

The face in Marfan syndrome: a 3D morphometric study

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Marfan syndrome (MFS) is a rare congenital disorder of the connective tissue mainly caused by mutations in the *FBN1* gene, resulting in an altered assembly of extracellular matrix microfibrils and TGF-beta signalling dysregulation. Major clinical manifestations of MFS involve the skeletal, ocular, and cardiovascular systems, with a high risk of life-threatening aortic dissection and rupture. An early recognition of the disorder is essential, but it could be difficult, due to the variable phenotypic expression of the disease and the current incomplete sensitivity of molecular genetic testing of *FBN1*. It has been suggested that craniofacial dysmorphism associated with MFS could facilitate obstructive sleep apnea, which in turn may promote aortic dilation. The study aimed to investigate the face in MFS through a 3D not invasive approach [1], identifying new morphometric features which could facilitate the early diagnosis of the disease. The 3D coordinates of 50 anatomical facial landmarks were obtained using a stereophotogrammetric system in 68 Italian subjects diagnosed with MFS, aged 4-64 years (27 males, mean \pm SD age 29.6 ± 18.2 years; 41 females, mean \pm SD age 37.2 ± 15.5 years). Subjects were divided in 11 non-overlapping age groups. Facial linear distances and angles were measured; z score values were calculated comparing patients with healthy Italian reference subjects (347 males, 388 females), matched for gender and age. Subjects with MFS showed a shorter mandibular ramus than controls (mean z score = -1.9), a greater facial divergence (mean z score = +2.0), a reduced ratio between posterior and anterior facial height (mean z score = -1.9), and a reduced ratio between facial width and facial height (mean z score = -1.5), together with an expected but overall mild increase of facial height (mean z score = +1.3). Noteworthy gender differences or age trends were not observed. Facial abnormalities pointed out in the current study could represent phenotypic traits of MFS; since they were observed also in young patients, their detection could facilitate the early recognition, management, and follow up of the disease. These promising findings need to be confirmed extending the study on more patients.

References

- [1] Sforza et al. (2013) Soft-and-hard tissue facial anthropometry in three dimensions: what's new. *J Anthropol Sci* 91: 59-84; doi:10.4436/JASS.91007.

Keywords

Anthropometry; Marfan syndrome (MFS); facial morphology; stereophotogrammetry.