

## CLINICAL AND RADIOLOGICAL EVALUATION OF TER HAAR SYNDROME: CASE REPORT OF A PATIENT WITH EXTREME LONGEVITY

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### ABSTRACT

Ter Haar syndrome (THS) is one of the most deleterious disorders known in medicine today. Cases of THS are rare in the literature, and dental treatment for these children has never before been reported. Here we discuss the preventive and conservative treatment we provided to a patient with THS, and review the literature on the subject.

### INTRODUCTION

Ter Haar syndrome (THS) was first described in 1982<sup>1</sup> and is characterized by congenital glaucoma, craniofacial abnormalities, hypertelorism, kyphoscoliosis, skeletal dysplasia, congenital heart defects, and developmental delay. THS has an autosomal recessive inheritance pattern. The cardiac components of the disease are the most significant, as they are the primary causes of mortality.<sup>1-4</sup> Typical radiologic findings with this syndrome are bony sclerosis of base structures, abnormal dental alignment, sclerosis and irregularity of clavicles and ribs, described as “band-like” deformity; and cortical irregularity of tubular bones, with metaphyseal flaring and diaphyseal bowing. The flat bones are broadened, particularly the iliac crest and vertebral bones.<sup>5</sup> Cases of THS are rare in the literature, and dental treatment for these children has never been reported.

Disabled individuals’ physical, cognitive, and behavioral limitations require modification of standard preventive and conservative dental practices, including the choice of materials and techniques used. On World Health Day at the World Health Organization in 1994, a new method for treating dental caries was presented for developing countries. This approach, called *atraumatic restorative treatment* (ART), was introduced as a potentially viable means of providing restorative and preventive care. The ART approach uses glass ionomer cement (GIC) as a restorative material and fissure sealant in permanent and primary teeth, and combines both preventive and restorative procedures.<sup>6</sup> GICs represent an alternative to resin-based materials, especially when resins are contraindicated, as in the clinical treatment of children with deeply pitted or fissured primary molars, or permanent first or second molars that have not fully

emerged and whose isolation can be difficult, such as in young children and the disabled.<sup>7</sup>

We have provided preventive and conservative treatment to the longest-lived patient with THS. In this report, we present his case and review the associated literature.

### REPORT

In July 2003, a 6-year-old boy was referred to the Department of Pediatric Dentistry of Marmara University in Istanbul, Turkey. The patient presented with discomfort on eating. He was genetically diagnosed with THS immediately after his birth. Clinical history revealed that he was born full term as the fifth child, with a birth weight of 2000 g. Although the boy had 2 living healthy sisters, the family experienced the loss of 3 children, possibly from heart failure at the ages of 4, 3, and 1. All 3 children had similar syndromic face overview, but none of them had genetic testing to define any possible syndromes. The patient’s parents are relatives—his mother and father are cousins. The mother used acetylsalicylic acid (aspirin) during pregnancy for hypertension.

We report on this patient with congenital glaucoma, congenital heart defects, brachycephaly with flat occiput, large anterior fontanel, hypertelorism, inguinal hernia, kyphoscoliosis, megalocornea, exophthalmos, large ears, short hands and feet, flexion deformity of the fingers, and clubfeet (**Figure 1**). He had a congenital mitral prolapse, severe mitral deficiency, pulmonary deficiency, severe pulmonary hypertension, and pericardial effusion. There was no mental retardation. In his history, he had trabeculectomies to decrease his intraocular pressure caused by glaucoma when he was 2 days old (however,

he had right-eye blindness); an orchiectomy for cryptorchidism when he was 4 years old, and frequent respiratory tract infections. He received a mitral valve repair at 6 years of age.

