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Case Report

AL Awadi Raas Rothschild Syndrome A Rare Case Report

Megha Bahal[1]*, Vanshika Saggar[2], Dushyant Rajput[2], Dr. Jaspreet Singh[3], Avishi Syal[4] and Ishmeet Sooch[4]

[1]Senior Lecturer, Department of Oral Medicine and Radiology, Baba Jaswant Singh Dental College, Hospital and Research Institute, Ludhiana, Punjab, India

[2]Associate Dentist Private Clinic, Ludhiana, Punjab, India

[3]Senior Lecturer, Department of Prosthodontics, Crown and Bridge in Baba Jaswant Singh Dental College, Ludhiana, Punjab, India

[4]Intern, Baba Jaswant Singh Dental College, Hospital and Research Institute, Ludhiana Punjab, India

***Corresponding Author:** Megha Bahal, Senior Lecturer, Department of Oral Medicine and Radiology, Baba Jaswant Singh Dental College, Hospital and Research Institute, Ludhiana, Punjab, India.

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Abstract

A genetic disorder is a health problem caused by one or more abnormalities in the genomic sequence. A syndrome is a recognisable complex of symptoms and physical findings which indicate a specific condition for which a direct cause is not necessarily understood. Rare diseases/syndromes are the ones which affect a small percentage of the population. Most rare diseases are genetic in origin and thus are present throughout person's life. Gross estimate of the rare or ultra rare diseases in South East Asian countries is 6-8% of the global population is widely accepted by researches. A head count of every rare/ultra rare disease is not possible as most of these diseases remain remain unreported. One such syndrome has been described here in this report which throws light on it being a deviation from normal yet non debilitating hence is considered a disorder.

Keywords: Genetic; Congenital; Oligodontia; Oligodactyly; Syndrome; Rare Disease

Introduction

Tooth agenesis is most recognisable developmental dental anomaly in humans and can be challenging to manage clinically. Recent advances in the field of molecular biology and human genetics have improved our understanding of the cause and its etiology [1]. Oligodontia that is the absence of more than 6 teeth in a human dentition can be syndromic/non-syndromic. Association between PAX 9 promoter polymorphisms and hypodontia in humans has been reported along with many other genetic abnormality in syndromic cases [2]. One such syndrome has been given symptomatic treatment only.

Al Awadi RAAS Rotshchild syndrome is a rare phocomelia syndrome characterised by limb pelvic hypoplasia with oligodactyly/syndactyly along with abnormal facial features and orofacial malformations. It is an autosomal recessive type of inheritance [3]. Severe limb recessive deficiencies have been reported in other well known genetic enteties such as Schinzel phocomelia syndrome, EEC syndrome, zimmer phocomelia syndrome, mand Baller Gerold syndrome but it's association with craniofacial malformations is classic feature of Al Awadi RAAS Rothschild syndrome with partial effects shown in Furrhamn's syndrome [4]. The aim of this report is to present a case of AARR syndrome with craniofacial anomalies and limb hypoplasia along with classic ectodermal abnormalities.

Case Report

The affected Case was of a 25-year-old Indian male who reported to the department of Oral Medicine and Radiology with the chief complaint of missing teeth, wanting them to be replaced. On further enquiring patient reported that he had not had his teeth erupted since childhood. Patient also revealed that the same condition existed with his elder brother who had scarce hair development and less fingers and absence of femur bone of the left along with no teeth development. The patient revealed that his sibling has got replacement of rod in place of femur bone of the left leg, 3 years back. On general examination of the patient he had only 3 fingers in his forelimbs and 2 fingers in his hind limbs with fusion of fingers leading to ologodactyly and syndactyly. The appearance looked like a lobster claw hand with median cleft in the hand and feet due to absence of central digital rays. There was no other dysmorphic feature in the hands and limbs and the anthropometry was within normal limits. Physical and systemic examination was within normal limits. On enquiring birth history he was a product of non consanguineous marriage and term normal delivery with

no significant perinatal events. The patient had distinctive facial features as prominent foreheads, thick lips and flattened bridge of nose. The patient reported to have sparse hair growth on head with growth of beard and moustache. He revealed that is deaf from the left ear since childhood and has less sweat production. He had pigmented lips and multiple moles and pigmentations seen the face.

On intra oral examination, patiënt only had 6 teeth present, had root stumps with respect to 17 and 23, Ellis class I fracture with respect to 13 and generalised recession present suggestive of chronic generalised periodontitis, generalised attrition and chronic irreversible pulpits with respect to 23 and 17. Soft tissue examination revealed a growth of smooth to firm pedunculated mass on the alveolar ridge on the left side with respect to 38 of size approximately 1cm in diameter which is non tender, non inflamed, pink in colour, suggestive of soft tissue fibroma or fibromatous polyp.

Orthopantomograph was done for radiological investigation which revealed micrognathia of maxilla and mandible, thin subcondylar region, partial edentulous maxillary and mandibular arch, severe residual ridge resorption, elongated stupid process bilaterally and round radio-opacity evident in relation to the right ramus of mandible, near the angle region of size 0.3-0.4 mm in diameter suggestive of a tonsillolith.

Informed consent of family members and patient was taken md mutation analysis was performed. Blood for collected as a sample for analysis which reveals.

Loss and mutation of T63 gene causing hypdontia and ectodermal abnormalities. Also frame shift mutation of PAX 9 gene was evident causing the etiology of oligodontia.

