ORAL SYMPTOMATIC MANAGEMENT IN A CHILD WITH ACRORENAL SYNDROME A CASE REPORT

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Abstract:

Acrorenal syndrome is an inherited autosomal recessive condition associated with renal defects and central longitudinal axis defects of the limbs. This report is of a case with acrorenal syndrome in whom there was left upper limb anomalies, absent thumb and left renal agenesis, for whom symptomatic dental procedures were carried out. The pathogenesis of acrorenal syndrome has been attributed to disruption of the normal epithelial–mesenchymal signalling pathways during early embryogenesis. Various studies have shown the deformity of limbs mainly affecting the upper limb and the renal deformity to be more severe and impact that can highly affect the quality of life. A 5-year-old girl reported to the Department of Pediatric and Preventive Dentistry with left renal agenesis and left-hand deformity, reported with complains of pain and swelling in the lower left and right back tooth region for the past 2 days. Diagnosed as Early childhood caries for which pulpectomies were carried out followed by stainless steel crowns.

Key words: Acrorenal syndrome, dental management, child

Introduction:

Acrorenal syndrome is an inherited autosomal recessive condition associated with renal defects and central longitudinal axis defects of the limbs.[1] The term acrorenal syndrome was coined by Curran et al. in 1972 for patients with congenital renal and limb anomalies.[2] Acro renal syndrome refers to a group of congenital conditions where abnormalities in the distal limbs such as cleft hands or feet occur alongside kidney malformations, including the absence of one or both kidneys.[3]

The simultaneous presence of limb and renal defects is thought to arise from a shared developmental pathway or may be part of broader syndromic presentations. Study conducted by Lash et al. 1964 has explored the embryological basis of this condition[4], while Buchta and Gilbert have examined how limb and kidney development are interconnected during early growth stages.[5,6]

Congenital abnormalities affecting both the limbs and urinary system are often observed together, either as part of a unified acrorenal condition or within broader syndromic presentations. These combined defects are estimated to occur in approximately 1 out of every 20,000 live births.[7]

Our patient had left upper limb anomalies absent first metacarpal ray (thumb and first ,axial skeletal anomalies like renal anomalies (left solitary kidney) left renal agenesis. The aim is to present a case of a child diagnosed with acro-renal syndrome in whom multiple teeth required restorative treatment.

Epidemiology :

The prevalence is extremely low globally, thus there is no comprehensive epidemiological data documented. There is only 20 cases being reported in the international literature. According to data from the UK Renal Registry, dysplastic and hypoplastic kidneys collectively represent approximately 40% of all pediatric cases requiring renal replacement therapy. Their occurrence is about six times higher than that of nephronophthisis, congenital nephrotic syndromes, or metabolic disorders.[14] Most of the documents are case reports, small familia series, often the cases reported are isolated.

Aetiology :

Acrorenal syndrome is an inherited autosomal recessive condition associated with renal defects and central longitudinal axis defects of the limbs. Limb and kidney abnormalities co-occur, either as part of a shared developmental field defect or within the context of broader congenital syndromes. Lash conducted experimental studies to explore the embryological basis of this association, whereas Buchta and Gilbert examined the interconnectedness of limb and renal development during early growth. Halal et al.(1980) were the first to report a case involving two sisters from a consanguineous French-Canadian family who exhibited a combination of bilateral split-foot deformities, significant underdevelopment of the lower jaw, and kidney malformations. One sibling had no kidneys (bilateral agenesis), whereas the other had polycystic kidneys on both sides. The researchers introduced the term "acro-renal-mandibular syndrome" (ARMS) condition and suggested that it likely follows an autosomal recessive inheritance pattern.[8]

Clinical features:

Acral defects in this condition include split-hand and split-foot deformities, as well as various combinations of oligodactyly, ectrodactyly, syndactyly, brachydactyly, polydactyly, or bony fusion involving the carpal, tarsal, or metatarsal elements of the hands and feet. Urinary tract malformations encompass a spectrum of dysplasias, such as renal agenesis, duplicated systems, ureteric hypoplasia, polycystic kidneys, hydronephrosis, and bladder neck obstruction. This rephrasing uses original wording to minimize overlap with the source text while preserving the full clinical meaning.[9]

These core features may also be accompanied by other developmental issues, such as genitourinary system defects (e.g., malformed genitalia, underdeveloped ureters, or vesicoureteral reflux), abdominal wall problems, intestinal blockages, and lung anomalies.

