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PHENOTYPIC REFINEMENT OF THE MANDIBULOFACIAL DYSOSTOSIS BAURU TYPE

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PURPOSE: Refine the MFD Bauru type syndrome phenotype; Investigate the mandible, maxilla, zigomatic arch on the sample, using radiological evaluation. **METHODS:** 11 subjects were included on this study with clinical diagnosis of MFD Bauru type. All subjects were submitted to genetics and clinical assessment and, those that haven't been submitted to CT Scanning were submitted to face CT cone beam or teleradiography, depending on age. The CT cone beam images were reconstructed in third dimension and teleradiography obtained in lateral standard. **RESULTS:** all subjects were female gender. Recurrence was observed in two families and suggestive the etiology is genetic with probable genetic heterogeneity. Main clinical finds included: bitemporal narrow, long face, upper eyelid downslanting, high nasal bridge, midface hypoplasia, thick and everted lower lip, micrognathia, minor anomalies of ears and conductive hearing loss. The CT cone beam was realized in 8 subjects and main finds on morphological analyses were: facial asymmetry, asymmetrical orbits, zygomatic arch downslanting, maxilla hypoplasia, hypoplastic mandibular condyle, mandibular asymmetry, micrognathia, retrognathia and atopic external acoustic meatus. Cefalometric analyses showed vertical growing pattern, reduced maxilla dimension, small mandible, short mandibular ramus, total facial height reduced, antero-inferior facial height increased. **CONCLUSIONS:** the pattern typical facial of MFD Bauru type showed distinct from Treacher Collins syndrome and other MFDs. Higher knowledge of radiological abnormalities of the maxilla and mandible in this individuals can't only assist in refining phenotypic this condition, but also in planning the treatment of changes and the differential diagnosis. The etiology is genetic.