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INTERNATIONAL JOURNAL OF ADVANCED RESEARCH (IJAR)

Article DOI:10.21474/IJAR01/20669
DOI URL: http://dx.doi.org/10.21474/IJAR01/20669

RESEARCH ARTICLE

DEFINING CHARACTERISTICS OF PRIMARY DENTITION IN A CHILD WITH ISOLATED PIERRE-ROBIN SEQUENCE: UNVEILING A NOVEL PHENOTYPIC EXPRESSION- A CASE REPORT

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Manuscript Info

Manuscript History
Received: 23 January 2025
Final Accepted: 26 February 2025
Published: March 2025

Key words:-
Pierre-Robin-Sequence, True Generalised Macrodontia, Tooth Agenesis

Abstract

The Pierre Robin sequence (PRS), initially identified in 1923, is a congenital condition defined by a triad of features: micrognathia, glossoptosis, and cleft palate (CP).[1] The occurrence of PRS is estimated to be between 1 in 8,500 and 1 in 30,000 live births.[2] This article explores a unique phenotypic expression of primary dentition in a child diagnosed with Pierre-Robin sequence.

Introduction:-

The Pierre Robin sequence is a well-established condition characterized by cleft palate, mandibular micrognathia, and glossoptosis, which refers to airway obstruction resulting from the tongue's displacement towards the lower, posterior part of the mouth.[34]

This condition can appear independently or be linked to different syndromes and anomalies. Stickler syndrome and Velo-Cardio-Facial syndrome are the two most frequently associated genetic disorders with Pierre Robin sequence. The condition was first described in 1891, and Pierre Robin published a case of an infant exhibiting these traits in 1923.[2]

This case report delivers a detailed assessment of a unique dental phenotype observed in the primary dentition of a pediatric patient diagnosed with Pierre-Robin sequence. It will detail the specific dental characteristics and developmental anomalies that may arise in children with this condition, highlighting the implications for dental management and orthodontic intervention.

Case Report

This is a case involving a 4½-year-old male child diagnosed with isolated Pierre-Robin sequence, who presented with complaints of crowding in both the upper and lower dental arches. He is the first child of a 35-year-old father and a 26-year-old mother, with no reported history of consanguinity within the family. The mother had been undergoing treatment for hypothyroidism during her pregnancy. The child had a birth weight of 3.2 kg and was delivered via C-section. Cleft palate repair was performed at the age of 1 year. Speech articulation delays were noted at 3 years of age, and he is currently receiving speech therapy at a nearby center. Additionally, he is undergoing behavioral treatment. However, his Karyotyping was of a typical male child.(Fig-V)

An extraoral examination revealed a convex facial profile with mandibular retrognathia,a short, thin upper lip, and an elongated philtrum (Fig. I). Height and weight measurements fell within age-appropriate standards.
A comprehensive intraoral examination revealed a striking case of generalized macrodontia affecting the primary dentition, characterized by significant crowding in both the upper and lower dental arches (Fig. II). The child's

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overall oral hygiene was notably poor, indicating a lack of consistent care. Multiple teeth exhibited smooth surfaces and proximal caries, suggesting areas of decay that had developed over time. Notably, there were nocaries on the occlusal surfaces, indicating a peculiar pattern in the dental health of this young patient.

Radiographic examination using OPG revealed an erupted macrodontic primary dentition, with all teeth having open apices. Fewer permanent tooth buds were noted in the maxillary and mandibular arches(Fig. III A &B).

Four primary canines were extracted to address the crowding and to improve oral hygiene practices. Furthermore, we provided dietary counselling and oral hygiene instructions, ensuring the patient is kept under constant follow-up (Fig IV A, B &C)

Discussion:-

Macrodontia refers to the presence of teeth that are significantly larger than what is typically considered normal.[5] It is estimated to affect between 0.03% and 1.9% of the global population. [6]A tooth or teeth called macrodontic may exceed the average size for their age and gender by more than two standard deviations..[7]

Odontogenesis is a complex process that relies on a coordinated interplay of genetic factors, growth signals, and transcriptional regulators. These components function through various signaling pathways to orchestrate the intricate stages of tooth development and ensure proper morphological and physiological formation. Any mutation in these genes and disruption of the regulatory molecules could result in a dental anomaly.[6]

True generalized macrodontia israre and is occasionally associated with conditions such as pituitary gigantism, Otodental syndrome, Ekman-Westborg-Julin syndrome, KBG syndrome, 47XYY syndrome, and Rabson-Mendenhall syndrome.[6]