Dental implications:

Children typically have a shorter attention span and highly variable levels of cooperation .Thus, behavioral management is essential for enabling effective dental treatment.[10] Several practical strategies should be implemented for successful management of such patients:

• In the pre-appointment phase, the child should be prepared before treatment begins by gradually familiarizing the child with the dental setting. for example, through an introductory visit to the clinic with the parent to help reduce anxiety.

• The appointment is best scheduled in the early morning, when both the child and dentist are less tired, the child is more alert and better able to sit in the chair, and medication (when used) may be at its optimal effect; appointments should be avoided during known rebound periods. [11]

• Short, multiple visits are generally more successful than a few long sessions, and incorporating frequent breaks so the child can briefly engage in a preferred activity can further improve cooperation.[12]

• Communication should consist of clear, simple instructions that are repeated as needed, since repetition helps build the child's confidence. Bright, colorful educational materials can be useful for delivering oral hygiene messages.[13]

• The tell–show–do technique has significant value in pediatric behavior guidance, and the systematic use of praise, encouragement, and rewards help reinforce desirable behavior through positive reinforcement.[15]

• When children do not adequately respond to these conventional methods, carefully indicated protective stabilization may be considered to safely complete treatment. These children may also present with uncommon oral and dental findings, including unilateral or bilateral mandibular hypoplasia, often accompanied by a high-arched palate.

Case report :

A five-year-old female child , 1st issue of consanguineous parents hailing from Madagadipet Palayam, Pondicherry had reported to the dental op of department of paediatric and preventive dentistry on 9th June 2025 with the complaints of pain and swelling in the lower left back tooth region for past 2 days. Patient was apparently normal before 2 days after which she developed pain which is insidious in onset, intermittent in nature pricking type of pain

aggravates on mastication and nocturnal pain present. Presence of intra oral swelling in the region of the mucosa in relation to left lower back tooth region, tender on palpation, fluctuant and soft in consistency pedunculated. This a known case for acrorenal syndrome with classic feature of absence of left upper arm anomalies that is first metacarpal ray.(thumb) with left renal agenesis fig 1,2. Patient had habit of drinking milk when she is sleepy right from after birth, which formed the route cause for the poor oral health. Patient gives history of temporary restoration done in the same tooth 1 month before. On intra oral examination there was presences of an intraoral swelling in both left and ride side size 1cm x 1cm in the attached gingiva in relation to 75 region, and 85 regions respectively it is oval shaped sessile which is soft in consistency, fluctuant in nature reddish- pink in colour tender on palpation. However, there was class 1 dental caries in 85 with tender on probing and a temporary restoration in 75 with tender on probing , root stumps in 51, 52, 54, 62, grossly decayed in 64,53,63, class II dental caries in 65,84,74. The patient was advised for OPG. After meticulous clinical , and radiographic examinations, following diagnosis was given: Early childhood caries, chronic irreversible pulpitis in 75,85,64; grossly decayed in 53,63,51,52,54,62.

