A CLINICAL CASE REPORT OF HYPODONTIA AND IMPACTED SUPERNUMERARY TEETH IN A PATIENT DIAGNOSED WITH YUNIS-VARON SYNDROME

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ABSTRACT

INTRODUCTION: Yunis-Varon syndrome (YVS) is an extremely rare autosomal recessive congenital disorder affecting many organs and systems (bones, ectodermal structures, the cardiorespiratory system, etc.). Abnormalities include aplasia or hypoplasia of the clavicles, facial dysmorphism, sparse hair, absence or hypoplasia of the thumbs, toes and distal phalanges, prominent auricles, shortened upper lip and other skeletal deformities, such as pelvic dysplasia, kyphosis, scoliosis, etc. Tooth structures and jaws are characterized by marked micrognathia, a narrow high-arched palate, hypodontia, impacted permanent teeth as well as supernumerary teeth.

AIM: The present case report aims to describe the clinical and radiographic changes in the masticatory apparatus of a patient diagnosed with Yunis-Varon syndrome.

CASE PRESENTATION: The subject of this study was a patient, suffering from pain in tooth #38. The clinical intraoral examination revealed hypodontia, but the radiographic image revealed many supernumerary teeth in the anterior and premolar region of both the maxilla and the mandible.

RESULTS: In addition to skeletal anomalies and impaired ectodermal structures, this rare syndrome can cause disturbances in the teeth and jaws. For this reason, dental specialists play an important role in the diagnosis and treatment of patients suffering from Yunis-Varon syndrome.

CONCLUSION: Early intervention is important in Yunis-Varon syndrome. Future investigation will permit identification of these skeletal and dental abnormalities.

Keywords: *Yunis-Varon syndrome, hypodontia, supernumerary teeth, impacted teeth*

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INTRODUCTION

Yunis-Varon syndrome (YVS) is an extremely rare autosomal recessive congenital disorder affecting many organs and systems (bones, ectodermal structures, the cardiorespiratory system, etc.). The syndrome was first described in 1980 by Emilio Yunis and Humberto Varon, hence the name of the disorder (1). Since its discovery, only a limited number of cases have been reported in literature,

detailing changes in internal organs, such as the heart and brain, as well as skeletal system anomalies affecting the phalanges, the skull, eyes, ears, lips, and other structures of ectodermal origin (2).

AIM

The present case report aims to describe the clinical and radiographic changes in the masticatory apparatus of a patient diagnosed with Yunis-Varon syndrome.

CASE REPORT

A male patient of apparent age of 45 years, suffering from pain in tooth #38, sought assistance at the University Medical and Dental Center of the Medical University of Varna, Bulgaria. The patient seemed auto- and allopsychically oriented and of normal physical and mental development. The extraoral examination showed a prominent frontal bone, medium build, narrow shoulders, shorter thumbs on both hands, and shorter distal phalanges of the fingers. The patient's medical history listed a double left clavicle.

The clinical intraoral examination revealed a high-arched palate, marked micrognathia, a tented upper lip, and hypodontia. The dental status showed hypodontia, with the following teeth missing: maxillary central and lateral incisors, maxillary canines and maxillary first premolar on the left. Teeth #17, #16, #15, #25, #26, and #27 were present in the oral cavity. Some of them had fillings, while tooth # 15 had not fully erupted. Mandibular teeth #35, #33, #43, and #44 were missing. The radiographic image revealed many supernumerary teeth in the anterior and premolar region of both the maxilla and the mandible as well as an extra tooth distal to each of the mandibular wisdom teeth. In the region of the mental protuberance, at the lower mandibular edge, 8 supernumerary teeth were radiologically visualized. Most of the supernumerary teeth in both jaws had a premolar shape (paramolars), with well-developed crowns and roots. The patient was offered a treatment plan involving surgical, orthodontic, and prosthetic interventions to restore the normal masticatory and speech functions as well as the aesthetics. Following consultation with an orthodontist, it was agreed that the surgical treatment should involve a stepwise extraction of the supernumerary teeth and those that did not seem eligible for orthodontic treatment. The ultimate aim was to restore the masticatory apparatus and achieve functional and aesthetic restoration through fixed and removable prostheses. The patient was informed of potential complications during surgery due to the location of some of the mandibular supernumerary teeth, especially those in the mentum region and the mandibular edge.

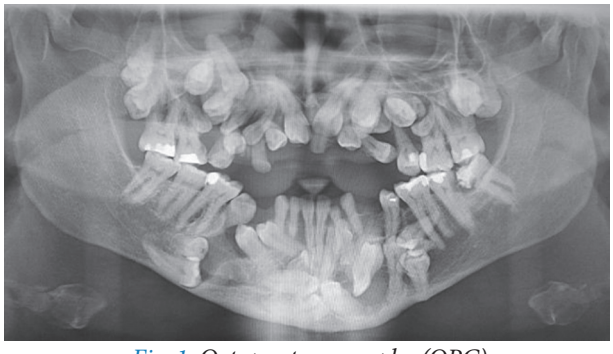

Fig. 1. Ortopantomography (OPG).

