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Occlusal morphology in Turner syndrome

Marit Midtbø* and Agnar Halse**

Departments of *Orthodontics and Facial Orthopedics and **Oral Radiology, School of Dentistry, University of Bergen, Norway

SUMMARY The prevalence of malocclusion in 32 Turner syndrome patients, age 7–16.7 years, was investigated. The sample was subdivided according to karyotype, and 72 normal girls, aged 7.1–16.1 years, served as controls. Compared with normal girls overjet did not differ significantly while overbite was significantly reduced in 45X patients. The prevalence of distal molar occlusion, anterior and lateral open bite and lateral crossbite was significantly increased. Most significant differences were found between 45X patients and controls. Mosaic and isochromosome for the long arm of X karyotypes showed the same pattern of malocclusion, but with greater variation. No significant differences were found comparing 45X patients with mosaic and isochromosome for the long arm of X karyotypes. The results indicate that patients with structural and/or numerical aberration of the X chromosome, develop a specific pattern of malocclusion with deviations in sagittal, vertical and transversal directions.

Introduction

In females with numerical and/or structural aberration of the X chromosome as in Turner syndrome, somatic development deviates from the normal pattern. The most predominant symptom is restricted growth caused by an aberrant skeletal growth pattern (Lubin *et al.*, 1990). The size and shape of several craniofacial structures are influenced. The posterior part of the cranial base is shortened and the cranial base angle flattened, resulting in a retrognathic position of the mandible. The mandible is short and broad compared with the narrowed maxillary arch (Jensen, 1974; 1985; Laine *et al.*, 1985; Laine and Alvesalo, 1986; Peltomäki *et al.*, 1989; Rongen-Westerlaken *et al.*, 1992).

Turner syndrome patients have an endocrine imbalance caused by gonadal insufficiency. Oestrogen deficiency is responsible for the absence of a pubertal growth spurt and probably also for the delayed skeletal maturity (Park *et al.*, 1983). It is assumed that the absence of gonadal activation secondarily influences growth hormone secretion which is normal in children, but lowered in prepubertal girls with the syndrome (Albertsson-Wikland and Rosberg, 1990).

The permanent dentition is characterized by a deviating pattern with respect to crown and root morphlogy, size as well as crown-root proportions (Alvesalo and Tammisalo, 1981; Townsend *et al.*, 1984; Mayhall *et al.*, 1987; Varrela *et al.*, 1988; Midtbø and Halse, 1994 a,b). Dental maturation is accelerated. Eruption problems have also been reported (Filipsson *et al.*, 1965; Midtbø and Halse, 1992).

The deviations from normal development are also reflected in an increased frequency of occlusal anomalies. The prevalence of distal molar occlusion, lateral crossbite and anterior open bite is increased (Horowitz and Morishima, 1974; Laine *et al.*, 1986; Harju *et al.*, 1989). From these earlier investigations it seems probable that patients with X chromosome monosomy are more severely affected than patients with mosaic and isochromosome karyotypes.

The present investigation compares the occlusal morphology of young Turner syndrome patients having different chromosomal constituents with normal girls to gain further knowledge of the influence of X chromosome aberration on malocclusion.

Subjects and methods

This investigation is part of a systematic study of Turner syndrome patients to evaluate growth and development before, during and after therapy with growth hormone and oestrogen.

The karyotyping was undertaken by chromosome analysis of peripheral lymphocytes. The karyotyping, the hormone therapy and the study of general parameters was performed at the Department of Pediatrics, University of Bergen.

The subjects were 32 Turner syndrome patients from different parts of Norway (Table 1). Before hormone therapy the patients were examined and five intraoral slides, hard stone casts in habitual occlusion and a panoramic roentgenogram were taken. Five of the Turner patients were undergoing or had finished treatment with fixed orthodontic appliances; two of these patients had the maxillary first premolars extracted. Congenitally missing maxillary lateral incisors were found in one patient, another had a missing mandibular first molar. In the remaining 28 patients all the permanent teeth were present. Third molars were not evaluated.

