3D stereophotogrammetric facial analysis of SMAII patients

Valentina Pucciarelli[1], Chiara Mastella[2], Simona Bertoli[3], Katia Alberti[2], Ramona De Amicis[3], Alberto Battezzati[3], Giovanni Baranello[4] and Chiarella Sforza[1]

[1] Dipartimento di Scienze Biomediche per la Salute, Università degli Studi di Milano, Milano, Italy
[2] SAPRE (Settore Abilitazione Precoce Genitori), Fondazione IRCCS Ca' Granda, Ospedale Maggiore Policlinico, Milan Italy
[3] ICANS (Centro Internazionale per lo Studio della Composizione Corporea), Dipartimento di Scienze per gli Alimenti, la Nutrizione e l'Ambiente, Università degli Studi di Milano, Milano, Italy
[4] UO Neurologia dello Sviluppo, Fondazione IRCCS Istituto Neurologico Carlo Besta

Spinal muscular atrophy (SMA) is a rare neurodegenerative disease, due to autosomal recessive mutations on SMN1 gene. It is clinically classified into 4 phenotypes (SMAI-SMAIV) and it is characterized by muscular weakness and atrophy of the voluntary muscles of the legs, arms and trunk [1]. No information is available about soft tissue facial characteristics in these patients. To better define their facial phenotype and to evidence possible modifications, the 3D facial reconstructions of 12 male SMAII patients (3-8 years) were evaluated. All of them were able to sit, but not to walk independently and had respiratory problems, chewing and swallowing difficulties. The facial reconstructions were obtained through a stereophotogrammetric system, after the non-invasive identification of 50 facial reference landmarks, whose 3D coordinates were used to calculate a series of linear measurements. Data were compared with those of healthy controls, matched for age and sex, through the calculation of z-score values [2]. Results show that patients have larger skull base, mandibular and facial widths (z-score = 1.5, 2 and 1.8 respectively), together with an increased height of the nose (z-score = 3) and mandibular body length (z-score = 2.1). The mandibular ramus length is reduced (z-score = -2.6). Results are of interest to define the facial anatomy of these patients, since a detailed knowledge of their facial features can be useful to create ergonomic devices, as respiratory masks, that these patients must daily use.

References

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Keywords

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