make it difficult to identify the genetic factors that determine the normal variation in facial appearance.

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