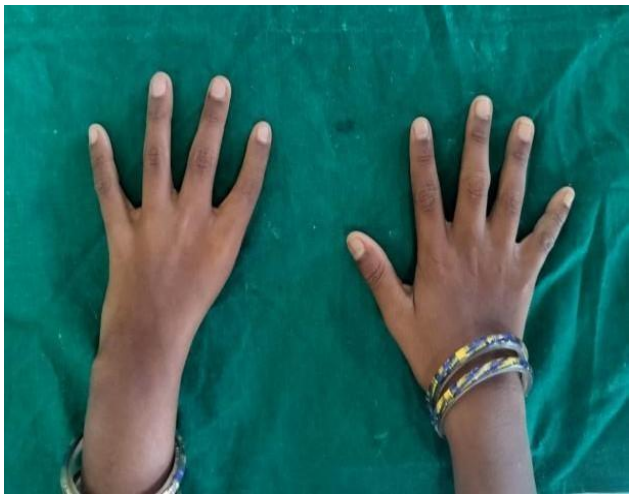

Fig 1: Absent left thumb

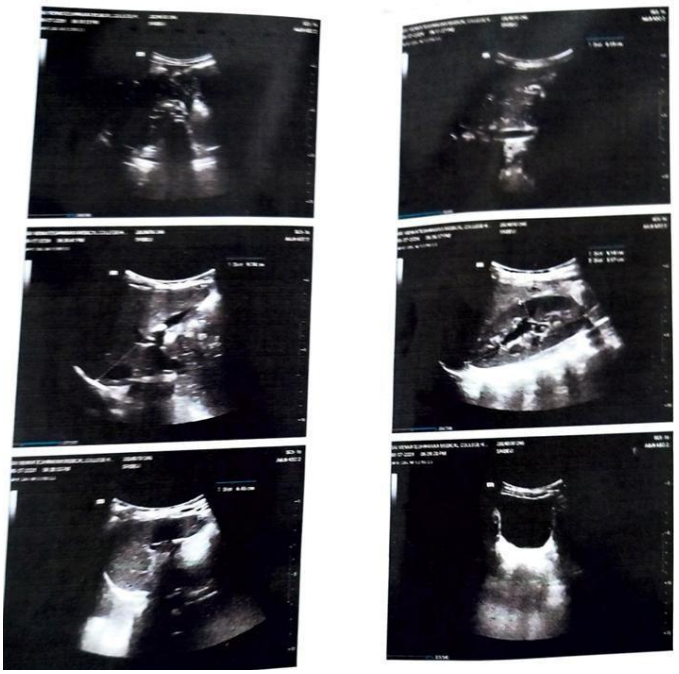

Fig 2: Renal ultra sonography

Therefore, the treatment plan was oral prophylaxis, pulpectomy with respect to 75,85,64 followed by stainless steel crown. Extraction of 51,52,54,64 followed by Gropper's appliance with restoration of 65, 84, 74.

The parents weren't willing for extraction of the tooth hence only symptomatic treatment was carried out. Fig 3

The treatment carried out were pulpectomy followed by placed of SScrown(3M™ Unitek™ Primary Stainless-Steel Crowns, USA) for both 75and85.

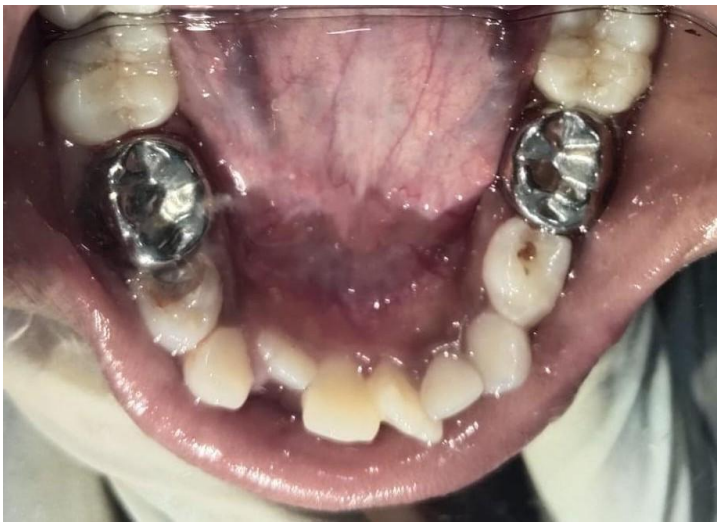

fig 3: Post symptomatic treatment intra oral image

Discussion :

Renal involvement in acrorenal syndrome shows a broad spectrum, extending from relatively mild abnormalities such as hydronephrosis associated with a duplex collecting system to advanced lesions, including bilateral renal hypoplasia and eventual renal failure. Although renal abnormalities are not extensively documented, most reported biopsy cases have revealed the presence of oligomeganephronia.[16,18]

Other associated anomalies may affect the oromandibular region, respiratory structures such as the trachea and lungs, and various skin-derived tissues including sweat and mammary glands. Involvement of the uterus, vas deferens, nasal placodes, and eyes has also been observed. Halal et al. (1980) were the first to report the occurrence of bilateral split-foot deformity, pronounced mandibular hypoplasia, and bilateral renal anomalies in two female siblings born to consanguineous French-Canadian parents. One of the siblings exhibited bilateral renal agenesis, while the other presented with bilateral polycystic kidneys.[19] Additionally, cleft hand deformity, apart from its association with acrorenal syndrome, has been documented in several other syndromes such as ectodermal dysplasia, ectrodactyly–

ectodermal dysplasia–clefting (EEC) syndrome, Cornelia de Lange syndrome, focal dermal hypoplasia, ectrodactyly with cleft palate syndrome, ectrodactyly–mandibulofacial dysostosis, and ectrodactyly with macular dystrophy.[20]

The pathogenesis of acrorenal syndrome has been attributed to disruption of the normal epithelial–mesenchymal signalling pathways during early embryogenesis.[17] These interactions are essential for the formation of the apical ectodermal ridge, which orchestrates proximal–distal limb outgrowth, and for the induction and branching of the ureteric bud that gives rise to the collecting system of the permanent kidney. In addition, the same epithelial–mesenchymal mechanisms contribute to the development of multiple craniofacial and ectodermal structures, including the palate, jaws, skin appendages, mammary glands, paramesonephric ducts, and ocular tissues, providing a unifying explanation for the constellation of skeletal, renal, and other anomalies observed in this syndrome.

