IOSR Journal of Dental and Medical Sciences (IOSR-JDMS)
e-ISSN: 2279-0853, p-ISSN: 2279-0861.Volume 24, Issue 6 Ser. 1 (June. 2025), PP 19-24
www.iosrjournals.org

Oral Rehabilitation Of A 4-Year-Old Child With Christ-Siemens-Touraine Syndrome

A. El-Missioui, H. Ramdi, H. El Khammal
Resident In Pediatric Dentistry - Faculty Of Dental Medecine Rabat - Mohammed V University In Rabat–Morocco; Mohammed V Military Teaching Hospital, Rabat Morocco
Professor Of Higher Education In Pediatric Dentistry- Faculty Of Dental Medicine Rabat - Mohammed V University In Rabat–Morocco
Associate Professor, In Pediatric Dentistry- Faculty Of Dental Medicine Rabat - Mohammed V University In Rabat -Morocco

Abstract:
Christ-Siemens-Touraine syndrome (CST syndrome), also known as Anhidrotic Ectodermal Dysplasia (AED) or Hypohidrotic Ectodermal Dysplasia (HED), is the most common form of ectodermal dysplasia (ED). It is a congenital and hereditary condition caused by mutations in the genes of the ectodysplasin/NF-kappa B signaling pathway, which are necessary for the proper development of several ectodermal structures including teeth, nails, hair, exocrine glands, adenohypophysis, skin and its appendages. All modes of transmission have been described and it can affect both sexes. . Its incidence is estimated at approximately 1 in 100,000. It is characterized by a triad of signs: hypohidrosis or anhidrosis, hypotrichosis, and hypodontia, oligodontia or anodontia.
This syndrome leads to oral and dental manifestations, including anomalies in the number and shape of teeth in both dentitions. These alterations cause functional difficulties and have a significant aesthetic impact, often requiring early prosthetic rehabilitation in children due to growth-related constraints.
We report the case of a 4-year-old child who was treated in our pediatric dentistry department at the university hospital center for prosthetic rehabilitation. The treatment restored functional occlusion while preserving facial growth and the child's psychosocial well-being. The prosthetic rehabilitation, carried out in several phases, aimed to improve mastication, phonation and aesthetics, with a planned follow-up until adulthood for a definitive prosthetic adaptation.
This case illustrates the importance of a multidisciplinary approach in the treatment of ectodermal dysplasia, as well as the central role of the pediatric dentist in early prosthetic rehabilitation, aiming to improve the quality of life of young patients.
Key words: *Anodontia, Oligodontia, Anhidrotic Ectodermal Dysplasia (AED), Dental management, Pediatric dentistry.*

Date of Submission: 23-05-2025 Date of Acceptance: 03-06-2025

I. Introduction:

Anhidrotic ectodermal dysplasia (AED), also known as Christ-Siemens-Touraine syndrome, is the most common form of ectodermal dysplasias (ED) which encompasses a heterogeneous group of rare genetic diseases or syndromes characterized by abnormalities in the development or homeostasis of at least two of the following structures, all derived from the embryonic ectodermal layer: teeth, nails, hair follicles, and certain glands.

AED is characterized by a triad of signs: hypohidrosis or anhidrosis (due to hypoplasia of sweat glands), hypotrichosis (sparse scalp and body hair) and hypodontia, oligodontia or anodontia.[1, 2]

The oral and dental manifestations of CST syndrome are primarily characterized by abnormalities in the number and shape of teeth, affecting both the primary and permanent dentition. These anomalies lead to significant functional impairments, such as chewing and speech difficulties, often requiring a semi-solid diet beyond the age of three or four years. The severe aesthetic impact generally justifies early management, raising the issue of prosthetic rehabilitation in young children, while considering growth-related constraints.

This article presents an initial prosthetic treatment plan for these rare but complex cases, illustrated by a clinical case of a four-year-old patient, referred by her geneticist at the request of her parents. A multidisciplinary approach ensures optimal management of the specific needs of these young patients.

