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## **Dental caries history in nine children with chromosome 18p deletion syndrome**

**Chromosome 18p deletion syndrome is caused by the deletion of a portion of genetic material on the short (p) arm of chromosome 18. Many of 100 prior case reports in the medical literature describing the dental health of subjects with this syndrome reported multiple caries associated with the syndrome. At the third annual international conference of The Chromosome 18 Registry & Research Society, dental examinations were carried out on nine children with chromosome 18p deletion syndrome and five of their unaffected siblings. The dental examination included an intra-oral evaluation of coronal decay and filled permanent teeth surfaces (DFS) and decayed and filled primary tooth surfaces (dfs) using a mouth mirror, explorer, and a high-intensity fiber optic light. An evaluation of the data revealed that five of nine children with 18p deletion syndrome (56%) were free of tooth decay or a history of tooth decay. Four of the nine (44%) had tooth decay or a history of tooth decay. The prevalence of decay was quite similar in the genetically unaffected siblings. Three of the five (60%) unaffected siblings of the children with 18p- were free of tooth decay, whereas two of the five (40%) had tooth decay. One of the affected children had a missing mandibular left central incisor. None of the children had abnormally shaped teeth. The caries pattern seems to be similar to that reported in the NHANES III data collected in the United States from 1988-1991. Analysis of these preliminary data suggests that the risk for caries in chromosome 18p deletion syndrome may be lower than previously reported.**

**C**hromosome 18p deletion syndrome (18p-) is caused by the deletion of a portion of genetic material on the short (p) arm of chromosome 18. This condition was first reported in 1963 by de Grouchy *et al.*<sup>1</sup> The female-to-male ratio for individuals with this syndrome is 2:1. Affected individuals have an average birth weight of 2600 grams (the low end of the normal range) and present with a variety of clinical findings, which can vary greatly in number and severity. Over 100 cases of this syndrome have been reported in the literature. The clinical characteristics can vary considerably among individuals. Several authors have reported multiple defects (> 10%) in persons with 18p deletion. These findings include mental retardation, short stature (< 5th percentile), abnormal external ears, cleft lip and/or palate, hypertelorism, flat nasal bridge, IgA deficiency, small mandible, epicanthic folds, short neck, microcephaly, ptosis, hypotonia, strabismus, broad trunk, and webbed neck.<sup>2</sup> Several recent case reports have also observed dystonia<sup>3,4</sup>. The dental findings in individuals with chromosome 18p deletion syndrome are reported to be: dental hypoplasia,<sup>5</sup> downturned corners of the mouth, late eruption of teeth, and irregular and often carious teeth.<sup>6-13</sup> It is unknown to what extent the chromosomal anomaly contributed to the dental caries, since dental caries prevalence was high in the years during which most of these case reports were published.

Given the large number of reports of dental caries associated with 18p-syndrome in the literature, a survey of

the dental caries was undertaken. Clinical dental examinations were offered to children of parents attending the third annual international conference of The Chromosome 18 Registry & Research Society, in Memphis, Tennessee. Some of the parents of children with chromosome 18p- expressed concerns that their children experienced more dental problems than their karyotypically normal siblings or other children. The purpose of this preliminary communication is to report data on the prevalence of dental caries (or dental caries history) in children with deletions of 18p and their unaffected siblings (normal chromosome 18).

### **Materials and methods**

Fourteen children from nine families attending the conference voluntarily participated in a dental examination in which data related to their dental and chromosome status were collected. This convenience sample of children with 18p- syndrome (n = 9) ranged in age from 3 to 9 years. The sample of unaffected siblings (n = 5) ranged in age from 6 to 13 years. All the children were non-institutionalized. Children who were unable to cooperate with the examination procedure were excluded. The dental examination included an intra-oral soft tissue examination, and an evaluation of coronal decay and filled permanent teeth surfaces (DFS), and of decayed and filled primary tooth surfaces (dfs). One dentist (CBH) carried out the examinations using a mouth mirror, #23 explorer, and a high-intensity light. A second individual (JTC)

recorded the dental findings and pertinent demographic data. All examinations were made on the same day and without the benefit of radiographs.

The 18p- diagnosis had been previously made by karyotyping. The subjects reported here are limited strictly to known 18p- deletions. Several children were examined who, in addition to 18p- deletion, had other chromosomal abnormalities, such as 18q+, 18q-, 15p+, and 8q+, but they are excluded from this report.

