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Dental and craniofacial manifestations in rare genetic thin bone disorders in SA

Manogari Chetty
University of the Western Cape, South Africa

Introduction: The prevalence of Osteogenesis imperfecta type III (OI III) as a category of the inherited connective tissue disorders in South Africa is of paramount importance. Although worldwide, autosomal recessive (AR) OI is rare, it had emerged that the frequency of OI III is relatively high in the indigenous Black African population of South Africa. A review of the literature revealed a paucity of information regarding the dental and craniofacial manifestations of the disorder in this ethnic group. For these reasons, the central theme of this project was the identification, documentation and analysis of these features in individuals with OI III in the Black African population of SA. Methodology: Documentation of the dental and craniofacial phenotype and the correlation with the genotype in affected persons is a major objective of this study. A total of 64 Black African affected persons with OI III were assessed. In addition 5 persons of Cape Mixed Ancestry (CMA) and 3 Indian individuals were investigated. By reason of their similarity to OI, three very rare autonomous genetic thin bone disorders, Pyle Disease, Osteolysis (Torg-Winchester Syndrome) and Osteoporosis-pseudoglioma Syndrome were also investigated and documented in this project. The study had a clinical, imaging and genetic component in which dental and craniofacial abnormalities in affected persons were documented. Although radiographic resources were limited, 15 CBCT images, 20 panorex and 20 cephalometric radiographs were obtained. Results: Specific mutations in the FKBP10 gene were detected in 27 Black African persons of the total 72 individuals with OI III. Autosomal recessive OI III in the Black African population of SA has been shown to be caused by mutations in the FKBP10 gene. FKBP10 is one of the newer members of an expanding list of AR OI genes with the gene map locus 17q21.2. This gene encodes an extracellular matrix protein FKBP65. In terms of genotypephenotype correlations in the Black African population group with OI III, 23 persons with the homozygous mutation, FKBP10_HOM_c.[831dupC][831dupC], 3 persons with the compound heterozygous mutation, FKBP10_CHET_c.[831dupC][831delC] and 1 person with the compound heterozygous mutation, FKBP10_CHET_c. [831dupC][1400-4C>G] were identified. Conclusion: In South Africa, a developing country, the allocation of resources in terms of specialized dental facilities is limited. Socioeconomic barriers also exist with patient access to dental care. The previously neglected dental and craniofacial abnormalities documented in this study emphasizes the importance of a raised level of awareness in terms of dental management and the possible challenges that may be encountered.

Osteogenesis imperfecta (OI) is represented by a gaggle of genetic disorders that mainly affect the bones, animal tissue and should increase skeletal fragility, also referred to as brittle bone disease. Among all cases, 85%–90% present a scarcity of type I collagen thanks to a mutation within the COL1A1 and COL1A2 genes that's inherited from the oldsters or develops de novo. With advances in genomic analysis and exome sequencing, several other gene mutations, like CRTAP, LEPRE1 PPIB, SERPINH1, and SP7 mutations, which could end in defects in collagen post-translational modifications or osteoblast differentiation, are found to be involved within the occurrence of OI.