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Case Report Open Access

Surgery Under General Anesthesia to Treat Gingival Hyperplasia Endanger the Life of Children with Mucolipidosis II - A Case Report

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Abstract

Background and case description: A 28 month old Caucasian boy, diagnosed with Muccolipidosis II, presented at the Pediatric Dentistry Department of the Hadassah medical center demonstrating characteristic features including hypertrophic gums with delayed teeth eruption. The child has tracheostomy, gastrostomy and hearing and eyesight deficiencies. Because of severe limitation on airway management that may cause respiratory and cardiovascular complications, surgical intervention was not performed. As the child has no pain from the gingival enlargement and there was no esthetic issue, a follow- up with active oral hygiene instruction was decided on.

Clinical implications: Surgical intervention should be highly avoided in children with Mucolipidosis II (I-cell disease).

Keywords: Gingival hyperplasia; Mucolipidosis II; I-cell disease; Dental treatment; General anesthesia

Introduction

Mucolipidosis II (ML II, also known as I-Cell Disease (I.C.D.)) is a rare, slowly progressive, autosomal recessive metabolic disorder of lysosomal storage [1] that affects approximately 1:125,000 up to 1:600,000 people [2] and usually becomes apparent within the first year of life, but is rarely diagnosed at birth [3]. The underlying pathophysiology is related to a deficiency in the lysosomal hydrolase N-acetylglucosamine-1-phosphotransferase (GNPTAB) enzyme [1], required for biosynthesis of mannose 6-phosphate recognition marker that is essential for efficient targeting of soluble lysosomal enzymes to lysosomes. Defects in this enzyme lead to miss-sorting of several lysosomal enzymes and lysosomal accumulation of non-degraded macromolecules [4], manifested in patients cultured skin fibroblasts as coarse granular cytoplasmic inclusions that are large lysosomes containing heterogeneous material [5].

The most common features of the disorder are mental and physical retardation with typical orofacial features including coarse facial features, puffy eyelids with slight exophthalmia, depressed nasal bridge, incompetent lips, thick tongue and gingival enlargement with buried teeth [3,6]. Radiographic examinations of the jaws usually demonstrate deeply buried teeth within the gingivae that frequently do not erupt. Often the teeth appear hypocalcified and mucolipid storage material that accumulates in the dental follicles may jeopardize eruption and cause gingival enlargement [7,8]. Postnatal growth is limited and often ceases in the second year of life. Delayed development is apparent early in life, as well as hypotonia, congenital dislocation of the hips and hernia. Contractures develop in all large joints, the skin is thickened and orthopedic abnormalities are already present at birth. Development does not seem to proceed further than sitting and standing without support, and a few social responses such as smiling and early vocalization. Neither unaided walking nor self-feeding is accomplished [9]. All children appear to have cardiac involvement. Progressive mucosal thickening narrows the airways and gradual stiffening of the thoracic cage contributes to respiratory insufficiency, the most common cause of death [1]. The life expectancy of children with Mucolipidosis is poor, as death typically occurs within the first decade of life secondary to cardiopulmonary complications [10]. There is no specific treatment available for ML II beside bone marrow transplantation as a source of structurally normal lysosomal enzymes [11].

The present case describes a 28- month old child diagnosed with Muccolipidosis II who presented with gingival hypertrophy and erupting teeth and was subscribed regular observation with supportive and preventive treatment.

Case Report

A 28- month old Caucasian boy, the youngest of three children, presented at the Pediatric Dentistry Department of the Hadassah medical center. The parents had noticed that one of his primary teeth had started to emerge and were concerned by his swollen gums and delayed teeth eruption. The child was born in a cesarean section (C-section) with polyhydramnios weighting 2.960 kilograms and was immediately hospitalized because of desaturation and a syndromic appearance. Imaging revealed multiple fractures of the long bones. He was diagnosed with Muccolipidosis II, manifesting the characteristic features of extreme psychomotor delay and failure to thrive, coarse facial features, gingival hyperplasia, extensive inguinal hernia that was repaired, cardiac problems and joint stiffness. His hearing and eyesight are impaired. Since the age of 9 months he had suffered several episodes of pneumonia and breathing deterioration led to a tracheostomy. His oral functions are inferior and he is fad through gastrostomy.

Examination on admission revealed a considerable developmental delay and facial features typical of Muccolipidosis II, including coarse facial features, exophthalmia, puffy eyelids, and a depressed nasal bridge (Figure 1). Intraoral examination revealed massive generalized gingival hyperplasia. The gingival enlargement was mainly in the vertical dimension with loss of space between the jaws, in addition to the space created by the use of the pacifier (Figure 2). Tooth H (Primary maxillary left canine) was the only tooth visible in the mouth and had minor gingival inflammation (Figure 2).