## Results

The DFS/dfs data from each of the subjects are shown in the Table. The dental examination revealed that five of nine children with 18p- (56%) were free of tooth decay or a history of tooth decay. Four of nine (44%) children had tooth decay or a history of tooth decay. The prevalence of decay was quite similar to that in the unaffected siblings. Three of five (60%) unaffected siblings of the children with 18p- were free of tooth decay, whereas two of the five (40%) had tooth decay. One of the affected children had a congenitally missing mandibular left central primary incisor. None of the children had

abnormally shaped teeth or cleft palate or cleft lip.

## Discussion

The prevalence of caries-free children with 18p- and their unaffected siblings compares favorably with that of children in the United States overall. In the third National Health and Nutrition Examination Survey (NHANES III),<sup>14</sup> data were collected by the National Institutes of Health from 1988 to 1991. The survey reported on weighted estimates for over 58 million children from ages 2 to 17. Of children aged 2 to 9 years, 62.1% were decay-free in their primary dentition. In children aged 5 to 17 years, 54.7% were decay-free in their permanent dentition.

Caries, or the history of caries, was clustered in 44% of the children with 18p- syndrome and 40% of their siblings. This pattern is similar to that reported in the NHANES III data, in which 80% of the decay in permanent teeth was found in 25% of children ages 5 to 17.

Since these data are from clinical examinations of a small convenience sample of individuals volunteering for a dental examination, the findings

must be considered preliminary and are not based on a randomized survey. Therefore, caution is advised in the generalization of these data.

However, analysis of the data suggests that, for nine children with chromosome 18p-, their caries pattern is similar to that in unaffected children. Since the majority of children with 18p deletion syndrome were caries-free (56%), the majority of the decay was found in a small percentage of individuals (44%). These findings are in conflict with previous case reports of a high rate of caries in children with chromosome 18p deletion syndrome. We speculate that previous case reports reflected the widespread caries present in the population during past decades and not a specific genetic influence of chromosome 18p deletion.

The majority of the individuals examined had a dentist of record and received regular dental examinations and preventive care. This care might ameliorate a genetic risk for dental caries if, in fact, chromosome 18p deletion results in increased caries risk. The interventions believed to be responsible for the decline in caries over the past several decades in the United States are appropriate for the

**Table. Dental examination data from nine children with chromosome 18p deletion and five of their unaffected siblings.**

Subject Age and Gender (yrs, M or F)	18p Deletion Subjects				Subject Age and Gender (yrs, M or F)	Unaffected Siblings			
	No. of Teeth Present		dfs/DFS			No. of Teeth Present		dfs/DFS	
	Primary Teeth	Permanent Teeth	Primary Teeth	Permanent Teeth		Primary Teeth	Permanent Teeth	Primary Teeth	Permanent Teeth
9, Fa	16	8	0	1	11, Fa	0	24	0	0
4, Mb	20	0	15	0	6, Mb	20	2	1	0
8, Fc	11	8	0	2	11.5, Mc	0	28	0	5
8.5, Md	mixed dentition		0	0	4, Fc	20	0	0	0
4, M	20	0	0	0	13, Md	0	28	0	0
3, F	16	0	0	0					
9.5, M	14	9	30	2					
8.5, M	17	7	0	0					
4.5, F	19	0	0	0					

Five subjects with 18p deletion had no sibling dental data. Abbreviation key: dfs = decayed and filled surfaces in primary dentition; DFS = decayed and filled surfaces in permanent dentition.

a, b, c, d = siblings within a family.

dental health of patients with 18p-Fluoride, sealants, oral hygiene, appropriate sucrose consumption, and regular dental care are all indicated to maximize their dental health. As with all children, the early identification of caries-susceptible children with chromosome 18p deletion is important.

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The Chromosome 18 Registry & Research Society is a non-profit educational/research organization that provides educational, referral, and family support for persons with chromosome 18 anomalies as well as research and

treatment-related services. Its address is 6302 Fox Head, San Antonio, Texas 78247. The e-mail address is: office@chromosome18.org.

The views expressed in this article are those of the authors and do not reflect the official policy of the Department of Defense or other Departments of the US Government.

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