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Insights into Axenfeld Rieger Syndrome: A Case Report

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ABSTRACT

Axenfeld Rieger syndrome is an extremely rare genetic disorder that occurs unprompted and is inherited in a way that is autosomal dominant. It has a frequency of 1 in every 200,000 individuals in the general population. This condition is defined by the abnormal development of the front part of the eye, as well as a range of dental, craniofacial, and somatic abnormalities. The wide range of patterns makes it difficult for health care professionals to diagnose. A comprehensive diagnostic evaluation and an interdisciplinary approach are crucial for identifying this uncommon condition. This article presents a case of Axenfeld Rieger syndrome with cerebral calcifications, which was detected by a thorough diagnostic investigation in a 10-years-old male patient.

KEYWORDS

Rieger syndrome; Ocular abnormalities; Dental anomalies; Genetic disorders; Multidisciplinary care

INTRODUCTION

Rieger syndrome, also known as Axenfeld-Rieger syndrome, is a rare hereditary condition affecting the eyes, teeth, and potentially other body parts. It's caused by mutations in the PITX2 and FOXC1 genes, which are crucial for embryonic development [1]. This condition causes ophthalmic abnormalities, dental irregularities, craniofacial issues, and heart problems, along with glaucoma, underdevelopment of the iris, and glaucoma displacement [2].

Rieger syndrome diagnosis involves a comprehensive assessment by experts like ophthalmologists, geneticists, and dentists. Imaging studies and genetic tests verify mutations. Treatment focuses on supportive care, with ophthalmic therapies like glaucoma and dental treatments like orthodontics. Frequent medical surveillance is crucial for early detection and management [3].

Rieger syndrome is a complex condition with a genetic basis that requires a comprehensive approach involving multiple disciplines. Despite its rarity, it is crucial to identify and treat it promptly to improve outcomes and overall well-being. Further research into the syndrome's genetic basis could lead to future therapeutic approaches.

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CASE REPORT

A 10-year-old male patient with missing teeth and vision abnormalities has been reported to the outpatient facility of the Department of Oral Medicine and Radiology. The patient's parents further gave a history, saying that only a few primary teeth erupted, and a few permanent teeth did not appear. The patient's prenatal history includes a full-term pregnancy and normal delivery, with no drug intake or trauma. On a detailed family history, his maternal uncle had similar symptoms but never received any assessment or treatment. The patient has an apparently normal older brother and a younger sibling with similar symptoms who died at the age of 2.

During the general physical examination, the team of experts from oral medicine and pediatric dentistry observed the salient features of short stature with a small build. Further, the patient also had prominent eyes with hypotelorism and prominent supraorbital ridges. Profound midfacial hypoplasia, and a beaded nose were also present (Figure 1). On intraoral examination, 16, 17, 23, 26, 27, 33, 34, 36, 37, 43, 46, and 47 were clinically visible, suggesting oligodontia as the other teeth were missing. The teeth present were microdontic and showed variable evidence of enamel mottling, suggestive of enamel hypoplasia. The patient also had a narrow palatal arch (Figure 2).

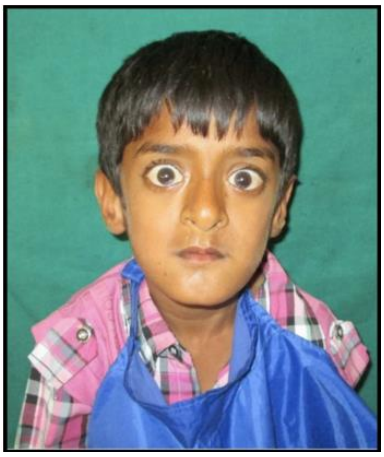

Figure 1: Extraoral frontal view of the patient showing the exophthalmos.

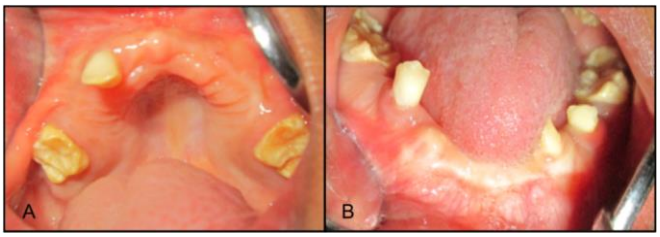

Figure 2: A) Intraoral maxillary arch; **B)** Intra-oral mandibular arch illustrating the oligodontia and mottling of enamel concurrent with the dysplastic changes.