DISCUSSION

All reported cases of YVS share many common characteristic features. These include cleidocranial dysostosis, micrognathia, absence of thumbs and distal phalanges of the fingers, hypoplasia of proximal phalanges, absence of the distal phalanges of the toes, hypoplastic nails, pelvic dysplasia, bilateral hip dislocation, and thin, retracted and poorly defined lips (1). Similar patterns of abnormalities were observed by Hughes and Partington, who proposed the eponym of Yunis-Varon syndrome (3). Subsequently, further pathological associations have been described that were not previously reported, such as hearing impairment, pyloric stenosis (4), atrophy of the left lobe of the liver, anomalous hepatic vessels (5), congenital heart malformations (6), osteodysplasty (7), physical and mental retardation. Other symptoms linked to YVS include hypoplastic or missing clavicles, thin fragile bones, craniofacial disproportion with severe micrognathia. Additional YVS abnormalities have also been reported, such as sparse eyelashes and eyebrows, loose skin, deformed earlobes, prominent eyeballs (8). Microcephaly is detected in most cases. More serious associations, such as brain malformations, sclerocornea, bilateral cataracts, flat metaphyseal bones, pathological fractures, etc., have also been documented (9).

Yunis-Varon syndrome is a genetic condition that has an autosomal recessive inheritance pattern where a mutated gene is passed down from both parents to a child. If a child receives one unaffected (dominant) gene from one parent and one changed (mutated) gene, causing the disorder, from the other parent, the person will be a carrier of the disease but will not typically display its symptoms. Some cases of Yunis-Varon syndrome reported consanguineously married parents, who are more likely to pass this autosomal recessive disease on to their offspring. The disorder is caused by mutation in the FIG4 gene, which does not necessarily predispose to the development of the YVS. Mutations of the human FIG4 have also been identified in individuals with Charcot-Marie-Tooth disease, amyotrophic lateral sclerosis (ALS) and bilateral temporooccipital polymicrogyria (10,11,12,13). The FIG4 is responsible for a protein regulating the transportation of vesicles inside cells by modifying molecules called phosphoinositides. According to another theory of the development of this genetic condition, there is evidence of lysosomal storage disturbances leading to metabolic disorder and accumulation of toxins in the cells, affecting various parts, organs, and systems (14). The diagnosis of Yunis-Varon syndrome-YVS can be confirmed only after thorough clinical and paraclinical examinations or by establishing mutation in the FIG4 gene.

Most patients with this syndrome, displaying severe multisystem abnormalities, do not reach adulthood. The expected prognosis of YVS is poor. However, diagnosed individuals do not always present with all clinical symptoms of the disorder. In some patients, disturbances in the limbs and clavicles are observed while others, as in the clinical case described, exhibit hypodontia and many impacted supernumerary teeth, leading to functional and aesthetic issues.

Differential diagnosis is commonly achieved when cleidocranial dysplasia (CCD) is confirmed. Cleidocranial dysplasia is a rare genetic bone abnormality characterized by hypoplastic or anaplastic clavicles, delayed closure of cranial sutures and multiple dental anomalies (15). Other conditions and diseases associated with hypo- or anaplasia of the clavicle, such as mandibuloacral dysplasia (MAD), should also be considered as a differential diagnosis (16). The presence of hypodontia and impacted supernumerary teeth is usually associated with other diseases and syndromes, such as cleft lip and cleft palate, trichorinopharyngeal syndrome, Gardner's syndrome, etc. (17).

The present case report concerns a patient who was diagnosed with YVS. It is a rare autosomal recessive hereditary disease affecting the development of the bone system and ectodermal structures. The syndrome is characterized by peri- and postnatal growth retardation, skeletal anomalies, hypo- or anaplasia of the clavicles, cleidocranial dysplasia, hypoplasia or absence of the thumbs and toes, and distal aphalangia. What was distinctive about this case was that, apart from the intraoral peculiarities, involving hypodontia and delayed eruption of teeth in the permanent dentition and many supernumerary teeth, there were no clearly marked extraoral features associated with the syndrome. Although the prognosis of YVS is poor, not all patients go on to develop the typical clinical picture of the disorder. The treatment is symptomatic. With regard to the periodontium, a multidisciplinary approach is necessary to fully restore the affected masticatory and speech function as well as the aesthetic looks to improve patients' quality of life.

CONCLUSION

In addition to skeletal anomalies and impaired ectodermal structures, this rare syndrome can cause disturbances in the teeth and jaws. For this reason, dental specialists play an important role in the diagnosis and treatment of patients suffering from Yunis-Varon syndrome.

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