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Abstract

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Apert syndrome, or acrocephalosyndactyly type I, is a craniofacial dysostosis, an autosomal dominant condition characterized by severe developmental disturbances of the craniofacial region including bilateral coronal synostosis associated with midface hypoplasia, exophthalmia, hypertelorism, and symmetric syndactyly of the hands and feet. The aim of this study is to assess the clinical and computed tomography imaging patterns of non-operated patients with Apert syndrome, correlating the bone abnormalities of the cranium, face and the skull base. The study population consisted of 5 patients with Apert syndrome. As part of the craniofacial assessment of the imaging center's routine, all patients underwent clinical evaluation and CT (computed tomograph) exam. Three-dimensional images were generated from helical CT scans, using an independent workstation, to evaluate the craniofacial abnormalities of the syndrome. Clinical exam determined that syndactyly of the hands and feet, pseudocleft in the midline palate and midface hypoplasia were features observed in all of the Apert patients. 3D-CT showed that some abnormalities such as bilateral coronal synostosis, calvarial midline defect and reduction in the antero-posterior dimension of the anterior, medial and posterior cranial fossae were present in all cases. In conclusion, the correlation of clinical and CT imaging findings can be useful to assess the main features observed in Apert patients, improving the criteria for examining the patient and diagnosing this condition, and contributing to the therapeutic planning and surgical follow-up.

Keywords : Tomography; X-ray computed; Skull; Acrocephalosyndactylia.

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