

3D morphometric evaluation of the face in Marfan syndrome: a better definition of dysmorphic features

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Marfan syndrome (MFS) is a rare genetic disorder of connective tissue caused by mutations in the *FBN1* gene, which result in alterations of fibrillin-1 and dysregulation of TGF- β bioavailability in the extracellular matrix. MFS is a clinical diagnosis. It is crucial to prevent acute aortic dissection; nevertheless it is difficult, due to the high degree of clinical variability of the disease. A quantitative definition of craniofacial abnormalities associated with MFS is not available and they are usually evaluated through a qualitative impression. The study aimed to better characterize the facial phenotype associated with MFS, identifying new morphometric features useful for the diagnosis of the disease. 3D facial images of 61 Italian subjects diagnosed with MFS, aged 16-64 years (21 males, mean \pm SD age 38 ± 15 years; 40 females, mean \pm SD age 41 ± 13 years), and divided in 6 non-overlapping age groups, were obtained by stereophotogrammetry [1]. From the coordinates of 50 soft-tissue facial landmarks, linear distances and angles were measured; z score values were calculated comparing patients with healthy Italian reference subjects (400 males, 379 females), matched for gender and age group. Statistical comparisons were performed by Student's t test. All subjects with MFS showed a greater facial divergence ($p < 0.001$; mean z score = +1.9) and a reduced facial height index ($p < 0.001$; mean z score = -1.9), being both the values mostly influenced by a shorter mandibular ramus ($p < 0.001$; mean z score = -1.9) and an overall mild but significant increase of facial height ($p < 0.001$; mean z score = +1.2). Gender differences or age-specific trends were not observed. Quantitative facial abnormalities pointed out in the current study enrich the information about the phenotypic expression of MFS and suggest their usefulness in the recognition of the disease.

References

- [1] C. Sforza et al. (2013) Soft-and-hard tissue facial anthropometry in three dimensions: what's new. *J Anthropol Sci* 91: 59-84.

Keywords

Facial anthropometry, Marfan syndrome (MFS), stereophotogrammetry