Dental Abnormalities Related to Roifman Syndrome: A Case Report

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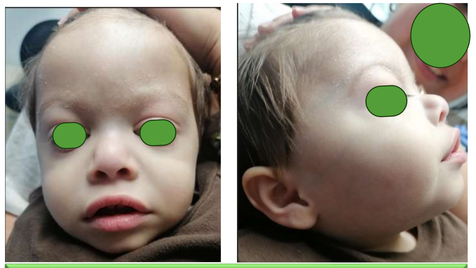
ABSTRACT

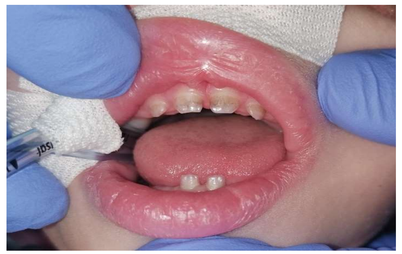
This clinical case report's objectives were to discuss the proper diagnosis, treatment, and follow-up of a 23-month-old male with a known medical history of Roifman syndrome. No similar cases have been reported in the medical literature. This is the first case in which the dental characteristics are documented, and the patient's dental treatment is discussed.

MATERIALS AND METHODS

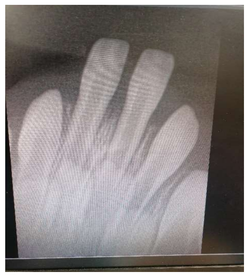
This patient was referred to the Children's of Alabama Hospital Dental Clinic by the Gastrointestinal team with a chief complaint of discolored teeth. He has a significant medical history of Roifman syndrome, Asthma, Ichthyosis Vulgaris, IgG deficiency, Ventricularulomegaly of the brain, and severe iron deficiency. The child received treatment under general anesthesia as part of a collaborative effort between the dental and gastrointestinal teams. The gastrointestinal team successfully placed a G-tube, while the dental resident conducted a thorough oral examination, including radiographs. Then the dental resident removed the external stains via polishing, performed an incision on an eruption cyst, drained the blood, and applied fluoride varnish on erupted teeth.

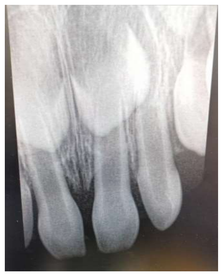


Facial Dysmorphism

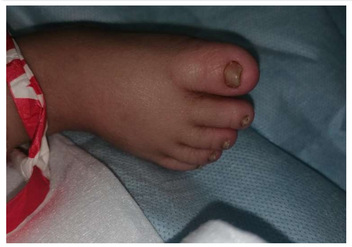


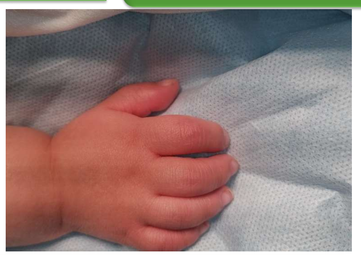
Teeth Discoloration
Wide Spaced Dentition



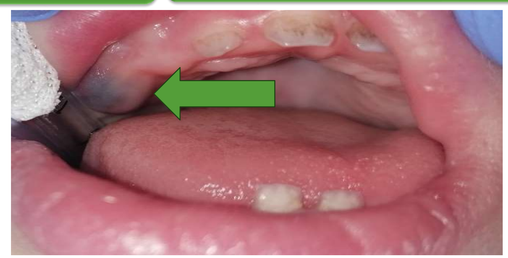


Microdontia





Body Dysmorphic Features



Eruption Cyst

RESULTS

On extraoral examination microcephaly, sparse scalp hair, symmetrical eyes with shallow orbits, hypertelorism, an upturned nasal tip, a wide mouth, thin upper lip, downturned lips, and dry scaly lips were noted. Several interesting dental findings were observed on intraoral examination. The child is in the primary dentition stage, with delayed dental age. He has a high-arched palate, but there was no frank clefting. The teeth were small (Microdontia), and pointed, with wide inter-dental spacing. Severe internal and external teeth discoloration was noted, and the surface texture was grayish and rough. An Eruption cyst in the area of tooth #B was observed . Radiographically the enamel appeared to be thin and poor anatomic contours of the primary teeth were evident. The pulp spaces appear to follow the anatomic contours of the pointed crowns. Follow-up visits showed improvement in the external teeth staining, but no improvement in internal staining was observed.

CONCLSUION

Roifman syndrome is an extremely rare genetic disorder characterized by a combination of antibody deficiency, spondyloepiphyseal dysplasia, facial dysmorphism, growth retardation, and retinal dystrophy. To date, fewer than twenty cases of Roifman syndrome have been documented. This case report aims to highlight the dental manifestations associated with the syndrome, offering valuable insights and establishing a foundation for further research as we continue to expand our understanding of this rare condition.

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