Robustness of distinctive facial features in Prader-Willi Syndrome: a stereophotogrammetric analysis and association with clinical and biochemical markers in adult individuals .

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Background: Prader-Willi syndrome (PWS) is a rare genomic imprinting disorder associ ated to a complex neurodevelopmental phenotype and a distinctive facial appearance. The study investigated the relationships between the quantitative facial dysmorphism in PWS and clinical and biochemical markers of the disease and its treatment.

Methods: facial images of 15 Caucasian adult individuals with PWS (8 males, 42 ± 5 years; 7 females, 37 ± 8 years; BMI 38.87 ± 8.92 kg/m^2) were acquired through stereophotogrammetry. From the 3D coordinates of 38 landmarks, linear distances and angles were calculated; they were expressed as z-score values by referring to 403 healthy subjects matched for age and sex and compared by Student's t-test with Bonferroni correction for multiple testing. Patients underwent auxological and biochemical assessment of endocrine/metabolic dysfunction and nocturnal respiratory function. An exploratory correlation analysis was performed to investigate their associations with the facial phenotype; uncorrected *p*-values were used.

Results and conclusions: individuals with PWS showed decreased bifrontal diameter, facial depths, palpebral fissures, mandibular ramus length, lower vermillion height, and modified relative position of exocanthia and nasion. Since these characteristics did not show any associations with clinical and biochemical markers of PWS, they could constitute robust distinctive facial features and contribute to the diagnosis of the disorder. Individuals with PWS showed also a larger mandibular width with smaller gonial angles, thinner upper vermillion, greater inclination of the orbit relative to the Frankfurt plane, and a smaller angle of the auricles versus the facial midplane. Relationships between these facial anthropometric features and body composition, glucidic metabolism indexes, nocturnal hypoxemia episodes, or duration of GH treatment were found, suggesting their potentially useful role in the clinical monitoring and management of the disease. However, they need to be confirmed by subsequent dedicated studies.