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Association between morphometric facial features and other systemic manifestations in patients with Marfan syndrome

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Marfan syndrome (MFS) is a rare hereditable disorder of connective tissue which results from mutations in the gene encoding fibrillin-1 (FBN1), an extracellular matrix component found in non-collagenous microfibrils. To date, MFS is a clinical diagnosis mostly based on family history and alterations in skeletal, ocular and cardiovascular systems. Early recognition of MFS is essential to prevent its most severe complications but it can be difficult, because of the phenotypic variability of the syndrome. In a recent study we analyzed facial dysmorphism in MFS, defining some quantitative morphometric features which could elude a mere visual inspection of the patients [1]; in the current study we investigated their association with other systemic manifestations of the syndrome. Facial linear distances and angles of 41 Italian adult subjects with MFS (14 males, 36±16 years; 27 females, 41±14 years) were computed from the 3D coordinates of soft-tissue landmarks obtained by stereophotogrammetry [2]. Corresponding z score values were calculated comparing patients with 779 healthy reference subjects matched for sex and age. For each facial measurement, patients were assigned into two groups on the basis of z-score values lower or higher than ½1.5½. Patients underwent multidisciplinary clinical examinations in order to evaluate the presence/absence of systemic signs of the syndrome, according to revised Ghent nosology. Patients with ectopia lentis (61%) or pectus deformities (51%) showed a more reduced facial width/height ratio (Student's t test, $p<0.05$), the value being mainly influenced by a longer face in patients with ectopia lentis ($p<0.01$) and a narrower face in patients with pectus deformities ($p<0.01$). A significant association between ectopia lentis and facial width/height ratio was found (Fisher's exact test, $p<0.05$). Results are promising and suggest that quantitative features of the face should be included among information which clinicians should be aware to improve the recognition of MFS; nevertheless they need to be confirmed on more patients.

References

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Key words

Facial anthropometry, Marfan syndrome (MFS), stereophotogrammetry, clinical correlation.

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