From the files of screening patients at the Department of Orthodontics, University of Bergen, 72 girls without known genetic or hormonal disorders were selected to match the Turner patients by age (Table 1). For each of these patients hard stone casts and a panoramic roentgenogram were available. Ten patients had finished or were undergoing treatment with fixed orthodontic appliances, six of them with extraction of permanent teeth. Seven other patients lacked permanent teeth. In the controls a total of 35 permanent teeth had been extracted and seven were congenitally missing.

The occlusion was assessed twice by one investigator (MM) according to the criteria of Bjørk *et al.* (1964) on the basis of the hard stone casts. The limits selected for extreme maxillary overjet and anterior deep bite were 6 and 5 mm respectively. The average of the double registrations was used in the calculations.

In the statistical analyses the Turner patients were grouped according to karyotype as: (i) 45X and (ii) isochromosome of the long arm of X and mosaics. Differences in overjet and overbite were evaluated by analysis of variance and Tukey's multiple comparison test was used for intergroup comparisons. Differences in sagittal, vertical and transversal occlusion were evaluated by the χ^2 test. The calculations were performed by a computer program (Minitab, 1991).

Table 1 Patients distributed on the basis of age and karyotype.

		Age (years)	
Karyotype	*n*	range	mean
Monosomy X			
45X	23	7.0–16.7	
Mosaics			
45X/46XX	3	12.5–15.3	
45X/46XY	1	14.7	
45X/46X,i(Xq)	1	12.8	
45X/46X,r(Xq)	1	15.8	
Isochromosomes			
46X,i(Xq)	3	8.7–12.8	
Turner	32	7.0–16.7	12.2
Controls	72	7.1–16.1	12.5

Results

The group of Turner patients presented reduced overbite as well as increased prevalence of distal molar occlusion ($P<0.01$), anterior and lateral open bite ($P < 0.001$) and lateral crossbite ($P<0.001$). A common pattern of malocclusion is illustrated in Fig. 1.

45X karyotype

Overbite was on average smaller ($P<0.01$) than in normal girls (Tables 2 and 3). Distal molar occlusion occurred in 60.9 per cent of the 45X

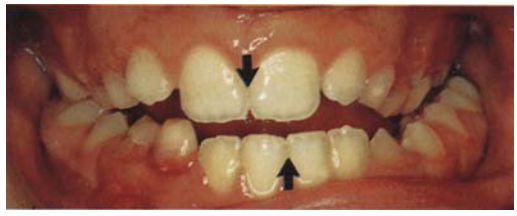

Figure 1 Patient with 45X karyotype showing lateral crossbite with midline deviation, distal molar occlusion and anterior open bite.

Table 2 Overjet (mm) for Turner patients subdivided on the basis of karyotype.

	n	Mean	SD	Min	Max	F	*P*
45X	23	4.8	2.02	1.5	10.0		
46X,i(Xq)	3	3.2	1.25	2.0	4,5		
Mosaics	6	4.4	2.05	2.0	7.0	2.06	0.132
Controls	72	4.0	1.61	1.0	8.0		

Table 3 Overbite (mm) for Turner patients subdivided on the basis of karyotype.

	n	Mean	SD	Min	Max	F	*P*
45X	23	2.0	2.08	−2.0	5.4		
46X,i(Xq)	3	0.8	2.97	−2.6	2.6		
Mosaics	6	3.6	1.25	2.0	5.5	5.94	0.004
Controls	72	3.3	1.46	0.0	8.3		

Tukey's multiple comparison test: 45X versus controls $P<0.01$.

patients which was significantly ($P<0.05$) more often than in controls (Table 4). In 21.7 per cent of the cases the distal occlusion was combined with extreme maxillary overjet. Mesial molar occlusion occurred in one patient.

