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3D facial features in Andersen-Tawil syndrome: a family report

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Andersen-Tawil syndrome (ATS) is an inherited potassium channelopathy affecting cardiac and skeletal muscle, which results in periodic paralysis and symptomatic arrhythmias. Distinctive facial and skeletal features also characterize the disorder. Variability in expression of heterozygous mutation in the KCNJ2 gene and the rarity of the syndrome (estimated worldwide prevalence 1:100.000) may lead to misdiagnosis. The recognition of facial dysmorphic features can improve the early diagnosis of ATS. However, facial features may be mild and they may be easily overlooked on routine physical examination. Today a detailed evaluation of craniofacial structures can be performed through the application of noninvasive 3D image acquisition systems. The study aims to better characterize the facial phenotype of ATS in order to improve the early diagnosis of the syndrome. We studied 4 members of a family with genetically confirmed diagnosis of ATS. Computerized acquisitions of the 3D facial image through stereophotogrammetric procedures were performed on a 39-year-old mother and 3 of her 4 sons (aged 5, 12, and 14 years respectively, the youngest one with a different father) without previous history of craniofacial trauma. From the digital 3D coordinates of 50 landmarks, facial linear distances and angles were compared with those collected in healthy subjects matched for age and sex. All members of the family showed 1) larger intercanthal width and smaller length of eye fissure than controls, 2) larger alar base width and smaller nasal tip protrusion, 3) smaller forehead and lower facial height including shorter and wider philtrum, 4) smaller anteroposterior facial distances and wider facial convexities in the horizontal plane. The present family study allowed to better characterize the facial features associated with ATS. The results are in accordance with the already known facial features of the syndrome, except for the extension of the forehead. 3D morphometric facial analysis pointed out characteristics of the philtrum which were not described previously in subjects with ATS, and could be more than a mere somatic familial trait.

References

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Keywords
Anthropometry; Andersen-Tawil syndrome; face; stereophotogrammetry.