II. Case Presentation:

M.T. a 4-year-old Moroccan girl, was born from a third-degree consanguineous marriage, and resides in a region 550 km away from Rabat. She was referred for the first time in February 2022 to the Department of Pediatric Dentistry at the Consultation and Dental Treatment Center of the Ibn Sina University Hospital in Rabat - Mohammed V University of Rabat.

At the age of 1 year and 9 months, she was initially referred by a geneticist at the Children's Hospital of Rabat, who strongly suspected anhidrotic ectodermal dysplasia. During this first consultation, no therapeutic intervention was possible due to patient's young age and lack of cooperation.

In June 2024, at the age of 4 years, she was seen again to start prosthetic rehabilitation.

During the medical interview, the mother reported that the pregnancy had not been monitored by a doctor, no medication had been taken during pregnancy, and it had been carried to term without complications. No family history of **AED** was reported. Her younger 2-year-old sister was in good general health and did not present any signs of the condition.

The parents reported that the girl suffered from heat intolerance with the absence of sweating, even in summer. She also experienced mastication and speech difficulties, as well as frequent gastroesophageal reflux disease**.** They also mentioned that she was under dermatological follow-up.

The parents expressed psychological distress and social concerns, as people around them mocked their daughter, claiming she was possessed by spirits due to her edentulous condition. Her peers also teased her about her appearance. This situation deeply affected the child, leading her to refuse school attendance, as she perceived her edentulism as a major aesthetic and psychological issue.

The general and extraoral clinical examination revealed the classic characteristics of ectodermal dysplasia. The patient had thin and sparse hair, a prominent forehead, reduced lower facial height, protruding lips, a saddle-shaped nose with crusty secretions in the nasal cavities, diffuse skin dryness (xerosis), and eczema on the hands. (Figures 1 & 2)

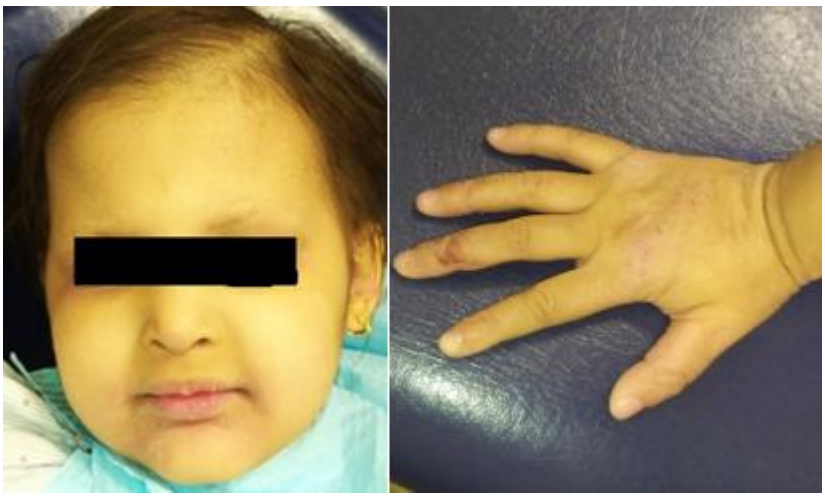

Figure 1: Front view showing classical features of child with AED
Figure 2: View of the right hand showing eczema and skin dryness

The intraoral examination revealed oligodontia, with the presence of conically shaped primary central incisors and the right and left primary first molars in the maxillary arch, along with anodontia in the mandibular arch. The examination also showed alveolar bone hypoplasia in the edentulous areas. (Figures 3 & 4)

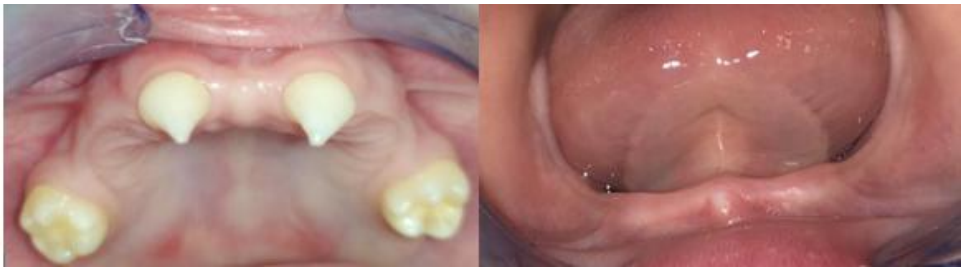

Figure 3: Intraoral view showing oligodontia of maxillary arc
Figure 4: Intraoral view showing anodontia in the mandibular arch with alveolar bone aplasia in the edentulous areas