Vertical anomalies were found significantly ($P<0.001$) more often in the monosomy X patients compared with controls (Table 5). Anterior open bite was registered in 17.3 per cent and lateral open bite in 8.7 per cent of the cases. No complex open bites involving both the anterior and lateral segments were found. The lateral open bites were unilateral and often associated with submerged maxillary premolars and/or first molars (Fig. 2). Only one patient had an anterior deep bite.

The prevalence of lateral crossbite was significantly ($P<0.01$) increased (Table 6). In 39.1 per cent of the patients either a unilateral or bilateral crossbite was found. None of the 45X patients had scissors bite, while this anomaly was registered in one of the control patients.

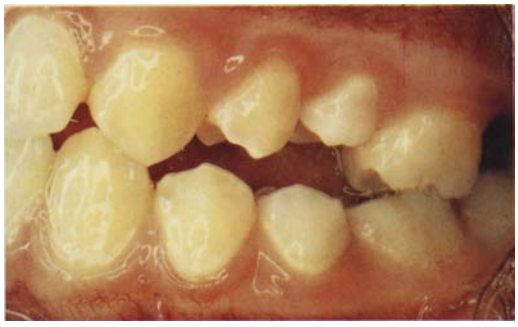

Figure 2 Lateral open bite involving maxillary premolars in a 45X karyotype.

Table 4 Sagittal occlusal anomalies in Turner patients subdivided on the basis of karyotype.

	Distal		Mesial		Extreme maxillary overjet	
	n	(%)	*n*	(%)	*n*	(%)
45X ($n=23$)	14	(60.9)	1	(4.3)	5	(21.7)
46X,i(Xq) ($n=3$)	1	(33.3)	0	(0)	0	(0)
Mosaics ($n=6$)	4	(66.7)	0	(0)	2	(33.3)
Turner ($n=32$)	19	(59.4)	1	(3.1)	7	(21.9)
Controls ($n=72$)	19	(26.4)	5	(6.9)	10	(13.9)

Turner versus controls: $\chi^2=10.42$, $P<0.01$.
45X versus controls: $\chi^2=9.16$, $P<0.05$.

Table 5 Vertical occlusal anomalies distributed on the basis of karyotype.

	Deep bite		Open bite			
	n	(%)	Anterior		Lateral	
			n	(%)	*n*	(%)
45X ($n=23$)	1	(4.3)	4	(17.3)	2	(8.7)
46X,i(Xq) ($n=3$)	0	(0)	1	(33.3)	1	(33.3)
Mosaics ($n=6$)	0	(0)	0	(0)	1	(16.7)
Turner ($n=32$)	1	(3.1)	5	(15.6)	4	(12.5)
Controls ($n=72$)	7	(9.7)	0	(0)	1	(1.4)

Lateral and anterior open bite, Turner versus controls: $\chi^2=18.81$, $P<0.001$.
Lateral and anterior open bite, 45X versus controls: $\chi^2=15.80$, $P<0.001$.
Anterior open bite, Turner versus controls: $\chi^2=12.71$ $P<0.01$.

Table 6 Transversal occlusal anomalies in Turner patients subdivided on the basis of karyotype.

	Crossbite			
	Unilateral		Bilateral	
	n	(%)	*n*	(%)
45X ($n=23$)	7	(30.4)	2	(8.7)
46X,i(Xq) ($n=3$)	0	(0)	1	(33.3)
Mosaics ($n=6$)	3	(50.0)	0	(0)
Tumer ($n=32$)	10	(31.3)	3	(9.4)
Controls ($n=72$)	5	(6.9)	1	(1.4)

Turner versus controls: $\chi^2=15.47$, $P<0.001$.
45X versus controls: $\chi^2=12.43$, $P<0.01$.
Isochromosomes and mosaics versus controls: $\chi^2=9.64$, $P<0.01$.

Mosaic and isochromosome of the long arm of X karyotypes

Similar deviations in occlusion as for the 45X patients were observed, but with greater variation.

Mean overjet was smaller than in 45X patients (Table 2). Isochromosome patients also showed greater reduction in overbite than 45X, mosaics and controls (Table 3).