The etiology of PRS is typically separated into isolated (non-syndromic) and syndromic PRS. Non-syndromic PRS has been associated with chromosome 2, 4, 11, or 17 mutations. Some evidence suggests SOX9 or KCNJ2 mutations (on chromosome 17) may affect the development of facial structures and cartilage, leading to this condition.[2] The syndromic PRS has recently been reported to account for 60% of cases. [8]There have been 34 syndromes associated with it.[9]

However, in some cases, individuals with PRS may have a normal karyotype, meaning their chromosomes are not visibly abnormal. At present, the exact cause of PRS is unknown. The most widely held view is that multiple contributing factors lead to a sequence of physical changes within the oral cavity. These changes are thought to occur in a series of steps rather than as isolated events.[2]

Regardless of the cause, the Pierre-Robin sequence presents as a triad of mandibular micrognathia, glossoptosis, and upper airway obstruction, often associated with a U-shaped cleft palate. These findings are consistent with the current case.Thechild's chromosomal study was that of a normal male 46XY; any underlying systemic conditions were not reported.The available literature data about the dental phenotype associated with a non-syndromic PRS is scarce.

Antonarakis & Suri (2014) and de Smalen et al. (2016) identified dental agenesis as a dental phenotype in individuals with nonsyndromic PRS. The prevalence rates they reported for tooth agenesis were 32.5% and 47.8%, respectively. [101112]

In another study, Jose Francisco et al. (2018) noted that taurodontism was a newly recognized and more common dental phenotype in non-syndromic PRS. They also found that the prevalence of tooth agenesis in non-syndromic PRS in their research was 22.72%.

This patient's OPG also showed the initiation of only a few permanent tooth buds. The child was 4 ½ years old at the time of the initial examination.Permanent tooth buds of four lower incisors were absent,containing permanent tooth buds of 11,16,21,26,33,34,36,43,44, and 46.However, a full complement of generalised Macrodontic primary teeth was present, and the radiograph showed that all the erupted teeth had open apices.

Conclusion:-

This patient experienced significant crowding in the upper and lower dental arches due to true generalized macrodontia associated with micrognathia. A comprehensive, multidisciplinary strategy is crucial for addressing developmental delays, speech difficulties, craniofacial disorders, and dental irregularities in these children, and ongoing oral hygiene education and prevention programs are equally important.

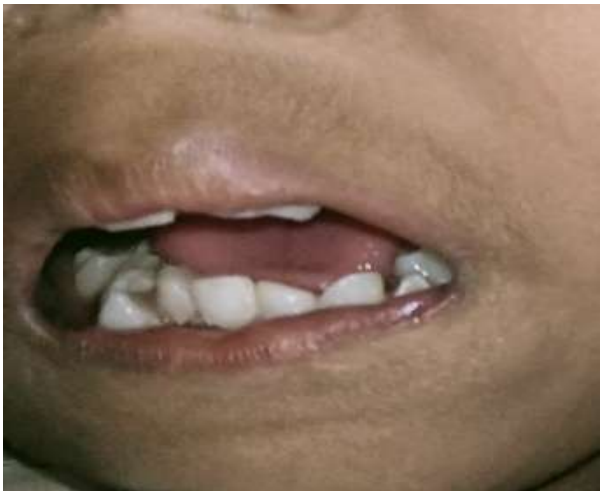

Fig I:-Extra-oral view,thin upper and lower lip with Micrognathia of the mandible.

Fig II:- Intra-oral view,true generalised Macrodontia ofprimary dentition.

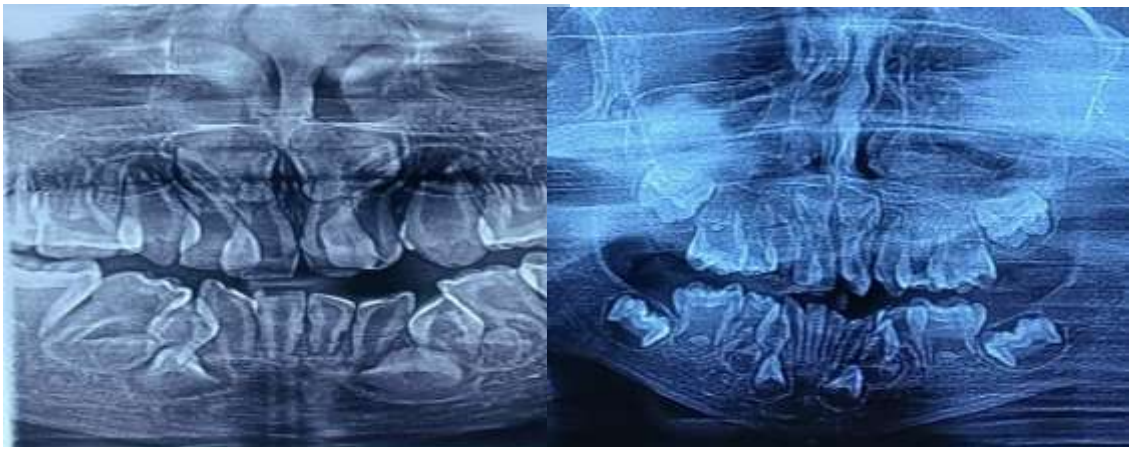

Figure III:-OPG of the Child- Full complement of macrodontic primary dentition with open apices. Tooth buds of 11,16,21,26,33,34,36,43,44, and 46 can only be seen.

