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Case Report

Dentistry Section

Unveiling the Rare Facets of Oral Manifestations of Radial Ray Syndrome: A Case Report

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ABSTRACT

Radial Ray Syndrome (RRS) is a rare congenital anomaly characterised by non progressive horizontal ophthalmoplegia and various systemic features, including abnormalities of the thumb, radial-sided carpals, and metacarpals. To date, there are not many documented cases that report oral manifestations associated with RRS. Hereby, the authors present a case of an 18-year-old female diagnosed with RRS. The patient exhibited skeletal Class III malocclusion, with maxillary retrusion and mandibular protrusion, counter-clockwise mandibular rotation, asymmetrical mandibular condyles, and multiple dental caries. A posteroanterior X-ray revealed asymmetry in the craniofacial structure. This case report aims to highlight the oral manifestations observed in a patient with RRS.

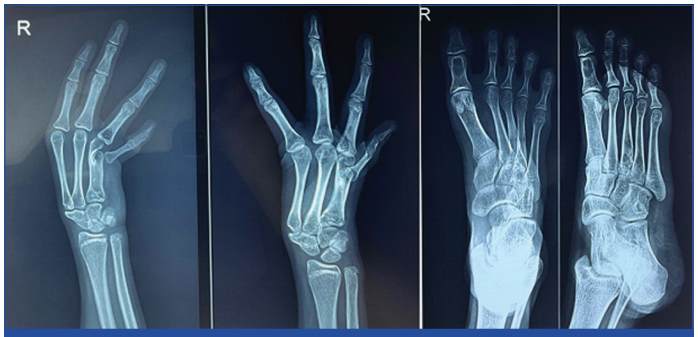
Keywords: Biochemistry, Chemical biology, Developmental biology, Duane-radial ray syndrome, Human

CASE REPORT

An 18-year-old female presented to the hospital with a complaint of malalignment of her teeth and expressed a desire to have it corrected. The patient reported a history of pulmonary tuberculosis two years prior, for which she had received treatment but was not on any medication at present. She also provided a history of being born out of a consanguineous marriage, resulting in an eutocic birth after a normal pregnancy.

A general examination of the patient revealed a petite and slender build, with asymmetry in posture due to bilateral limb deformities, as shown in [Table/Fig-1,2]. Extraorally, the patient had a distinct concave facial profile with pronounced micrognathia of the maxilla and a significantly protruded mandible, resulting in an Angle's Class III skeletal pattern. This was confirmed through cephalometric analysis, which will be discussed in the upcoming paragraphs. Her nasal bridge appeared depressed, and her ears were low-set, as shown in [Table/Fig-3].

[Table/Fig-1]: The patient exhibited a petite and slender build, with asymmetry in posture due to bilateral limb deformities.



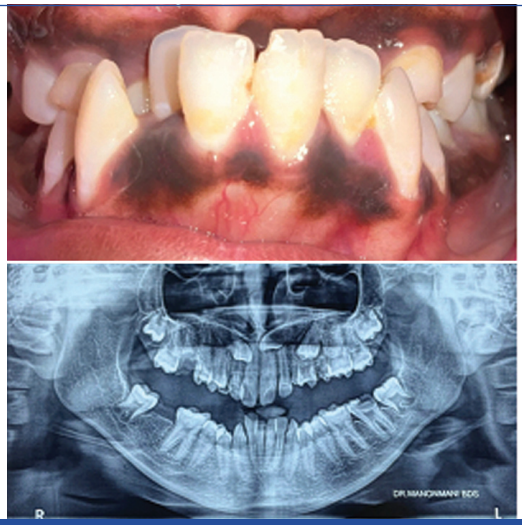
[Table/Fig-2]: Bilateral limb deformities are shown in radiographs of the patient suggesting a classic appearance of clinical features of the syndrome. X-rays of the forearms confirmed bilateral radial hypoplasia, with marked dysplasia of the carpal and metacarpal bones, resulting in radial deviation of the hands.