The panoramic radiograph revealed the agenesis of all teeth, except for the buds of the permanent central incisors, the primary maxillary central incisors, and the primary maxillary first molars, each presenting with a single root and a single wide endodontic canal. Additionally, a nasal septum deviation was observed. (Figure 5)

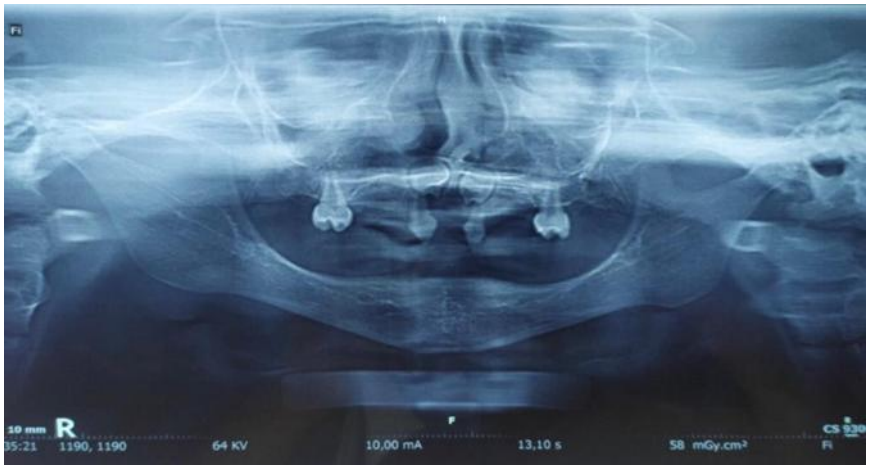

Figure 5: Orthopantomogram (OPG) confirming the clinical diagnosis

Regarding treatment, the young age of the patient guided us towards a partial maxillary and total mandibular prosthetic rehabilitation, designed with an evolutionary approach.

Before proceeding with the definitive prosthetic phase, the following preliminary treatments were performed:

- Prophylactic cleaning of the dental surfaces, along with the application of sealant on the pits and fissures of the present molars (Figure 6).
- Coronoplasty of the maxillary central incisors using composite to give them a broader shape rather than a conical one, aiming to enhance both aesthetics and function. (Figure 9)

To achieve this:

1. A primary alginate impression was taken to obtain a study model.
2. On this model, a wax-up was created to reconstruct the coronal shape of the central incisors (Figure 7).
3. An elastomeric key was fabricated to reproduce the final tooth appearance in the mouth through composite coronoplasty. (Figure 8)

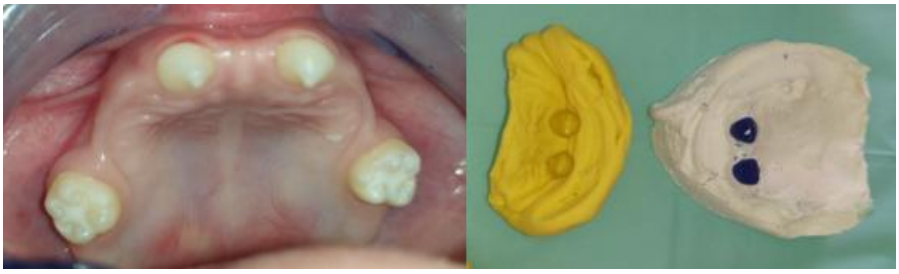

Figure 6: Intraoral view showing sealant application on molars pits and fissures
Figure 7: Crown Wax-Up on plaster model and Elastomeric key fabrication

