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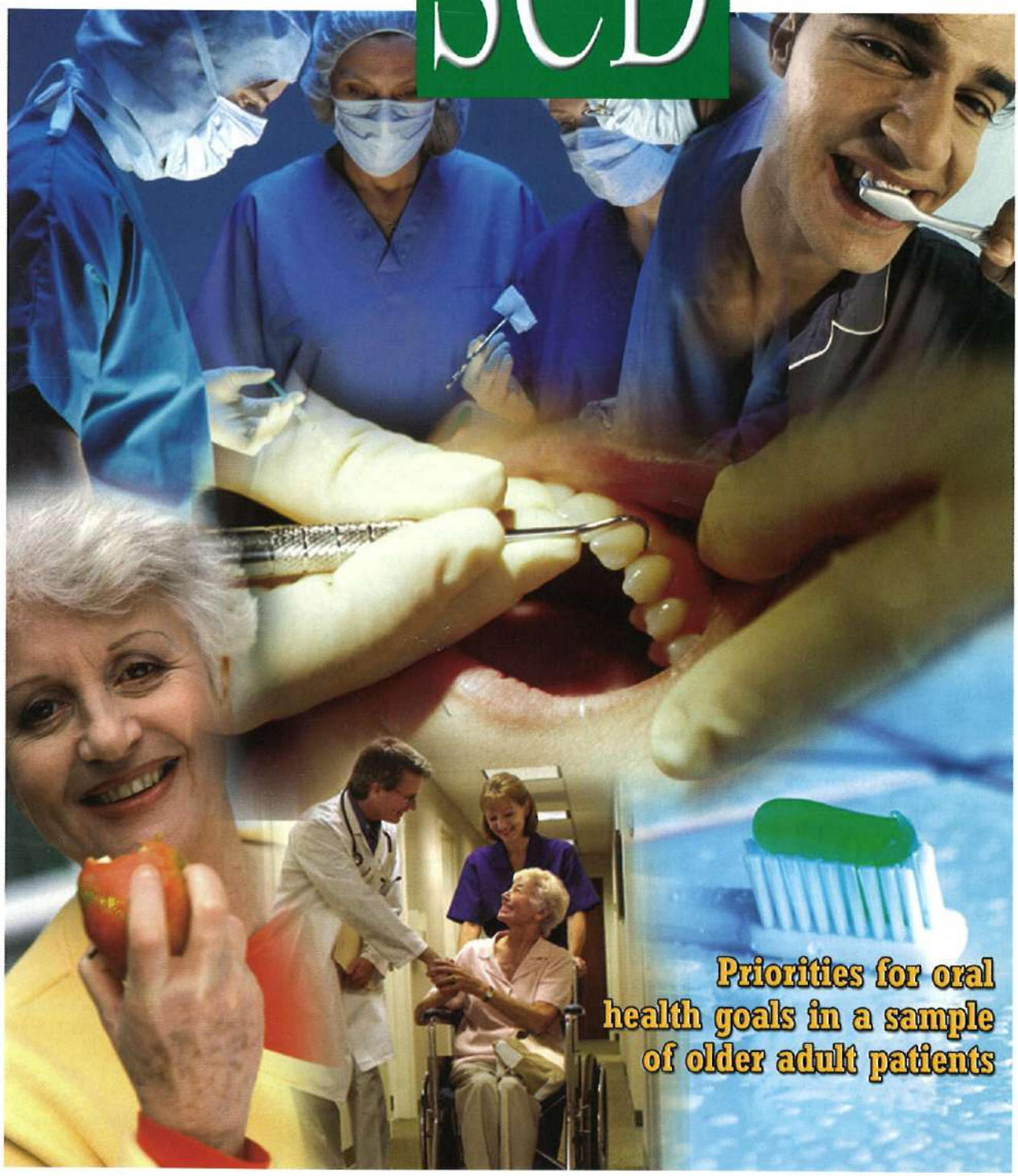

Priorities for oral health goals in a sample of older adult patients

Dental characteristics of the Wolf-Hirschhorn Syndrome: a case report

Use of cryotherapy to enhance mouth opening in patients with cerebral palsy

Treatment of muscle spasticity in patients with cerebral palsy using BTX-A: a pilot study

ARTICLE

ABSTRACT

Little attention in the dental literature has been given to the dental characteristics of patients with the Wolf-Hirschhorn Syndrome (WHS). The syndrome is caused by deletions of the terminal portion of the short arm of chromosome 4. This case report provides information on dental anomalies noted in a child with WHS. The dental findings include agenesis of multiple permanent teeth, particularly premolars and molars, taurodontism, and over-retained primary teeth. This syndrome exhibits variable clinical expressivity, possibly due to the extent and the specific locus of the chromosomal deletion. Further studies are required to obtain a clearer view of the clinical oral/dental manifestations of this syndrome.