[Table/Fig-3]: Extraorally, the patient had a distinct concave facial profile with pronounced micrognathia of maxilla and a significantly protruded mandible, resulting in an Angle's Class III skeletal pattern. Her nasal bridge was depressed, and her ears were low-set but otherwise proportionate to her craniofacial structure

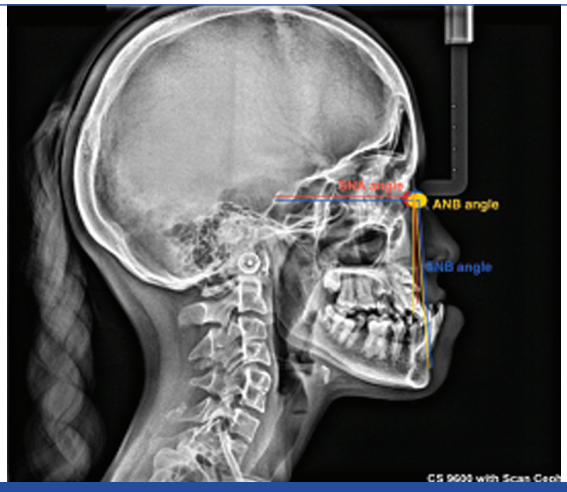
Intraorally, the patient displayed severe dental crowding in both the maxillary and mandibular arches. The malocclusion was identified as Angle's Class III, attributed to mandibular prognathism, with the mesiobuccal cusp of the upper first molar positioned behind the buccal groove of the lower first molar. Additionally, there were multiple findings of impacted permanent teeth and retained deciduous teeth.

Radiological investigations provided a detailed understanding of her skeletal abnormalities. The Orthopantomogram (OPG) revealed

multiple permanently impacted teeth, as shown in [Table/Fig-4], and retained deciduous teeth. A lateral cephalogram demonstrated a steep mandibular plane angle and a protruded chin, as shown in [Table/Fig-5]. X-rays of the forearms confirmed bilateral radial hypoplasia, with marked dysplasia of the carpal and metacarpal bones, resulting in radial deviation of the hands, as shown in [Table/Fig-2]. The lower limb radiographs revealed metatarsal and phalangeal anomalies, including notable medial deviation of the hallux and hypoplastic toenails.



[Table/Fig-4]: The malocclusion was identified as Angle's Class III, attributable to mandibular prognathism with multiple permanent impacted teeth and multiple retained deciduous teeth.



[Table/Fig-5]: A lateral cephalogram demonstrated maxillary retrusion and a protruded chin.

The lateral cephalometric analysis using Ricketts and Tweed norms [1] highlights significant deviations from normal craniofacial skeletal relationships. The Sella-Nasion-A point (SNA angle) was measured at 78.66°, which is significantly lower than the normal range of 82°±2°, indicating a retruded maxilla in relation to the cranial base. This suggests that the maxillary bone is underdeveloped or positioned posteriorly. The Sella-Nasion-B point (SNB angle) exceeded the normal range of 80°±2°, being measured at 84.09°. This finding indicates a prognathic mandible, meaning the lower jaw is excessively forward in relation to the cranial base, which contributes to a dominant lower facial profile.

The angle between A-point, Nasion, and B-point (ANB angle), calculated as the difference between SNA and SNB, was -5.43°, falling significantly below the normal range of 2°±2°. A negative ANB angle signifies a skeletal Class III malocclusion, typically characterised by a prominent mandible and/or a retruded maxilla. This skeletal pattern indicates significant disharmony, with a protruded mandible and an excessively retruded maxilla leading to an imbalanced facial profile.

The clinical investigations of the complete blood count revealed mild normocytic anemia, with a haemoglobin level of 11.1 g/dL, a low Red Blood Cell (RBC) count of 3.94 million/cmm, and a slightly reduced Packed Cell Volume (PCV) of 34.4%. The Mean Corpuscular Volume (MCV) and Mean Corpuscular Haemoglobin (MCH) are within normal ranges at 87.3 fL and 28.2 pg, respectively, indicating normocytic and normochromic red blood cells. The platelet count and total white blood cell count, including differential counts, are within normal limits. An SNP-Array underlined a microdeletion located in the long arm of chromosome 2, specifically in the 2q22.3 region. Given all these results, combined with clinical features consistent with Roberts syndrome, a rare congenital condition involving skeletal and craniofacial anomalies, these findings reflect the complex interplay of craniofacial, dental, and skeletal anomalies seen in RRS. This case highlights the critical need for multidisciplinary care involving orthodontists, maxillofacial surgeons, and orthopaedic specialists to address the patient's functional, aesthetic, and psychosocial concerns.

DISCUSSION

The RRS is a rare congenital anomaly characterised by the hypoplasia or aplasia of the radius, often accompanied by abnormalities in the thumb, radial-sided carpals, and metacarpals [2]. While its systemic and skeletal manifestations have been extensively studied, the oral and craniofacial features associated with this syndrome remain largely unexplored. This case report presents an 18-year-old female with RRS, highlighting significant oral and craniofacial anomalies that have not been previously documented in the literature.