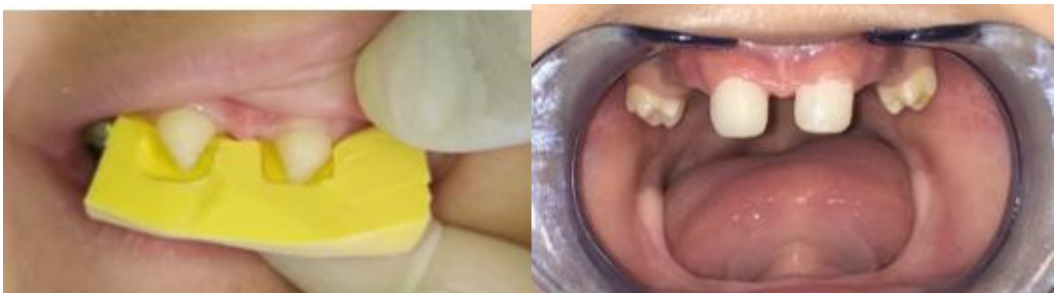

Figure 8: Adaptation of the silicone Key in the mouth
Figure 9: Upper central incisors coronoplasty with composite

A first series of alginate impressions of both arches was taken to facilitate the study of models. Based on these models, individual impression trays were fabricated.

After trial fitting, adjustments, and relining of the edentulous areas with Kerr paste, a secondary impression was taken using polyether material, which ensures precise and detailed capture of soft tissues and remaining dental structures. (Figure 10)

This material has the advantage of offering high dimensional stability, ensuring a reliable foundation for the subsequent prosthetic design. The secondary impression enabled the creation of an accurate working model, on which occlusal wax rims were fabricated (Figure 11).

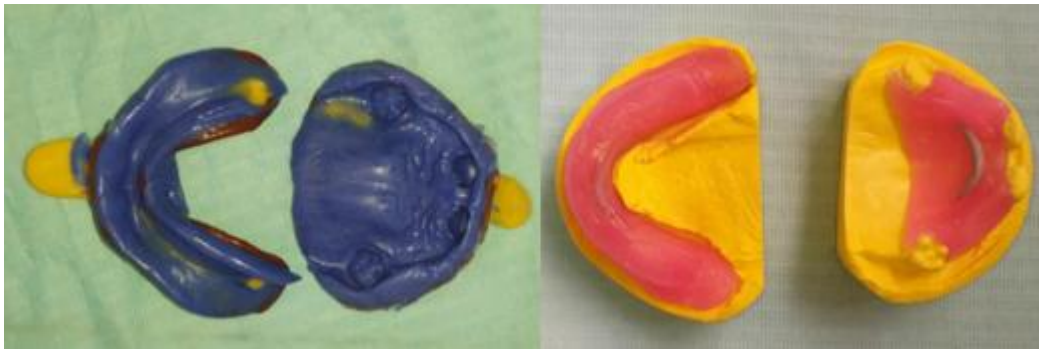

Figure 10: Secondary impressions showing precise structural details
Figure 11: Fabrication of wax occlusion rims

The occlusion recording and articulator mounting of the wax occlusion rims on the secondary models were performed following standard removable prosthetic protocols. This step was essential for determining the vertical dimension and guiding the fabrication of the final prosthesis (Figure 12).

The retained vertical dimension was determined based on the child's physiological rest vertical dimension, measured during the clinical examination. This approach was chosen to ensure proper occlusal function while accommodating the patient's future growth.

Particular attention was given to lip position and facial aesthetics, considering labial prominence and reduced lower facial height. The goal was to restore a functional occlusion while maintaining a harmonious facial appearance.

Due to time constraints—as the patient resides 550 km away from the hospital—the wax try-in phase was omitted from the clinical steps of the prosthetic fabrication.

The maxillary partial denture and mandibular complete denture were inserted and adapted intraorally. Adjustments were made to ensure optimal retention, stability, and occlusal balance. Any pressure areas or occlusal interferences were carefully eliminated to enhance comfort and functionality for the patient. (Figures 13 & 14)

Ongoing monitoring is recommended to modify or replace the prostheses as needed to accommodate the development of the patient's maxilla and mandible.