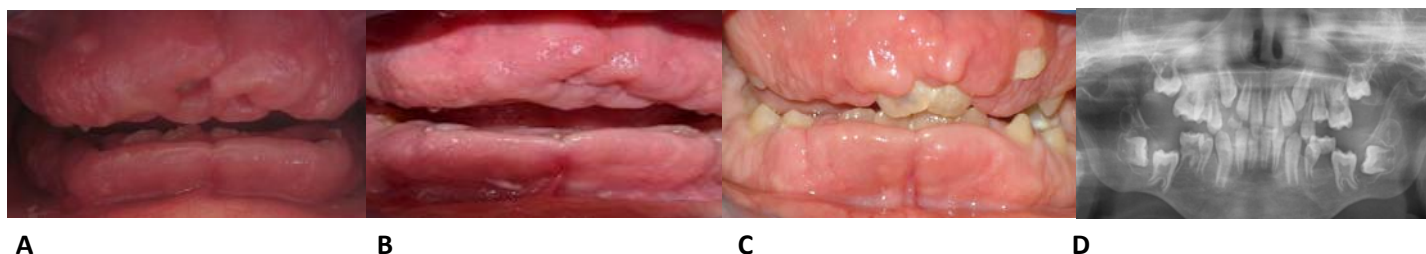
Glass ionomer capsules in the Fuji Triage and IX kits were mixed according to the manufacturer's instruction and a "pressed finger" (according to ART)<sup>6</sup> was used to spread GIC to seal the pits, fissures, and cavities. The pits and



**Figure 1.** Facial views(A) and hands - feet(B) of the patient

Intraoral finding was hyperplastic gingiva (**Figure 2**). Radiographs showed carious involvement of the pulp of tooth 54. Radix was present in 52, 62, 64, 73, 74, 83, and 84. Dentin caries were found in 55, 65, 75, and 85. Lack of congenital germs was also noted in 35 and 45. We observed rapid bone resorption at his six months checkup (**Figure 2**).

fissures were slightly overfilled. To enhance adherence and penetration of the material, a gloved finger with petroleum jelly was used to push the material into pits and fissures. The excess was removed by moving the finger sideways after a few seconds. Topical fluoride varnish was applied in 6-month intervals for prevention.



**Figure 2.** (A) Clinical appearance of the patient in 2003. (B) Clinical appearance of the patient in 2005. (C) Clinical appearance of the patient in 2009. (D) Panoramic radiograph showing absence of 35-45 and severe alveolar bone loss

The surface protection material (Fuji Triage, GC Europe NV) was placed on caries-free first permanent premolars, and restorative GIC material (Fuji IX, GC Europe NV) was used for teeth with caries (**Figure 3**).

Treatment was preventive and conservative only, and the patient lived free of acute symptoms for 6 years. Unfortunately, he died of cardiac pathology at age 13 years.



**Figure 3.** GC Fuji Triage was applied to the Second primary

## DISCUSSION

Originally considered a form of Melnick-Needles syndrome (MNS), THS is now recognized as a separate entity that, in addition to the Melnick-Needles phenotype, includes congenital heart defect and glaucoma.<sup>1,2</sup> In 1982, ter Haar et al reported on 3 patients in a family with an autosomal recessive form of MNS. This diagnosis was based on the striking resemblance of the craniofacial and skeletal characteristics seen in these 3 patients to those seen in MNS. A remarkable difference is the mode of inheritance: classical MNS has X-linked dominant

inheritance, with mortality in affected males.<sup>1,8</sup> THS occurs more frequently in males, whereas the male-to-female ratio in MNS is 1:7. Another noteworthy difference is prognosis: MNS is usually a benign condition, whereas all patients in the family described died of cardiovascular anomalies at an early age.<sup>2</sup>

Both syndromes are extremely rare—only 11 patients with THS and fewer than 60 patients with MNS have been reported. The largest series of THS includes the 3 patients defined by ter Haar et al.<sup>1-3</sup> The family history of our patient revealed the deaths of 3 siblings with similar facial appearance and symptoms, suggesting that their heart failure was most likely due to THS, but we were not able to consider genetic testing.

When caregivers and parents were questioned, it was clear that they had paid no attention to the oral hygiene of these children. Grannath and Widenheim<sup>9</sup> and Nicolai and Tesini<sup>10</sup> also have reported that caregivers are not familiar with how to maintain oral hygiene in children with special needs. Therefore, the main focus of dental care for these patients should be promotion of oral health, early diagnosis, and intervention when problems occur. To achieve this, the dental staff must have good communication skills and be able to support the families and encourage them to maintain daily oral health care. However, there is very limited, even insufficient, research on effective ways to prevent dental caries and periodontal disease in patients with disabilities.

## CONCLUSION

This is the first case in the literature of a patient with THS undergoing dental treatment, the 11th case whose cardiac pathology was corrected by Barlas and colleagues,<sup>3</sup> and the first male with THS to survive 13 years.

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