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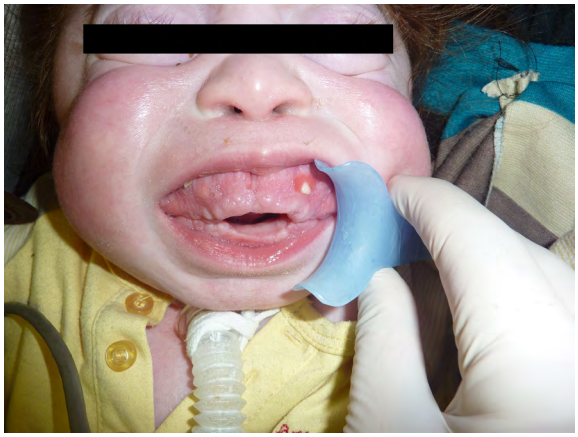

Figure 1: Clinical appearance of typical faces of Mucolipidosis II disease.

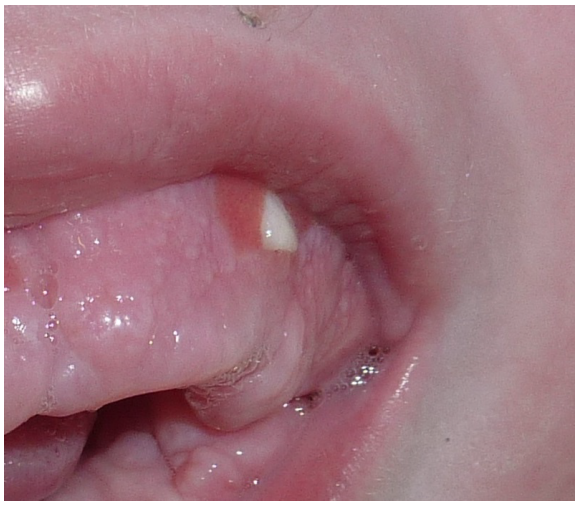

Figure 2: The buccal aspect of teeth H is seen. The inflammation around the tooth that erupted can be seen.

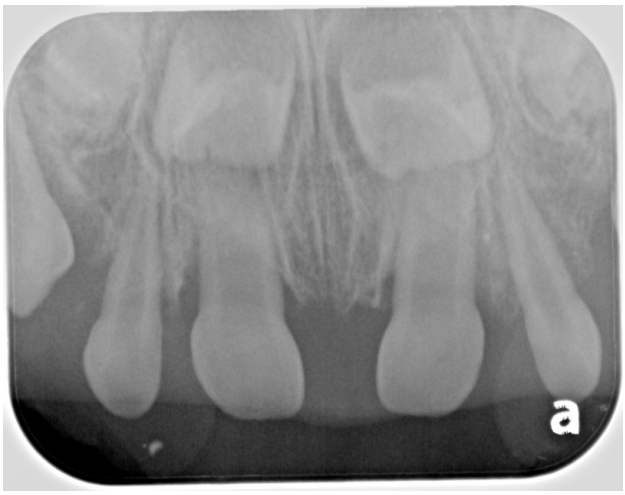

Figure 3: Periapical radiograph of the pre-maxilla. All upper primary incisors are out of the osseous tissue but are still enclosed by gingival tissue. Permanent tooth germs are poorly formed.

An anterior periapical radiograph revealed all upper deciduous incisors normally developed and corresponding to the age of the child, permanent incisors teeth buds appeared in the correct developing stage. There was no evidence of hypocalcification (Figure 3). Surgical intervention at this point was ruled out due to the following considerations:

1. The child is fad through gastrostomy with no nutritional difficulties.
2. The gingival enlargement is not painful.
3. There is no esthetic issue.
4. Reoccurrence of gingival enlargement is un-predictable after surgical removal.
5. Because of concerns about airway management, surgical intervention should be avoided as much as possible in children with Muccliopidosis II [1].

A follow up with oral hygiene instructions was suggested. Parents were given oral hygiene instructions and the use of Chlorhexidine in the area of the erupting teeth was recommended. A follow-up visit was scheduled in three months.

Discussion

We present a 28 month old young boy diagnosed as Muccolipidosis II with gingival hypertrophy and erupting primary teeth. Surgical intervention at this point was not advised due to concerns about airway management. Children with ML II have a small airway due to reduced tracheal elasticity by connective tissue stiffening and progressive narrowing of the airway from mucosal thickening. Poor compliance of the thoracic cage and progressively sclerotic lung parenchyma further complicate airway management. Extubation may also be challenging [1].

This rare case of Mucolipidosis II (ML II, I-cell disease) is of importance regarding the different considerations taken into account when deciding upon a treatment of medically compromised children. In this case surgical intervention to reduce gingival overgrowth would present a life threatening risk. Similarly to Lee and O'Donnell's case report the child will be observed with supportive and preventive treatment to keep him as dentally comfortable as possible [12].

Why this Clinical Report is Important to Paediatric Dentists

- Because of concerns about airway management being limited by respiratory and cardiovascular complications, surgical intervention should be highly avoided in children with Mucolipidosis II (I-cell disease).

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