

A Morphological Classification Scheme for the Mandibular Hypoplasia in Treacher Collins Syndrome

Cassandra A Ligh MD, Jordan Swanson MD, Jason Yu MD DDS, Fares Samra MD, Scott P Bartlett MD, Jesse A Taylor MD

Background: Mandibular hypoplasia is a hallmark of Treacher Collins syndrome (TCS), and its severity accounts for the main functional morbidity. There is scant literature quantifying and characterizing the mandibular deformity in TCS, and the purpose of this study is to develop a morphological classification scheme of the TCS mandible.

Methods: We identified 20 patients with TCS, ages 1 month to 20 years, with at least one 3D-CT prior to mandibular surgery. We examined 28 3D-CT scans from 20 patients and ordered them from least to most severe phenotype. We then performed a rigorous morphological analysis of the mandible and its relation to the face and skull base. The mandibles were then categorized based on morphology of the condyle, Co-Go-Me angle, and SNB.

Results: TCS mandibles demonstrated three consistent characteristics: a large antegonial notch, a steep mandibular plane angle, and retrogenia. TCS mandibles were graded from I to IV based on degree of condylar hypoplasia (I=normal, II= morphologically normal but small, III=condylar remnant that may not translate to the glenoid fossa, IV = No condyle), the Co-Go-Me angle in degrees (I= <150, II= 151-160, III= 161-170, IV= > 170 degrees), and SNB in degrees (I=>67, II=62-67, III=56-61, IV =<55). The overall mandible classification was determined by the median value among the three characteristics. Among the twenty-eight 3D CT exams studied, 12 (43%) were class 1 (least severe), 10 (36%) were class 2, 4 (14%) were class 3 and 2 (7%) were class 4 (most severe). Three patients had at least 3 longitudinal scans encompassing 5 to 11 years of growth. Despite increasing age, mandibular severity remained stable in those patients.

Conclusion: This provides a classification scheme of the TCS mandible in the era of 3D-CT. It takes into account three key morphological features of the TCS mandible—degree of condylar hypoplasia, Co-Go-Me angle, and SNB—that each appear to correlate with overall phenotype. While there is a natural progression of the mandible with age, we believe the mandibular deformities seen in longitudinal scans may also be inherent to the primary pathology of TCS. Further work is needed to determine the classification scheme's validity, generalizability, and overall utility.