An orthopantomograph was taken as a screening radiograph, where 13, 14, 15, 16, 17, 23, 26, 27, 33, 34, 35, 36, 37, 43, 44, 45, 46, and 47 were appreciated. Growing tooth buds of 37 and 48 were also noted in the mandible, which confirmed the diagnosis of oligodontia. Reduced radiolucency of enamel and dentin, along with large pulp chambers, was noted, which confirmed the diagnosis of enamel hypoplasia (Figure 3). A lateral skull view was also advised, which showed clear cut calcifications in the cranial cavity without involving the sella region (Figure 4).

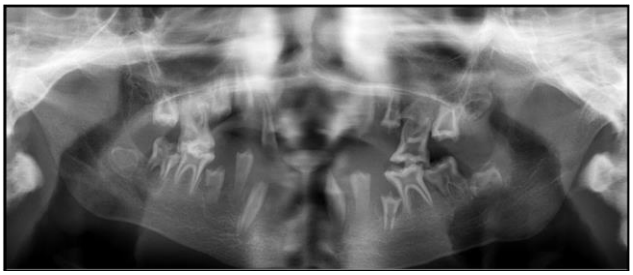

Figure 3: Orthopantomograph showing oligodontia and dysplastic enamel and dentin.

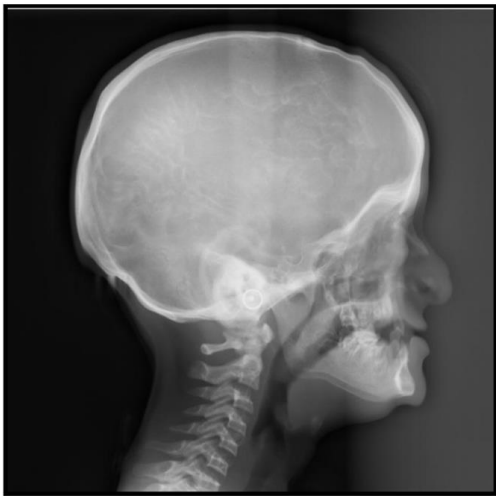

Figure 4: Lateral Cephalogram showing the tram-like calcification in the cranial cavity.

A computed tomography (CT) scan was done to confirm the extent of calcifications in the brain, which demonstrated extensive calcifications involving the basal ganglia and gyral calcifications of the frontal, parietal, temporal, and occipital regions (Figure 5). Radiographic examination of the extremities revealed irregular ossification of the femoral heads and distal femoral and distal tibial epiphysis. The patient underwent various laboratory tests, including a complete hemogram, thyroid profile, and serum tests, which were all within normal ranges, and was also seronegative for HIV.

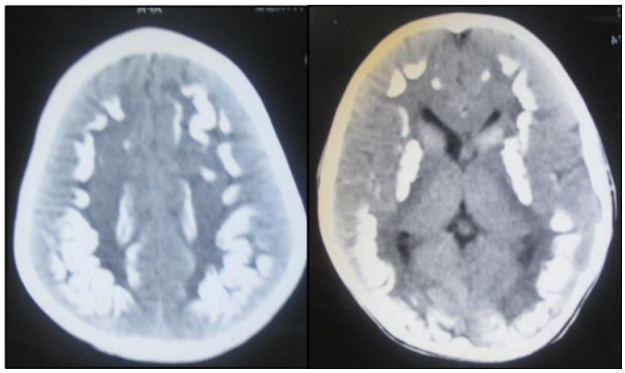

Figure 5: Computed tomographic images illustrating the extensive calcification involving basal ganglia, gyral calcifications of the frontal, parietal, temporal, and occipital regions.

The patient was also diagnosed with an atrophic hole in the right eye and corneal opacities in both eyes by ophthalmologists (Figure 6). A pediatrician confirmed partial failure of the periumbilical skin to involute and advised abdominal ultrasonography to rule out an inguinal hernia.

