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Case Report

Dentistry Section

Oral Manifestations and Conservative Clinical Management of a 2-year-old Child with Congenital Ichthyosis: A Case Report

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ABSTRACT

Congenital Ichthyosis (CI) is an uncommon genetic condition affecting tissues of ectodermal origin, including the skin, nails and tooth enamel. The thickening of the stratum corneum impairs the skin's ability to act as a protective barrier due to aberrant epidermal cell differentiation, as well as lipid synthesis, metabolism and transport. This set of keratinisation disorders is clinically and etiologically diverse, with severe forms characterised by skin dryness, flaking and peeling, leading to a wide range of complications. CI involves mutations in various genes and presents with different symptoms that can significantly affect the patient's quality of life. Although it affects tissues of ectodermal origin, little is known about its oral manifestations. Lamellar Ichthyosis (LI) is a type of CI that typically manifests at birth, with the infant found to be covered in a membrane. Xerostomia is one oral symptom that may increase the risk of dental caries. While many individuals with CI may present with normal dentition, some may experience dental defects, delayed tooth eruption and a higher risk of dental caries. In the context of CI, the primary consideration in dental care should include the presence of friable and tender skin, which necessitates extra caution during handling. It is crucial to ensure that any procedure minimises irritation and injury to the skin, as it is more vulnerable than usual. The present case report highlights a case of a two-year-old female child diagnosed with CI who presented with early childhood caries. Management may include a conservative approach for rehabilitation, utilising delicate handling of soft tissues to reduce patient discomfort and facilitate successful treatment.

Keywords: Autosomal recessive, Collodion baby, Lamellar ichthyosis, Non pharmacological management

CASE REPORT

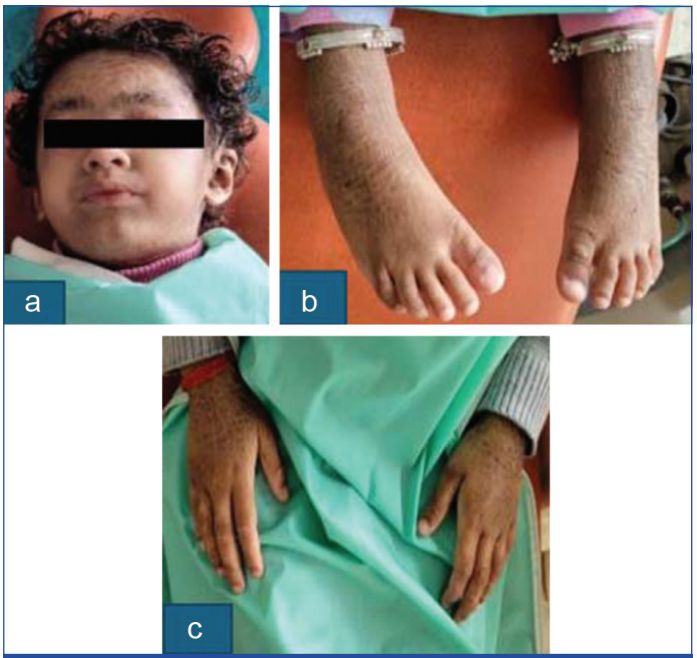
A two-year-old female child presented with her parents to the Department of Paediatric and Preventive Dentistry, with the chief complaint of blackish discolouration in the lower right back tooth region and the upper front tooth region for the past few months. The child did not report any associated pain, discomfort, or concerns regarding aesthetics.

Upon eliciting the personal history, it was revealed that the child was born prematurely (34 weeks and 2 days) via caesarean section to parents with a consanguineous marriage and was a "collodion baby" at birth, shedding the membrane at 10 days of age. Although there was no blistering at birth, the mother later noticed the development of scales causing roughness of the skin. After consulting a paediatrician, the child was diagnosed with LI. However, the parents reported a negative family history for the condition.

On general physical examination, dark brown scaly striations were observed on the forehead, bridge of the nose, chin, in front of both right and left ear tragus and on the hands and legs [Table/Fig-1a-c]. Intraoral examination of the soft tissue revealed pale pink gingiva, which was firm in consistency and showed no bleeding on probing. Hard tissue examination revealed the presence of primary dentition, with caries and pulp exposure in teeth 51, 52, 61 and 62, as well as caries involving dentin in tooth 54. Cervical caries were also noted in the mandibular primary anterior teeth [Table/Fig-2a-d].

Considering the patient's cooperation, clinical findings and the parents' acceptance, the treatment was planned to be carried out in a non pharmacological setting and to be as minimally invasive as possible. Pulp therapy, along with full mouth caries stabilisation, was planned and explained to the parents. The paediatrician's consent was obtained and full mouth rehabilitation was carried out over multiple visits.

