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Wiedemann–Rautenstrauch Syndrome: Case Report

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Case Report

History

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ABSTRACT

Neonatal-progeroid syndrome known as Wiedemann Rautenstrauch syndrome (WRS) is an extremely rare, autosomal recessive disorder. Neonatal progeroid disease characterized by progeroid appearance growth retardation, lipodystrophy, an unusual face (triangular shape, sparse hair, small mouth, macrocephaly, pointed jaw), thin skin, hard and thick joints and dental anomalies (newborn tooth; hypodontics). A 5-year-old boy case diagnosed with hypodontics is presented. In this case, a child prosthesis was performed to facilitate the nutrition of the patient and to increase the quality of life.

Keywords: Alkaline Phosphatase, Orthodontics, Osteoclasts, Risedronic Acid, Tooth Movement.

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Introduction

Neonatal-progeroid syndrome known as Wiedemann–Rautenstrauch syndrome (WRS) is an extremely rare, autosomal recessive disorder [1] and was first described by Rautenstrauch and Snigula in 1977 and delineated by Wiedemann in 1979.[2] Neonatal progeroid disease characterized by progeroid appearance growth retardation, lipodystrophy, an unusual face (triangular shape, sparse hair, small mouth, macrocephaly, pointed jaw), thin skin, hard and thick joints and dental anomalies (newborn tooth; hypodontics).[3] Although Jay *et al.*[3] reported bi-allelic truncating variations in *POLR3A* in a single patient and hypothesized these to be causal for WRS, the main cause for WRS still remain unknown.[4] It is inherited autosomal recessively and biallelic pathogenic variants in the POLR3A gene form the basis of the WRS phenotype.[5] Most of these patients die in the first days and months after birth[6] but there are reports presenting survival into 16-17 years of age.[7,8]

Here in this case, we report a 5-year-old male that has classical clinical features of WRS.

Case Report

A 5-year-old boy presented to Sivas Cumhuriyet University Faculty of Dentistry Department of Pedodontics with a complaint of not being able to eat due to lack of teeth. The parents did not have any congenital anomalies and he was the first child of the family. According to the anamnesis taken from the parents, it was learned that the parents had the same syndrome in their second child. In extra-oral examination performed in our clinic, it was observed that hair formation was rare, scalp veins were prominent and facial features was abnormal. In addition, cranio-facial disproportion was observed according to age due to micrognathia, hypodontia, small mouth with thin upper vermilion, prominent scalp veins, widespread hair loss, deep set eye (Figure 1). The patient's mental development was also normal.

In intra-oral examination of the patient, only right maxillary canine (53), left maxillary lateral (62) and mandibular central teeth (71, 81) were seen in the mouth and shape anomalies were observed in these teeth. The mandibular crest had a knife edge appearance and the tongue size was normal. When the patient's panoramic radiograph was examined, it was seen that the deciduous teeth did not have root development and there were no permanent tooth germs underlying deciduous teeth. However, mandibular permanent 1st molar germs were seen in panoramic radiograph (Figure 2).

As a result of the examinations and the patient's complaint of inability to feed, a child prosthesis was planned for this patient. A 3-month follow-up was recommended to the patient, and his parents were informed that the prosthesis could be renewed depending on the eruption of the teeth and growth and development of jaws (Figure 3).

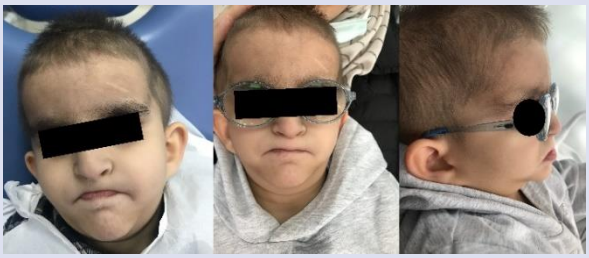

Figure 1.

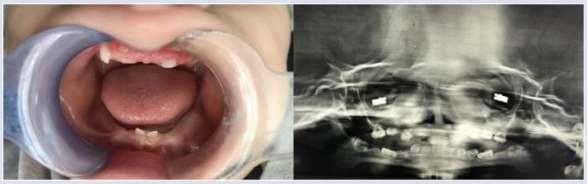

Figure 2.

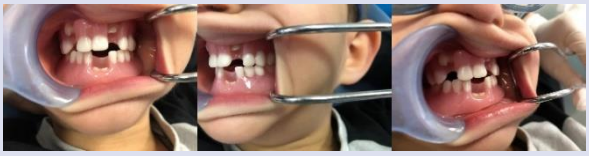

Figure 3.

Discussion

WRS is a rare autosomal recessive disease of unknown pathogenesis with progeroid appearance from birth.[9] Patients with this syndrome can be diagnosed at birth due to a range of abnormalities such as short stature, growth retardation, progeroid appearance, large fontanelles and sutures, prominent skin vessels, hypoplasia of facial bones, sparse scalp and eyebrows.[2,10] Pathogenic variants in the POLR3A gene are associated with hypomyelination, hypodontics, and hypogonadotropic hypogonadism.[5] Hutchinson-Gilford progeria syndrome (HGPS), Cockayne syndrome, Hallermann - Streiff syndrome (HSS), Werner's syndrome or Pelizaeus Merzbacher disease may phenotypically mimic WRS, but WRS is different from other premature aging syndromes[2,11], because all these changes are present at birth.[6,7,10,12,13]

HGPS has different facial features and includes prominent eyes and a beaked nose, with a characteristic bird-like facial appearance. Typical craniofacial features include easy visualization of scalp vessels, alopecia, micrognathia, and craniofacial disproportion.[14] Disorder in the growth of both maxilla and mandible, tooth crowding, irregular tooth eruption and localized enamel hypoplasia are seen.[15-18] Although the dental symptoms are partially similar with the patients with WRS, their phenotypic features are different. Characteristic features of Cockayne syndrome; microcephaly, cachectic dwarfism and progressive neurological degeneration, sensorineural hearing loss, cataracts, pigmentary retinopathy, photosensitivity and dental caries.[19,20] It differs from WRS because of these features. In addition, patients with WRS have hypodontia and oligodontia[3] and dental caries are uncommon.

HSS patients typically have bird-like facial features. Hypotrichosis, various ophthalmic disorders and dental abnormalities such as oligodontia, natal-neonatal teeth, enamel hypoplasia and supernumerary teeth are seen.[21,22] Although dental anomalies seen in this syndrome are also seen in patients with WRS, they do not show the ocular findings of HSS and are slightly similar in terms of facial features.

In our case report, patient has hypodontia and the erupted teeth had abnormal morphological structures and the lack of tooth structure may cause speech disorders, problems in the temporomandibular joint.[23,24] Due to the patient's nutritional problem, the progress of body development may slow down and a child prosthesis can overcome this problem by eating different kinds of nutritional products. A child prosthesis can increase the upper face height and get rid of the patient from elderly facial seem. For these reasons, a child prosthesis was performed to facilitate the nutrition of the patient and to increase the quality of life.

Conclusions

As a result, child prosthesis can be made to maintain the physical and functional development of these patients, to support them psychologically, and thus to increase their quality of life.

Consent

Informed consent was obtained from the patient parent for publication of this case report and accompanying images.

Acknowledgements

None

Conflict of Interest Statement

The authors deny any conflicts of interest related to this study.

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