In present case, the patient exhibited skeletal and craniofacial abnormalities, including micrognathia, a protruded mandible, a concave facial profile, low-set ears, and a depressed nasal bridge, contributing to an Angle's Class III skeletal pattern. Intraorally, the patient presented with severe dental crowding, class III malocclusion, multiple impacted permanent teeth, and retained deciduous teeth. These findings suggest that RRS, beyond affecting limb development, may also have significant implications for craniofacial and dental structures.

Cephalometric analysis further confirmed a skeletal Class III relationship, characterised by a reduced Sella-Nasion-A (SNA) angle (78.66°), an elevated Sella-Nasion-B (SNB) angle (84.09°), and a significantly negative ANB angle (-5.43°). Additionally, radiological investigations revealed bilateral radial hypoplasia with carpal and metacarpal dysplasia, as well as lower limb anomalies, including medial deviation of the hallux and hypoplastic toenails. The extraoral features in the limbs observed in this patient are consistent with previous reports on syndromic forms of radial ray anomalies, particularly in conditions associated with SALL4 mutations [3].

The SALL4 gene, known to play a critical role in limb and skeletal development, has been implicated in RRS [4]. Mutations in this gene can result in midface deficiencies, which were evident in this patient [5]. However, the presence of severe dental anomalies, including significant malocclusion and multiple impacted permanent teeth with retained deciduous teeth, further suggests that RRS may influence odontogenesis—a feature not previously emphasised in the literature.

A similar case of Duane-Retraction Syndrome (DRS) reported by Sicurezza E et al., described craniofacial abnormalities, including facial asymmetry, a fissured tongue, hypotrophic gingiva, microdontia, and enamel defects [6]. Although DRS is genetically distinct from the case presented here, the overlap in craniofacial and dental findings raises the possibility that radial ray anomalies may have a broader impact on oral structures than previously recognised.

Temtamy SA et al., (1975) and Temtamy SA and McKusick VA (1978) described a father and son with multiple congenital anomalies, with a notable emphasis on facial nerve weakness[7,8]. The

father presented with Duane anomaly, bilateral thenar and thumb hypoplasia, syndactyly of the index finger, a unilateral club hand deformity, and malrotation of both kidneys, with a partial horseshoe anomaly. The son exhibited significant facial nerve weakness along with congenital deafness, malformed pinnae, bilateral club hand with absent thumbs, and kidney abnormalities, including an absent right kidney and malrotation of the left kidney. Additionally, he had pectoral and upper limb hypoplasia. Despite the father's Duane anomaly, the son's ocular findings appeared normal [7,8].

In an Italian child with a mild form of DRS initially described by Parentin F and Perissutti P (2003), Miertus J et al., (2006) identified a heterozygous missense mutation in the SALL4 gene [9,10]. This mutation was also found in the affected father and maternal grandmother but was absent in the unaffected mother and twin sister. The proband exhibited mild features of DRS along with cranial midline defects, including facial dysmorphism, pituitary hypoplasia, and the presence of a single central incisor [9,10].

The complexity of present case underscores the need for a multidisciplinary approach in managing patients with RRS. The combined involvement of orthodontists, maxillofacial surgeons, and orthopedic specialists is crucial in addressing the functional, aesthetic, and psychosocial challenges associated with this condition. Early intervention, including orthodontic management, orthognathic surgery, and prosthetic rehabilitation, may improve both oral function and facial aesthetics, thereby enhancing the patient's quality of life. By documenting previously unreported oral manifestations of RRS, present case broadens the understanding of the syndrome's phenotypic spectrum. Further research is needed to establish the prevalence of craniofacial and dental anomalies in RRS and to explore the underlying genetic mechanisms contributing to these features.

CONCLUSION(S)

In conclusion, present case of an 18-year-old female with RRS highlights the distinctive oral manifestations of the condition, which have not yet been widely documented in the literature. Severe dental crowding, hypodontia, malformed dental roots, impacted teeth, and a high-arched palate, combined with significant craniofacial skeletal discrepancies such as mandibular prognathism and maxillary retrusion, represent critical findings that expand the understanding of this rare syndrome. These oral and craniofacial anomalies emphasise the importance of detailed documentation and reporting to enhance awareness and guide future management strategies. Furthermore, the present case underscores the necessity of a multidisciplinary approach, integrating orthodontic, maxillofacial, and orthopedic expertise to address the complex functional, aesthetic, and psychosocial challenges associated with RRS. By focusing on these underreported oral manifestations, this report contributes valuable insights to the existing body of knowledge and emphasises the need for further research in this domain.

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