The dento-prosthetic rehabilitation not only restored the young girl's smile but also provided valuable psychological support to both her and her parents. (Figure 15)

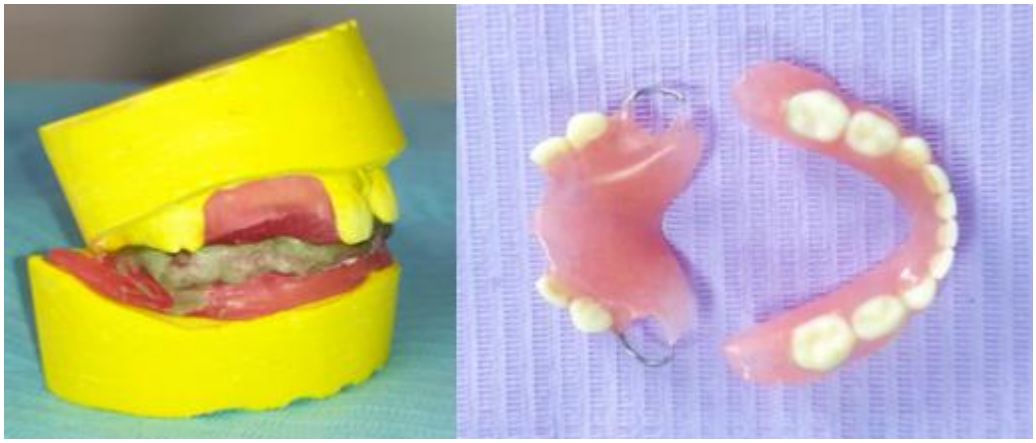

Figure 12: Occlusion Recording
Figure 13: Removable maxillary partial and mandibular complete dentures

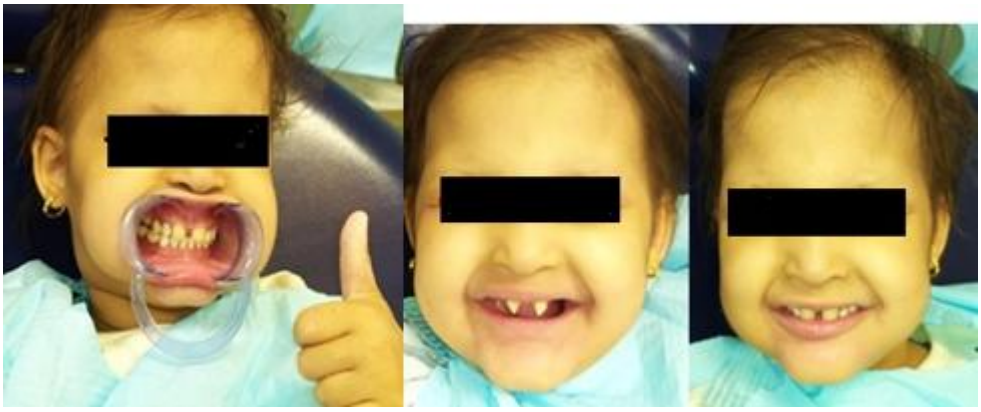

Figure 14: Insertion and adaptation of the maxillary partial and mandibular complete dentures
Figure 15: Improvements in facial appearance and esthetics after placement of prosthesis

III. Discussion:

Hypohidrotic ectodermal dysplasia, also referred to as anhidrotic ectodermal dysplasia or Christ-Siemens-Touraine syndrome, is a rare genetic disorder affecting ectodermal development, characterized by both systemic and oral manifestations.

Epidemiologically, Anhidrotic Ectodermal Dysplasia (AED) affects both sexes worldwide, with a generally male predominance and no specific geographical or ethnic predisposition. AED is a rare disease, with a global incidence estimated at 1 in 100,000.[3]

Genetically, all modes of transmission have been reported (autosomal dominant, autosomal recessive, and X-linked). HED results from mutations in genes involved in either the ectodysplasin/NF-kappaB pathway or the Wnt/beta-catenin pathway, both of which play a crucial role in the development of ectodermal appendages. Three genes have been identified as responsible for the disease: **EDA1** (chromosome Xq13), which accounts for X-linked forms, as well as **EDAR** (chromosome 2q13) and **EDARADD** (chromosome 1q42), which are associated with both autosomal dominant and recessive forms. Additionally, the **WNT10A** gene has been recognized as a key factor in various autosomal recessive forms of ectodermal dysplasias. These 4 genes are responsible for 90% of cases and the **EDA1** gene accounts for more than half the cases and explains the male predominance.[3, 4, 5, 6].