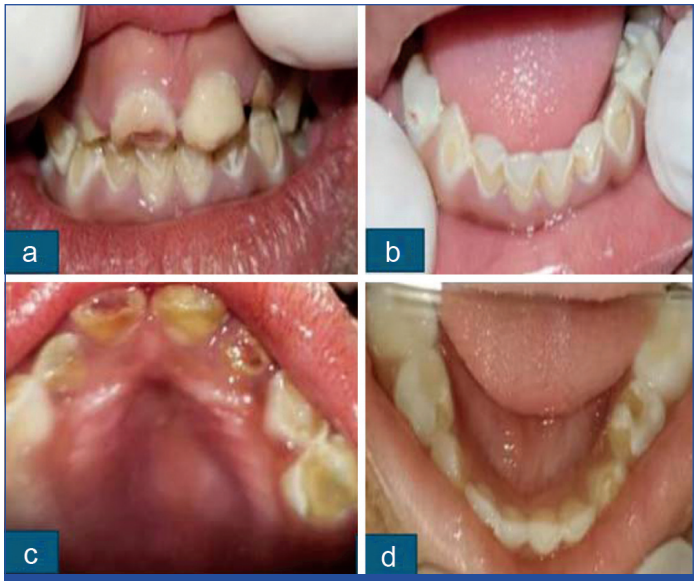
During the first visit, a restoration with glass ionomer cement was performed on tooth 54. Given the patient's cooperativeness, a multiple-visit pulpectomy was chosen as the treatment option.



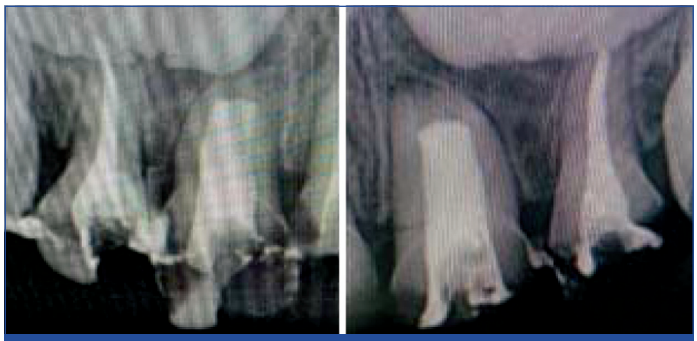
[Table/Fig-1]: a) Extraoral photograph showing dark brown scaly striations are seen on the forehead, bridge of the nose, chin, in front of right and left ear tragus; b) Extraoral photograph showing dark brown scaly striations are seen on hands; c) Extraoral photograph showing dark brown scaly striations are seen on legs.

Under local anaesthesia, an access opening was made in teeth 51, 52, 61 and 62, followed by pulp extirpation and placement of an intracanal medicament (triple antibiotic paste) to avoid flare-ups and eliminate any remaining bacterial infection. A temporary restoration was then provided [1].

In the second visit, the canals were meticulously irrigated with 3% Sodium Hypochlorite (NaOCl) and 5% saline and obturation was performed using a calcium hydroxide preparation (Metapex) for teeth 51, 52, 61 and 62 [Table/Fig-3]. To avoid skin irritation and enhance patient comfort during the procedure, Vaseline was applied and



[Table/Fig-2]: a) Intraoral photograph showing deep carious lesion in maxillary primary incisors; b) Intraoral photograph showing cervical caries in mandibular primary incisors; c) Preoperative occlusal photograph of maxillary arch; d) Preoperative occlusal photograph of mandibular arch.



[Table/Fig-3]: Postoperative radiograph showing obturation irt. 51, 52, 61, 62.

regular breaks were provided throughout the treatment. Restoration was also carried out on the mandibular teeth with cervical caries using glass ionomer cement.

Post-restoration, oral hygiene instructions were given and patient compliance was confirmed. The patient has been advised to return for regular follow-up in three months.

DISCUSSION

Congenital ichthyosis (CI) is an uncommon genetic skin condition that primarily manifests as autosomal recessive. It is known to result from aberrant epidermal cell differentiation, as well as lipid synthesis, metabolism and transport, which impacts the skin's barrier function and leads to noticeable thickening of the stratum corneum [2,3]. X-linked ichthyosis and ichthyosis vulgaris are considered to be more prevalent, affecting one in 6,000 male births and one in 250 female births, respectively [4]. However, other forms of Autosomal Recessive Congenital Ichthyoses (ARCI), such as Lamellar Ichthyosis (LI), congenital ichthyosiform erythroderma, harlequin ichthyosis, Self-Healing Collodion Baby (SCCB), acral SCCB and swimsuit ichthyosis, are rare and affect one in 200,000 births, with equal incidence in men and women [4,5].