The prevalence of distal molar occlusion and distal molar occlusion combined with extreme maxillary overjet was increased in mosaic patients compared with the other groups (Table 4). None of the patients with isochromosome karyotype showed extreme maxillary overjet. Mesial molar occlusion was not observed.

Anterior and lateral open bites occurred most frequently in the isochromosome karyotype (Table 5). In the mosaic group one patient had a unilateral open bite (Fig. 3), while none had an anterior open bite.

The prevalence of lateral crossbite was significantly ($P<0.01$) increased compared with controls. In mosaic patients only unilateral crossbite was observed. In isochromosome patients the situation was different; 33.3 per cent showed bilateral crossbite (Table 6).

The differences between 45X and the other karyotypes were small and none of them were statistically significant.

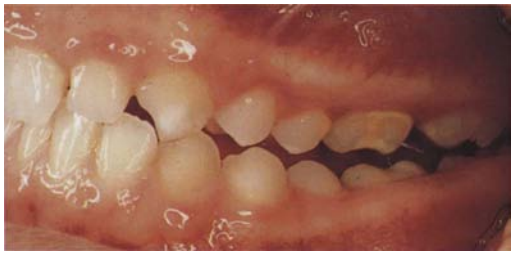

Figure 3 Lateral open bite in a patient with 45X/46XX karyotype. Both premolars and molars are involved.

Discussion

The investigation revealed increased prevalence of malocclusion in a group of young patients with Turner syndrome. The deviations comprised in the sagittal direction: increased prevalence of distal molar occlusion, in the vertical direction: reduced overbite and increased prevalence of anterior and lateral open bite, and in the transversal direction: increased prevalence of lateral crossbite. No significant differences were found between the karyotypes.

The criteria of Bjørk *et al.* (1964) for assessment of malocclusion were used in this and in two comparable Finnish studies of adult Turner patients (Laine *et al.*, 1986; Harju *et al.*, 1989). The findings are similar but the frequencies of malocclusion differ, especially for the isochromosome and mosaic patients. Small sample size as well as differences between the subjects with respect to age, chromosomal constituent and stage of dental development may have influenced the observations. The results indicate, however, that specific patterns of malocclusion are part of Turner syndrome.

When studying malocclusion in Norway, as in many other countries, it is no longer possible to determine the prevalence unaffected by previous orthodontic treatment or extractions. Early orthodontic treatment has usually eliminated or modified some malocclusions while extractions may have improved the space conditions. In this group of Turner patients 15.2 per cent of the individuals were undergoing or had finished orthodontic treatment. Large overjet and lateral crossbite were the malocclusions treated early, resulting in reduced frequencies of these traits.

Several investigations have documented an interrelationship between cranial base flexion and the number of X chromosomes. A reduced cranial base angle is associated with an increased number of X chromosomes and an increased angle with X chromosome deficiency (Jensen, 1974; 1985; Ingerslev and Kreiborg, 1978; Peltomäki *et al.*, 1989; Rongen-

Westerlaken *et al.*, 1992; Babić *et al.*, 1993; Brown *et al.*, 1993; Midtbø and Halse, 1996).

It is likely that the variations in the cranial base angle influence facial prognathism and thereby the basal sagittal jaw relationship (Jensen, 1974, 1985; Ingerslev and Kreiborg, 1978). According to a hypothesis of Gorlin *et al.* (1965) the number of X chromosomes also has an effect on mandibular growth relative to maxillary development. This has later been confirmed by investigations of occlusal anomalies in adult patients with 45X and 47XXY karyotypes. Increased frequency of distal molar occlusion and extreme overjet are observed in Turner syndrome patients (Laine *et al.*, 1986) while increased prevalence of mesial molar occlusion is found in 47XXY, Klinefelter syndrome (Alvesalo and Laine, 1992). The prevalence of distal molar occlusion is also significantly increased in young patients with Turner syndrome, as shown in this investigation.