On the systemic level: Hypohidrosis or anhidrosis (reduced or absent sweat glands), leading to increased sensitivity to high temperatures. Hypotrichosis, characterized by fine, sparse, fragile, and pale hair, which may progress to alopecia after puberty. A characteristic facial appearance (marked facial dysmorphia with prominent frontal bossing, a flattened nasal bridge, prominent lips, protruding supraorbital ridges, and hollow cheeks), as well as smooth, dry, slightly wrinkled skin with pronounced pigmentation around the eyes. Other signs may include reduced gland function, eczema, asthma, dryness of the oro-nasal mucosa, onychodystrophy (thin, brittle, and deformed nails), ophthalmological involvement, and in some cases, intellectual delay.[1, 2, 6, 7]

On the oral and dental level: The main dental phenotypic characteristics include hypodontia, oligodontia, or even anodontia. The existing teeth may be conical, exhibit taurodontism, or have supernumerary cusps, affecting both the primary and permanent dentitions. Severe oligodontia can lead to chewing difficulties, promoting an unbalanced diet and, in some cases, underweight. This phenomenon is particularly pronounced in children whose diagnosis and management are delayed. Hypoplasia of the salivary glands may be also present, along with the absence of oral accessory glands, contributes to the development of xerostomia.[1, 2, 6, 8]

The management of anhidrotic ectodermal dysplasia is multidisciplinary, including symptomatic medical treatment at a general level, ranging from the application of hygiene and dietary guidelines to the management of frequently associated pathologies. From a dental perspective, prosthetic rehabilitation plays a predominant role. This management is carried out in several stages, introducing different types of prostheses to compensate for the consequences of edentulism. The objectives include the restoration of mastication, phonation, swallowing, and aesthetics, as well as the reestablishment of occlusal balance and the vertical dimension of occlusion.

The prosthetic rehabilitation of patients with hypohidrotic ectodermal dysplasia (HED) represents a clinical challenge, primarily due to the rarity of the disease and the limited clinical experience.

The placement of removable dentures is recommended from the age of 2 years, although the median observed age is around 4 years. In edentulous children, complete dentures are commonly used. However, implant-supported removable dentures can be considered in the edentulous mandible, with one to four interforaminal implants. Although dental implant placement is generally contraindicated in growing children, it

may be beneficial in certain cases of severe anodontia. The use of mini-implants is a viable temporary solution, offering several advantages, including lower cost, a simplified surgical protocol, immediate restoration, and no need for submerged healing.

The choice of treatment depends on the remaining dentition. In cases of oligodontia, removable partial dentures with clasps are the standard solution, while overdentures with attachments can be an effective alternative, as they provide better retention and stability. When conical teeth are present, direct composite reshaping is frequently performed. If the child has at least six natural teeth, fixed tooth-supported restorations such as crowns or bridges are preferred.

From a craniofacial perspective, the lack of posterior tooth replacement as well as certain fixed implant rehabilitations may have a negative impact on craniofacial development and masticatory function, particularly in cases of skeletal Class III malocclusion.

Overall, a personalized and multidisciplinary approach is essential to optimize occlusal function, aesthetics, and craniofacial development, while considering the constraints related to bone growth. [1, 9, 10, 11]

Our young patient received a removable partial resin denture for the maxilla and a complete denture for the mandible, restoring both occlusal functions and aesthetics. This rehabilitation enabled her to socially reintegrate into her educational and social environment. However, she will be closely monitored to address any concerns or discomfort and to adjust her prostheses as she grows.

IV. Conclusion:

Anhidrotic ectodermal dysplasia is a rare disorder characterized by a polydysmorphic syndrome with both aesthetic and functional implications. Its management requires a multidisciplinary approach, in which the pediatric dentist plays a key role, particularly through prosthetic rehabilitation. This intervention aims to restore essential functions while enhancing the patient's quality of life and social integration.

