Masticatory Muscle Defects in Hemifacial Microsomia: a New Embryological Concept

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Craniofacial surface setting and skull 3D reconstructions of a patient with hemifacial microsomia

- Hemifacial microsomia corresponds to human latero-facial congenital anomalies affecting Cranial Neural Crest Cells (CNCC) derivatives of the first pharyngeal arch (PA1) (ie. the ascending mandibular ramus, the temporo-mandibular joint, the external and middle ear).

Hemifacial microsomia : skeletal and muscular jaw defects

- The masseter muscle is absent in the affected side in 7 patients.
- CNCC are a necessary source of molecular cues essential to maintain the myogenic program in CMMC leading to masticatory muscles formation: genetic lesions affecting only CNCC can prevent muscularization of the jaws (Ref 1).

Embryonic cellular mechanisms at the origin of masticatory muscle formation

- CNCC give rise to skeletal components of the head while craniofacial muscles derive from Cephalic Myogenic Mesodermal Cells (CMMC).

Method

Craniofacial CT-scan and skull and masticatory muscles 3D reconstructions of a patient with hemifacial microsomia (A-B) right (unaffected side) and left (affected side) view of a skull 3D reconstruction. (C) CT-scan section (at the indicated level in A and B) with the defined masticatory muscles. (D-E) right and left view of muscular jaw defects 3D reconstruction.

The main three groups of masticatory muscles

- The absence of masseter is correlated neither with the age of the patients nor with the volume and shape of the affected ramus.

The muscular defects in hemifacial microsomia might derive from an anomaly in the CNCC/CMMC interaction

- Recently, we demonstrated that PA1 CNCCs, which give rise to skeletal element of the jaws, are required for the differentiation and patterning of PA1 CMMCs leading to masticatory muscle formation (Ref 1).

Results

- Based on experimental results from animal models, we propose that the hypoplasia or agenesis of masticatory muscles derives from a defect in the CNCC/CMMC communication during early embryonic development (Ref 2).

To evaluate the involvement of CNCC/CMMC interaction in human craniofacial development, we performed a quantitative analysis of masticatory muscle and mandibular bone volumes on craniofacial CT-scans from 8 children affected by hemifacial microsomia.

The size of the hypoplastic structures are approximately 50% significantly reduced

- Another fact supporting an early defect is continued growth of the hypoplastic muscles during postnatal development, suggesting that their basic physiology is preserved and that they do not regress due to the abnormal skeleton.

- This new embryological concept could contribute to improved understanding of the aetiology for hemifacial microsomia, and it might be useful for prenatal diagnosis.

Linear regression analysis of the volumes of affected versus unaffected masticatory muscles and hemi-mandible volumes

- In all cases, the pterygoid and the temporal muscles are either reduced or absent.

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References