A short posterior cranial base, a posteriorly rotated mandible and reduced posterior face height also indicate a skeletal basis for the vertical anomaly (Midtbø and Halse, 1996). Additionally increased anterior face height is a common finding in vertical anomalies but is reported to be normal or decreased in Turner patients (Jensen, 1985; Rongen-Westerlaken *et al.*, 1992; Midtbø and Halse, 1996). However, when compared to the generally reduced craniofacial dimensions the relative size of the anterior face height may be increased.

Anterior open bite has also been associated with other hereditary defects such as amelogenesis imperfecta (Rowley *et al.*, 1982), and a type with X-linked dominant mode of transmission has been distinguished from a form with similar transmission but without an open bite. Persson and Sundell (1982) suggest, in a cephalometric study of amelogenesis imperfecta patients, that the open bite is of skeletal origin. Their findings indicate further that these patients have smaller dimensions of the posterior cranial base structures, which is in accordance with findings in Turner syndrome, and further support an X-chromosomal basis for this trait.

Failure of eruption is also known to cause open bite anomalies. Eruption disturbances have been reported in the lateral segments of Turner syndrome patients (Midtbø and Halse, 1992) and may contribute to the increased prevalence of posterior open bite. Several genetic disorders involving tooth eruption anomalies have been reported by Sauk (1988) and comprised defects involving inherited amelogenesis imperfecta, syndromes with enamel involvement and growth retardation syndromes. Two of these characteristics, growth retardation and enamel involvement are also part of Turner syndrome.

A new theory on tooth eruption (Cahill *et al.*, 1988) suggests that the enamel organ of each tooth times and induces the primary activities of its dental follicle in prefunctional eruption, namely bone resorption causing an eruption pathway and bone formation that moves the tooth through the eruption pathway. Alterations in the enamel organ may thus influence the timing and induction of tooth eruption. Our findings of eruption problems in Turner syndrome may be explained by such interrelationships between enamel involvement, timing of eruption and eruption disturbances (Alvesalo and Tammisalo, 1981; Midtbø and Halse, 1992).

Endocrine disturbances have been associated with delayed or failed eruption. Hypothyroidism and hypopituitarism are the two most commonly encountered examples (Hall, 1994). An association between the growth hormone deficiency in prepubertal girls with the syndrome and eruption disturbances of posterior teeth may thus be possible.

Primary failure of eruption as a cause of posterior open bites has been discussed by Proffit and Vig (1981). They list several characteristics of patients with this disorder which correspond with our findings in Turner syndrome: (i) posterior teeth are involved more often than anterior teeth; (ii) involved teeth may erupt all the way into occlusion and then cease to erupt; (iii) deciduous as well as permanent molars are likely to be involved; (iv) the condition is rarely symmetrical and frequently unilateral.

Our findings of increased frequency of lateral crossbites are in accordance with those of Laine *et al.* (1986) and Harju *et al.* (1989) and may be caused by disharmony in width between the maxilla and the mandible (Jensen, 1985; Laine and Alvesalo, 1986; Laine *et al.*, 1985). The position of tooth buds and the path of eruption are also of significance in development of transversal anomalies (Proffit, 1986).

Transversal growth of the maxilla occurs

mainly in the midpalatinal suture. The influence of the nasal cartilage on postnatal maxillary growth is not clarified. However, it is known that the nasal cartilage is important for prenatal and early postnatal growth of the midface and for maintenance of normal midfacial form (Persson and Thilander, 1985). On the basis of deviations in most cartilage derived craniofacial structures in Turner syndrome (Rongen-Westerlaken *et al.*, 1992) alteration in the development of the nasal cartilage appears probable.

In conclusion, the present investigation supports earlier findings that patients with Turner syndrome develop specific patterns of malocclusion. Several deviations may be explained by alterations in form, shape and position of certain craniofacial structures. Local factors such as disturbance in eruption probably also contribute to an increased prevalence of malocclusion.

Address for correspondence

Marit Midtbø
Department of Orthodontics and Facial Orthopedics
School of Dentistry
University of Bergen
Årstadveien 17
N-5009 Bergen, Norway

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