Characterizing craniomaxillofacial anomalies in achondroplasia : from Jean Delaire to 3D morphometrics analysis

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Achondroplasia is the most common cause of rhizomelic dwarfism, and is linked to FGFR3 activating mutations, leading to alterations in endochondral ossification processes, characterized by growth plate disorganization and premature fusion of skull base synchondrosis. The craniofacial phenotype is characterized by a large skull and prominent forehead, maxillary retrusion, and nasal root saddle, of variable severity. Heterogeneity of craniofacial phenotype and its causes in achondroplasia needs to be elucidated.
The aim of our study was to quantify craniomaxillofacial anomalies in a retrospective series of 15 achondroplasia patients. Cephalometric analyses were performed according Delaire's method, as well as 3D morphometrics studies based on 3D CT-scans. Correlation studies were then performed between several skull base and maxillofacial parameters, to better understand the link between reduction of anteroposterior skull base length, midface retrusion, and the severity of the obstructive sleep apnoea syndrome.