Intra oral appearance

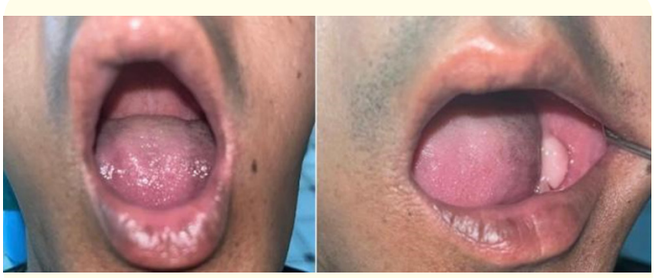

Figure 1: Clinical intraoral appearance.

Extra oral photographs

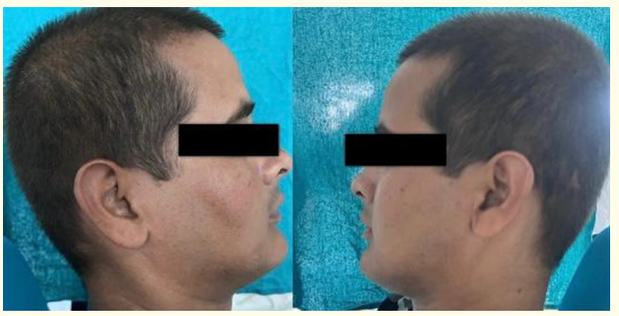

Figure 2a: Facial profile of the patient.

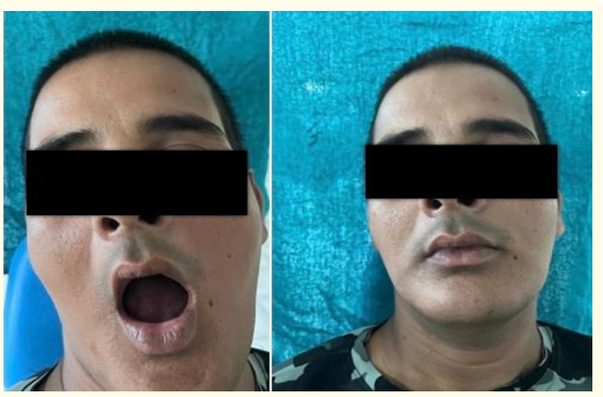

Figure 2b: Facial profile of the patient.

Radiographic investigations

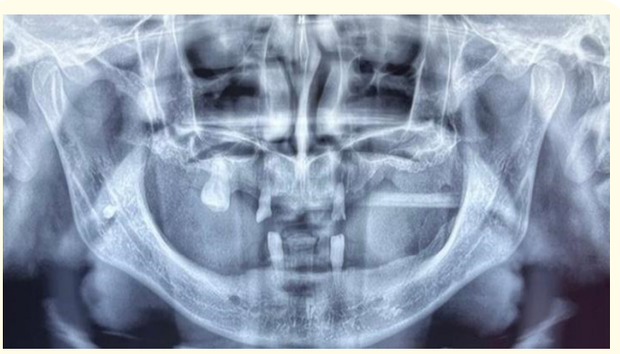

Figure 3: Orthopantamograph revealing the intraoral appearance.

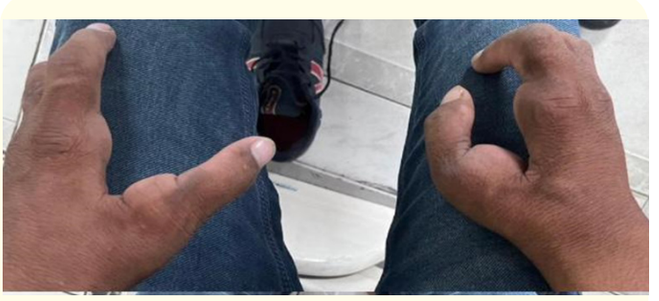

Figure 4: Syndactyly in upper and lower limbs.

Case Discussion

Craniofacial developmental defects are always intriguing. Al Awadi RAAS Rothschild syndrome when accompanied with dental and craniofacial abnormalities give several horrifying rare autosomal recessive limb malformation caused by WNT7A mutation [5]. Whole genome sequencing of the patient ruled out mutations in 3 hypodontia associated genes including WNT7A, PAX 9 and TP63 [6]. The classical features of Al Awadi RAAS Rothschild syndrome in abnormality of limbs having lobster hand growth abnormality, abnormality of craniofacial structure, tooth agenesis, abnormality of skin, hair and nails and less frequently Genito-urinary syndrome [7]. Partial loss of WNT7A gene leads to milder form of this syndrome termed as Furrhaman syndrome. All the states features along with aplasia of the fore and hindlimbs wads to Al Awadi RAAS Rotshchild syndrome [8].

Treatment options of these rare genetic and syndromic anomalies acquire symptomatic treatment mostly. Treatment depends on severity of specific symptoms of each affected person but may include surgical options. The clinical features of Al Awadi RAAS Rothschild overlaps with various other syndromes including Furhaman's syndrome, schinzel phocomelia syndrome and ECC syndrome [9].

Conclusion

Al Awadi RAAS Rotshchild syndrome is a distinct multiple malformation syndrome characterized by limb/pelvic hypoplasia/aplasia, renal anomalies such as horseshoe and polycystic kidney, and abnormal facial features including cleft palate, hypertelorism and micro-retrognatia [10]. This complex disorder affects the patient's overall Heath both physically and mentally. The treatment for the same requires a multidisciplinary approach.

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