KEY WORDS: Wolf-Hirschhorn Syndrome; chromosome 4 deletion; tooth agenesis; taurodontism; pediatric dentistry

Dental characteristics of the Wolf-Hirschhorn Syndrome: a case report

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Introduction

First described in 1965,[1,2] Wolf-Hirschhorn syndrome (WHS) is a genetic condition caused by a partial deletion of the terminal portion of the short arm of chromosome 4, in the p16 region. Clinical manifestations of the distinctive WHS phenotype include pre- or postnatal growth retardation, psychomotor delay, congenital hypotonia, seizures, and characteristic facial dysmorphism, in particular, "a Greek warrior helmet appearance" of the nose (that is, a broad bridge of the nose continuing to the forehead).

A wide range of clinical findings and manifestations occur in WHS. For example, major congenital malformations, such as cleft lip/palate, occur in 25% to 50% of patients,[3] dental abnormalities occur in 50% to 75%,[3] and the nonspecific findings of developmental disability and growth delay are consistent features in all WHS patients.[3] Within the first two years of life, the mortality rate is 21%.[4] There is some disagreement, possibly due to the limited number of patient reports, whether such a range in the severity of WHS is related to the size and the specific site of chromosomal deletion in distal 4p.[3] However, few studies using molecular cytogenetic techniques have correlated 4p16.3 microdeletions with the milder forms of WHS,[5,6] and a positive correlation between deletion size and overall risk of death has been reported.[4] Furthermore, the recent finding of two distinct critical deletion regions of chromosome band, 4p16.3 may explain the range in clinical manifestations of WHS.[7] Apparently, WHS is best described as a contiguous gene syndrome with an undefined number of genes contributing to the phenotype, rather than a syndrome resulting from deletion of a single, pleiotropic gene.[3,8]

There is little information on the oral/dental manifestations of WHS. Burgersdijk and Tan[9] reported severe hypodontia of the permanent dentition in a 14-year-old girl with WHS. The dental features of a 13-year old boy with WHS, as described by Breen,[10] also included numerous congenitally missing permanent teeth, as well as taurodontism in eight primary molars. In a review of 48 cases of patients with WHS, Battaglia and Carey[11] observed altered tooth development in more than 50% of the sample. The more common abnormalities included agenesis of mandibular lateral incisors and maxillary cuspids, delayed eruption of teeth with persistence of deciduous teeth, peg-shaped teeth, and taurodontism in the primary dentition.

Battaglia and Carey[12] recommended the need to establish guidelines for health supervision and guidance for patients who have WHS. However, the oral disorders are often overlooked or misinterpreted by non-dental investigators. This case report presents additional information on the dental manifestations of a child with WHS.

Case Report

An 8 1/2-year-old female patient sought dental treatment in April 2003 at the Long Island College Hospital, Brooklyn, New York (Figure 1). Although the child did not have a cleft lip or palate, she did show some common dysmorphic facial features characteristic of WHS, including micrognathia, mild ocular hypertelorism, and mildly protruding large eyes (proptosis). The mother reported her daughter's diagnosis of WHS, which was confirmed by fluorescent *in situ* hybridization (FISH) analysis. The patient was non-verbal and unable to cooperate during dental treatment in a routine setting. She therefore was treated in the operating room under general anesthesia. There was no complication of malignant hyperthermia during her sedation.

No caries were detected during the dental examination, but the patient did present with gingivitis and an extremely heavy accumulation of calculus covering the surfaces of her posterior teeth. Evaluation of the radiographs confirmed agenesis of some of her permanent teeth (Figure 2). The maxillary first and second premolars and the mandibular first molars (nos. 4, 5, 12, 13, 19, and 30) were absent. A panoramic radiograph may have afforded a better appreciation for the presence or absence of additional permanent teeth or tooth buds. However, this was not possible due to the lack of cooperation from the patient and necessity to treat the patient in the operating room. Over-retained primary teeth were noted in the area of the mandibular central incisors. Taurodontism was noted on the first and secondary primary molars, teeth K, S and T.

Discussion

As most health care providers have focused on the medical, rather than dental, aspects of WHS, little information is available on abnormalities involving the teeth of children with this syndrome. Although agenesis of permanent teeth appears to be common in children with WHS, the specific missing teeth differ per patient. Interestingly, deletions on chromosome 4p are correlated with tooth agenesis.[13,14] Over-retained primary teeth and taurodontism of the first and second primary molars appear to be consistent parameters of WHS.