References:

[1] Schnabl D, Grunert I, Schmuth M,Kapferer-Seebacher I. Prosthetic Rehabilitation Of Patients With Hypohidrotic Ectodermal Dysplasia: A Systematic Review. J Oral Rehabil. 2018;45:555–570. Https://Doi.Org/10.1111/Joor.12638

[2] Anbouba GM, Carmany EP, Natoli JL. The Characterization Of Hypodontia, Hypohidrosis, And Hypotrichosis Associated With X-Linked Hypohidrotic Ectodermal Dysplasia: A Systematic Review. Am J Med Genet Part A. 2020;1–11. Https://Doi.Org/10.1002/Ajmg.A.61493.

[3] Bashyam MD, Chaudhary AK, Reddy EC, Reddy V, Acharya V, Nagarajaram HA, Devi AR, Bashyam L, Dalal AB, Gupta N, Kabra M, Agarwal M, Phadke SR, Tainwala R, Kumar R, Hariharan SV. A Founder Ectodysplasin A Receptor (EDAR) Mutation Results In A High Frequency Of The Autosomal Recessive Form Of Hypohidrotic Ectodermal Dysplasia In India. Br J Dermatol. 2012 Apr;166(4):819-29. Doi: 10.1111/J.1365-2133.2011.10707.X. Epub 2012 Mar 5. PMID: 22032522.

[4] Cluzeau C, Hadj-Rabia S, Jambou M, Mansour S, Guigue P, Masmoudi S, Bal E, Chassaing N, Vincent MC, Viot G, Clauss F, Manière MC, Toupenay S, Le Merrer M, Lyonnet S, Cormier-Daire V, Amiel J, Faivre L, De Prost Y, Munnich A, Bonnefont JP, Bodemer C, Smahi A. Only Four Genes (EDA1, EDAR, EDARADD, And WNT10A) Account For 90% Of Hypohidrotic/Anhidrotic Ectodermal Dysplasia Cases. Hum Mutat. 2011 Jan;32(1):70-2. Doi: 10.1002/Humu.21384. PMID: 20979233.

[5] Https://Www.Orpha.Net/ Orphanet: Dysplasie Ectodermique Hypohidrotique

[6] Wright JT, Grange DK, Fete M. Hypohidrotic Ectoder Mal Dysplasia. 2003 Apr 28 [Updated 2017 Jun 1]. In: Adam MP, Ardinger HH, Pagon RA, Et Al. Eds. Genereviews® [Internet]. Seattle: University Of Washington; 1993–2019. Https://Www.Ncbi.Nlm.Nih.Gov/Books/NBK1112/.

[7] Https://Www.Has-Sante.Fr/ Protocole National De Diagnostic Et De Soins (PNDS) Dysplasie Ectodermique Anhidrotique

[8] Mokhtari S, Lotfi A. Christ-Siemens-Touraine Syndrome: A Case Report And Review Of The Literature. Case Reports In Dentistry. 2012;2012:1-3. Disponible Sur: Http://Www.Hindawi.Com/Journals/Crid/2012/586418/

[9] Shigli A, Sarkar PA. Prosthodontic Management Of Patients With Christ-Siemens-Touraine Syndrome. BMJ Case Rep. 2012 Jun 28;2012:Bcr1120115226. Doi: 10.1136/Bcr.11.2011.5226. PMID: 22744257; PMCID: PMC3448356.

[10] Alnuaimi R, Mansoor M. Prosthetic Rehabilitation With Fixed Prosthesis Of A 5-Year-Old Child With Hypohidrotic Ectodermal Dysplasia And Oligodontia: A Case Report. J Med Case Rep. 2019 Nov 8;13(1):329. Doi: 10.1186/S13256-019-2268-4. PMID: 31699141; PMCID: PMC6839255.

[11] Bala S, Nikhil M, Chugh A, Narwal A. Prosthetic Rehabilitation Of A Child Suffering From Hypohidrotic Ectodermal Dysplasia With Complete Anodontia. Int J Clin Pediatr Dent. 2012 May;5(2):148-50. Doi: 10.5005/Jp-Journals-10005-1155. Epub 2012 Aug 8. PMID: 25206157; PMCID: PMC4148747.