A newborn with LI is usually covered in a collodion membrane that breaks down within the first 10-14 days of life, eventually developing large, black, plate-like scales. Complications may include ectropion, eclabium, glued and primitive ears, sparse, dry, ringlet and brittle hair, variable degrees of hypohidrosis and palmoplantar keratoderma, which may lead to macrodactyly [6,7]. The incidence of LI is as low as 1 in 200,000-300,000 and typically affects children born to consanguineous marriages [5,8].

Recent research has highlighted several aetiological genes linked to CI, with Keratin 1 (K1), K2, K10, Transglutaminase 1 (TGM1), Adenosine Triphosphate (ATP) Binding Cassette Subfamily A Member 12 (ABCA12), Arachidonate 12-Lipoxygenase, 12R Type (ALOX12B) and NIPA Like Domain Containing 4 (NIPAL4) identified as key contributors [2]. Among these, the gene transglutaminase 1 (TGM1) is considered a primary cause of LI [9]. TGM1 plays a crucial role in skin barrier function by catalysing the formation of cross-links between precursor proteins and ceramides, which are essential for the integrity of the Cornified Cell Envelope (CEE) [5]. TGM1 enzymatic action involves the crosslinking of N (glutamyl) lysine and co-hydroxy ceramide, helping to form the structural framework of the skin's outermost layer. This process is integral in maintaining the skin's water retention and protection [5]. Beyond the skin, TGM1 has other roles, such as forming covalent bonds with salivary components, particularly in the parotid salivary gland, suggesting that TGM1 influences mucosal pellicle formation, which is important for maintaining oral health [8,9].

Enamel hypoplasia, which is the underdevelopment of enamel, is a common issue among hereditary skin disorders, as both skin and teeth share the same ectodermal origins and are affected by overlapping gene expression [2]. Although many individuals with CI may present with normal dentition, some may experience dental defects, delayed tooth eruption and a higher risk of dental caries. Other oral and dental conditions may include gingivitis, periodontitis, enamel hypoplasia, delayed primary and secondary eruptions, bruxism, alveolar ridging, bifid teeth, irregular tooth morphology, mouth breathing and xerostomia, with rare occurrences resulting in squamous cell carcinoma and hyperkeratotic plaques affecting the oral mucosa [10]. One significant oral health complication due to the dysfunction of TGM1 in LI is xerostomia (dry mouth), caused by reduced salivary secretion, which results in an increased incidence of dental caries and exacerbates oral discomfort [10]. Understanding the genetic cause of CI, particularly TGM1, is crucial for addressing oral health challenges and improving management strategies for affected individuals.

The literature regarding dental abnormalities in ichthyoses is sparse and primarily consists of isolated case reports. One case involved a two-year-nine-month-old Japanese boy with CI who developed severe dental caries and required full mouth rehabilitation under general anaesthesia, as reported [2]. Another case described a two-year-old diagnosed with Autosomal Recessive Congenital Ichthyosis (ARCI), who presented with reduced salivary flow and cervical caries; this child was treated with preventive and restorative measures and the parents were guided on proper oral hygiene maintenance [9]. Additionally, a case was reported of a one-year-old female with Keratitis-Ichthyosis-Deafness (KID) syndrome who had natal teeth [11].

In the context of CI, the primary consideration in dental care should be the friable and tender skin that requires extra caution during handling. It is important to ensure that any procedure minimises irritation and injury to the skin, which is more vulnerable than usual. Key recommendations for managing such patients include salivary flow evaluation, frequent hydration, fluoride use and practicing preventive dentistry as early as possible to address the rise of rampant caries in childhood. For patients with LI who have difficulty cooperating or require extensive dental treatment, general anaesthesia might be necessary for a successful treatment approach.

CONCLUSION(S)

In conclusion, the present case report emphasises the successful management of a child needing pharmacological intervention, reflecting the positive outcome when patient cooperation and behavior are conducive to non invasive methods. While the primary concern in patients with CI, especially LI, is skin-related, oral health issues should not be overlooked. Close attention to salivary function, preventive dental care and customised treatment planning are crucial for maintaining the oral health of these patients. However, it also points out that further research, especially genetic studies, would

be beneficial to fully understand the oral management of LI and to fine-tune preventive and treatment strategies for these patients.

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