A Range of Zygomatic Hypoplasia Exists in Treacher Collins Syndrome

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Background: Bilateral zygomatic hypoplasia characterizes Treacher Collins syndrome (TCS), and the susceptible malar subcomponents have been previously tabulated.[1] We sought to morphologically classify the pathology, volumetrically assess the TCS malar body versus norms, and analyze for symmetry.

Methods: Demographic and computed tomography data were recorded. The zygomata were digitally segmented using three-dimensional planning software (Materialise Surgicase CMF). Each zygoma was classified into one of three groups, based on the amount and location of agenesis. Volumes (mm^3) were calculated and sidedness ratios compared using two-sided t-tests.

Results: 58 sides were identified (24 TCS: 34 controls), both groups a mean age of 60.0 months and normally distributed. The dysmorphology ranged from small and partially intact cheekbones (Figure 1), to nearly-complete aplasia. Based on our classification, the degree of zygomatic deformity in our cohort was 17% type I (mild), 21% type II (moderate) and 63% type III (severe). The mean TCS zygoma was significantly smaller than controls: 851 ± 1004 mm^3 versus 2820 ± 868 mm^3 ($p<0.0001$). The sidedness ratio was 0.65 ± 0.28 for syndromic patients (Figure 2), compared to 0.97 ± 0.02 for controls ($p=0.002$).

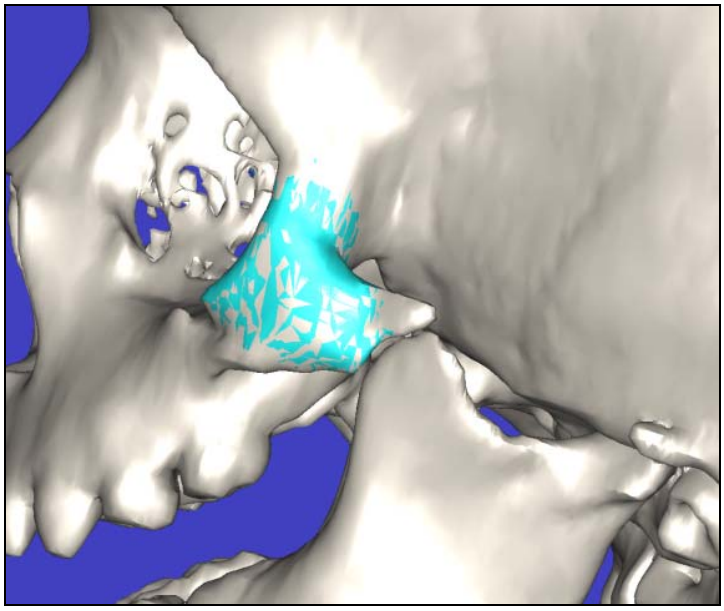

Figure 1. Example of malar bone hypoplasia in Treacher Collins syndrome.

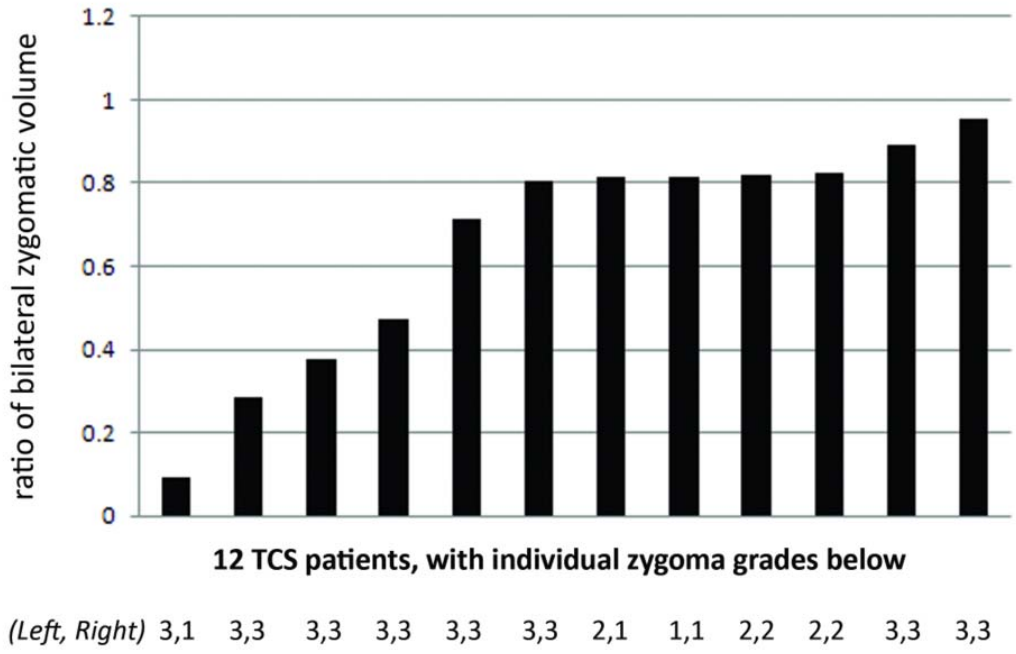

Figure 2. Zygomatic asymmetry and variability.

Conclusions: This study classifies the morphologic spectrum of Treacher Collins malar deformity and assesses the volume and sidedness. We found that an increasing grade of deficiency inversely correlates with volume. Structure is absent or significantly hypoplastic in a progressive fashion, beginning with the 1) zygomatic arch, followed by the 2) zygomaticosphenoid process, and lastly, in the most severe circumstances, the 3) zygomaticofrontal and zygomaticomaxillary processes. The malar bones, especially type III, are volumetrically deficient compared to controls. The type III subtype was most frequent in our series, in line with the near-total zygomatic agenesis thought to represent the syndrome. Though bilaterally affected, the amount of zygomatic hypoplasia from side-to-side is asymmetric in TCS.

References

1. Marsh, J.L., Celin, S.E., Vannier, M.W., Gado, M. The skeletal anatomy of mandibulofacial dysostosis (Treacher Collins syndrome). *Plast Reconstr Surg* 78: 460-470, 1986.

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