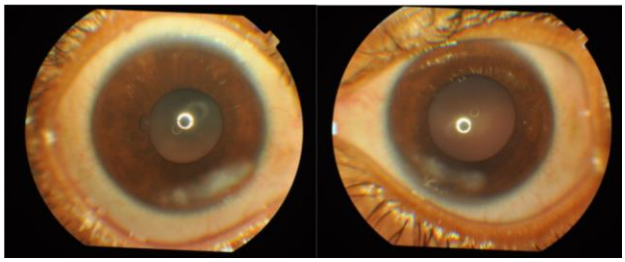

Figure 6: Ophthalmic evaluations, which show the corneal opacities.

After a comprehensive analysis of the history and other investigations, the patient was diagnosed with Axenfeld Rieger syndrome, and the family was educated about potential future complications. The patient was also advised of various preventive therapies, which included routine oral hygiene practices, fluoride application every three months, and routine mouth rinsing. Rehabilitative care was also planned for restoration using atraumatic restorative therapy (ART) and stainless-steel crowns for all the erupted permanent molars. Replacement of missing teeth of 11, 12, 21, 22, 31, 32, 41, and 42 was planned, followed by eruption of remaining permanent teeth for anchorage for a better prognosis.

DISCUSSION

Axenfeld Rieger syndrome, also known as Iris dental dysplasia, was initially documented by Rieger in 1934. It is a rare genetic ailment that follows an autosomal dominant pattern of inheritance. The prevalence of this condition in the general population is around 1 in 200,000, and there have been limited examples documented in scientific literature so far [4,5].

This syndrome is characterized by a diverse range of physical abnormalities and is inherited in an autosomal dominant manner with varying degrees of expression. The two genes associated with this condition have been identified as RIEG1, located at 4q25-q26, and RIEG2, located at 13q14 [6]. Although Axenfeld Rieger syndrome is typically described as having total penetrance, there have been a few case reports that demonstrate inadequate penetrance, such as the present instance [4].

This syndrome causes ocular symptoms such as underdevelopment of the iris, corneal adhesions, Schwalbe lines, corneal flaws, pupil abnormalities, cataracts, and early glaucoma in young individuals, with most symptoms observed in both eyes [1]. The presence of only ocular findings is called Rieger's anomaly [7]. The patient in question exhibited an atrophic hole in the iris of the left eye and corneal opacities in both eyes. These

symptoms can be linked to the varying expressivity of this condition.

The primary dental symptom that stands out is oligodontia, which affects both the primary and permanent teeth. The maxillary incisors, canines, and premolars are frequently absent in individuals. Additional dental manifestations encompass microdontia, enamel hypoplasia, conical teeth, delayed tooth eruption, taurodontism, and shorter dental roots [8]. The dental findings of oligodontia, enamel hypoplasia, and conical shaped teeth were consistent with the findings of the present case.

The patient displayed a lack of involution in the skin around the umbilicus and partial failure, indicating varying expressivity of the disease, leading to various presentations [9]. The patient had psychomotor impairment and irregular ossification of the humerus, which have been previously documented in association with this condition [6]. Additional characteristics that can be linked to this syndrome include hypospadias, inguinal hernia, Meckel's diverticulum, and empty sella syndrome [6]. However, these findings were absent in the present patient.

Moog et al. documented a case involving female siblings who had Rieger's syndrome and leptomeningeal calcification [10]. Thus far, there have been no recorded cases of Axenfeld Rieger syndrome involving thick calcifications of the brain. The first documented case of Axenfeld Rieger syndrome with such extensive brain calcifications is the present case, highlighting the need for increased attention and documentation by healthcare professionals.

CONCLUSION

Axenfeld Rieger syndrome is a rare genetic syndrome that is not often seen in clinical practice. This might make it difficult for health care practitioners to diagnose. Oligodontia can serve as the first indication that leads to the diagnosis of this disease. Therefore, as oral diagnosticians, it is essential for us to possess extensive knowledge and a strong desire to gather a meticulous family history. An exhaustive diagnostic investigation and an interdisciplinary approach are essential in order to definitively diagnose this disease.

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