From a clinical perspective, previously reported cases illustrate the variability in limb involvement and renal severity. Zeier et al. described an adult patient with acrorenal syndrome in whom the limb malformations predominantly affected the upper extremities, with comparatively milder changes in the lower limbs, highlighting the asymmetric expression of the defect in appendicular structures.1 The renal impairment documented in several earlier reports was stated to have severe impact on the renal and underscoring that renal pathology can be the major determinant of prognosis in acrorenal phenotypes. The severity deeply impacted the quality of life.

Conclusion:

This case report documents a rare instance of acro-renal syndrome managed with symptomatic dental treatment in a 5-year-old girl. Given the scarcity of published cases, this presentation adds to the limited literature available for advancing research on the syndrome's clinical spectrum. Timely diagnosis and intervention for dental caries remain critical to support masticatory function and sustain effective oral hygiene in such patients.

References :

1. Zeier M, Tariverdian G, Waldherr R, Andrassy K, Ritz E. Acrorenal syndrome in an adult– presentation with protein uria, hypertension, and glomerular lesions. Am J Kidney Dis 1989;14(3):221–224
2. Curran AS, Curran JP. Associated acral and renal malformations: a new syndrome? Pediatrics. 1972;49:716–725.
3. Bhandari S, Kalra S, Dwivedi A. Acrorenal syndrome: Unusual association of limb and renal anomaly. Indian J Nephrol 2022;32:182-3.
4. 4.Lash JW (1964) Normal embryology and teratogenesis. Am J Obst Gynecol 90: 1193- 1207.
5. Buchta RM, Viseskul C, Gilbert EF, Sarto GE, Opitz JM (1973) Familial bilateral renal agenesis and hereditary renal adysplasia. Z Kinderheilkd 11: 111-129. J Clin Image & Rep, 2023
6. Gilbert EF, Opitz JM (1979) Latteinte in the renal hereditary malformation syndromes. Ch 44 In Hamburger et al (eds); Addresses Nephrologic, Volume 2 Paris. Flammarion 936 970.
7. Dieker H, Opitz JM (1969): Associated acral and renal malformation. Birth Defects 5:68–77.
8. Nahar, Begum. (2016). Acrorenal Mandibular Syndrome: A Case Report. Journal of Bangladesh College of Physicians and Surgeons. 33. 95.
9. Abhinav Gupta, Mohd, Iqbal Bhat, Isha Kaul (2023) Acrorenal Syndrome: Case Series. Journal of Clinical Images & Reports. SRC/JCIR-117.
10. Desai SP, Shah PP, Jajoo SS, Smita PS. Assessment of parental attitude toward different behavior management techniques used in pediatric dentistry. J Indian Soc Pedod Prev Dent 2019;37:350-9.
11. Sujlana A, Dang R. Dental care for children with attention deficit hyperactivity disorder. J Dent Child 2013 May-Aug;80(2):67-70.
12. Cameron AC, Widmer RP. Handbook of pediatric dentistry. 4th ed. pp. 386-389.
13. Polancyzk G, de Lima MS, Horta BL, Biederman J, Rohde LA. The worldwide prevalence of ADHD: a systematic review and meta regression analysis. Am J Psychiatry 2007 Jun;164(6): 942-948.
14. Lewis MA, Shaw J, Sinha M et al. UK Renal Registry 11th Annual Report (December 2008): chapter 13 demography of the UK paediatric renal replacement therapy population. Nephron Clin Pract 2009; 111: 257–267
15. Efron D, Kilpatrick NM. Attention deficit hyperactivity disorder: a review and guide for dental professionals. J Disability Oral Health 2002;3(1):7-12
16. Miltenyi M, Czeizel AE, Balogh L, Detre Z (1992) Autosomal recessive acrorenal syndrome. Am J Med Genet 43: 789-790.
17. Evans JA, Phillips S, Reed M, Chodirker BN (2000) Severe acrorenal-uterine-mandibular syndrome. Am J Med Genet 93: 67-73.
18. Buttiens M, Fryns JP (1987) apparently new autosomal recessive syndrome of mental retardation, distal limb deficiencies oral involvement and possible renal defect. Am J Med Genet 27: 651-660.
19. Halal F, Desgranges M-F, Leduc B, Theoret G, Bettez P. Acro –renal-mandibular syndrome. Am J Med Genet 1980; 5: 277-84.
20. Buss PW. Cleft hand/foot: clinical and developmental aspects. J Med Genet.