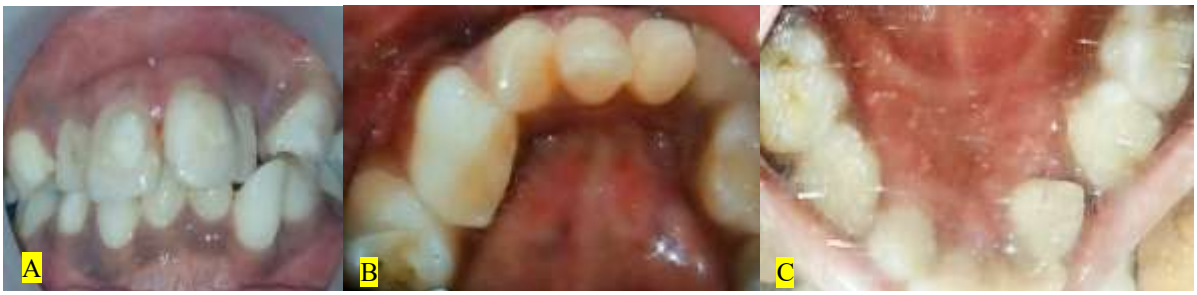

Fig IV A, B, C:-Intra-oral view after extraction of four primary canines.

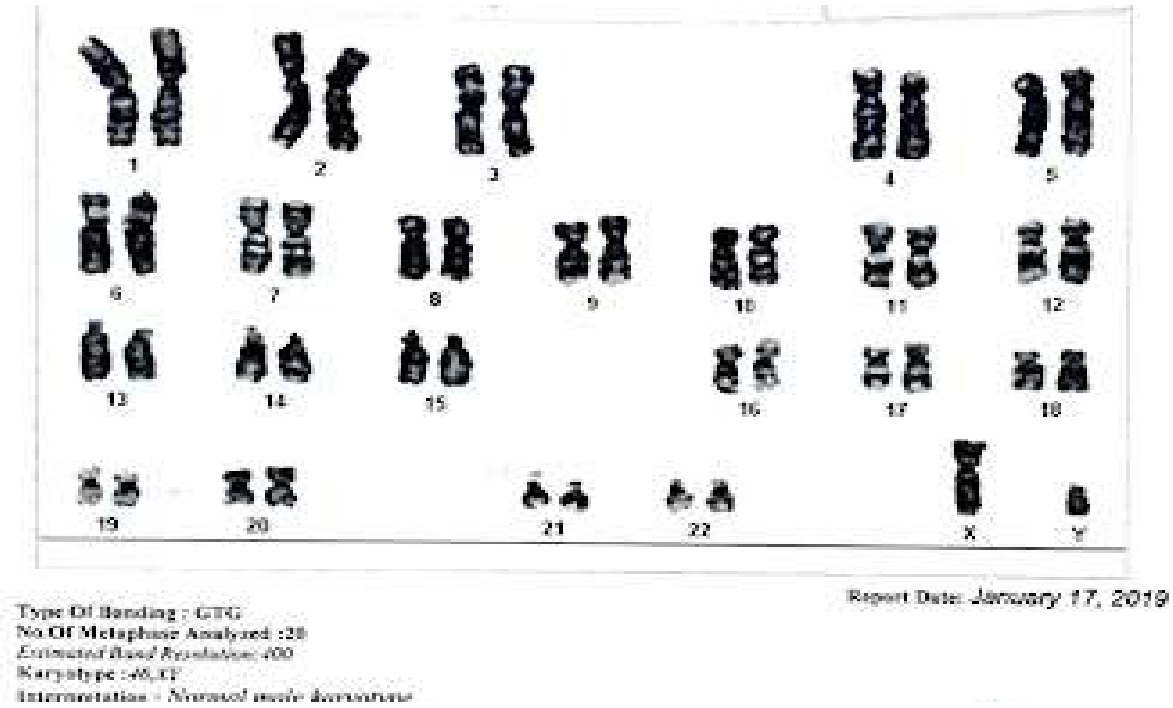

Fig. V:- Normal Karyotyping.

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