An aspect of WHS of importance to health care providers is the risk of developing malignant hyperthermia when general anesthesia is used.[15,16] Our patient did not develop malignant hyperthermia, an occurence that has been documented

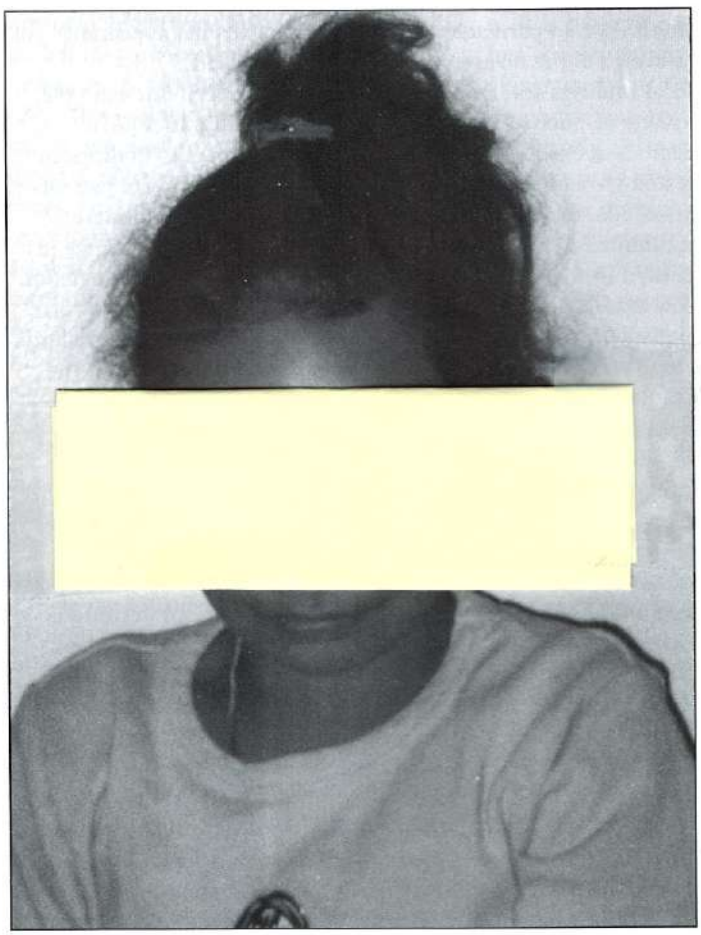

Figure 1. An 8 1/2-year-old girl with Wolf-Hirschhorn syndrome who sought dental treatment at the Long Island College Hospital, Brooklyn, New York.

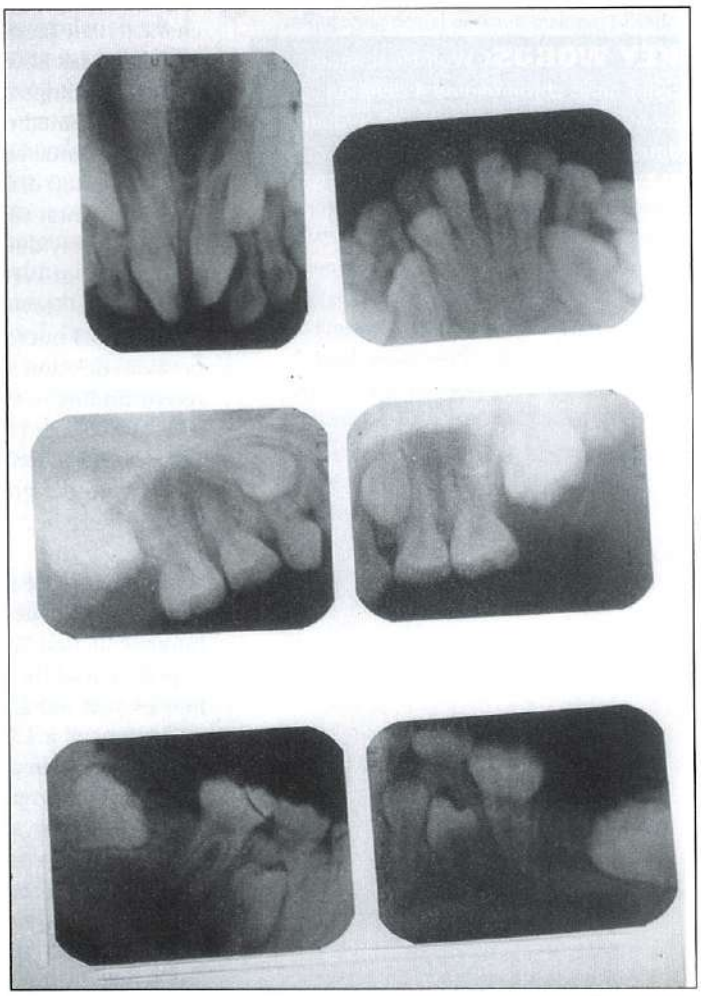

Figure 2. Dental radiographs of an 8 1/2 year-old girl with WHS.

by others.[17,18] The relationship between WHS and malignant hyperthermia requires further investigation.[17]

Summary

For the most part, research on the oral/dental aspects of WHS is extremely limited. Furthermore, patients with WHS manifest a wide spectrum of clinical symptoms, possibly due to the variable size and specific site of their chromosomal deletions. Thus, further investigations are required to attain a more complete picture of oral and dental anomalies in patients with WHS, which will ultimately lead to more effective treatment.

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