Oral changes in Alpha-Mannosidosis: case report

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Alpha-Mannosidosis (-Manosidosis) is an extremely rare autosomal recessive disease worldwide, with an approximate prevalence of 1 in every 500,000 - 1,000,000 births. It is characterized by alpha-mannosidase deficiency, caused by mutations in the MAN2B1 gene, this lysosomal enzyme is responsible for the degradation of mannose rich oligosaccharides, its absence leads to the accumulation of this substance in all tissues causing functional disturbances of cells. This syndrome can affect individuals of any ethnicity, the main features of the disease are mental retardation, hearing loss and gross facial appearance, including oral characteristics and changes, which are not yet fully understood. The objective of this case report is to demonstrate a case of a patient with this disease, especially the oral manifestations found during the dental examination. A 17-year-old male patient, white -skinned, diagnosed at age 6 by a geneticist from an alpha-mannosidosis urine test, attended the Dental Radiology Center and treated the patient with Piquet Carneiro Polyclinic Special Needs - PPC / UERJ, complaining of erythematous gum. Extraoral physical examination revealed changes such as: gross facial appearance, intellectual disability, skeletal deformation and hearing loss. The intraoral physical examination showed spaced teeth, healthy and gingivitis. Radiographic examination revealed molar taurodontics. This report shows the oral manifestations of the disease, and the need for further reports, as the disease is extremely rare.

Keywords: Alpha-Mannosidosis; Syndrome; Oral manifestations.

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