Palatine ridges and tongue position in Turner syndrome subjects

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SUMMARY Females with Turner syndrome (TS), X chromosome monosomy, are characterized with palates that are narrow in width, normal in height and which are commonly associated with the presence of lateral palatine ridges. The aim of the present study was to assess the relationship of tongue position, palatal dimensions, and the presence of the lateral palatine ridges in TS subjects. The study also aimed to evaluate the maternal contribution to the palatal dimensions of their TS daughters.

The subjects were 71 TS individuals and their female family members (n = 50). Tongue position was evaluated on lateral cephalograms, the palatal height and width measurements and observation of the presence of lateral palatine ridges on dental casts. Differences in tongue position and the palatal index were assessed by an independent sample's *t*-test, and the relationship between the presence of lateral palatine ridges, tongue position, and palatal dimensions by one-way analysis of variance (ANOVA) and Bonferroni *post hoc* multiple comparison test. Partial correlation analysis was used to determine the association of palatal dimensions between TS daughters and their mothers.

The distance of the tongue from the palate was significantly longer in the TS subjects compared with the controls [10.9 mm (standard deviation, SD, 4.0) versus 7.6 mm (SD 3.4), respectively, P < 0.001] indicating a low tongue position in TS. The TS subjects with prominent lateral palatine ridges had significantly narrower posterior palates compared with the TS subjects without lateral palatine ridges [29.5 mm (SD 3.1) versus 31.5 mm (SD 2.2), respectively, P < 0.05]. There was a trend for an association between mothers and their TS daughters in palatal width measurements at the level of the upper first premolars (r = 0.78, P = 0.07).

The tongue position in TS females is low. The presence of prominent lateral palatine ridges is associated with a reduced palatal width.

Introduction

A high-arched palate with lateral palatine ridges is a common non-specific feature of several syndromes. A description of a high-arched palate is usually based on subjective judgement. When palatal dimensions of such subjects are measured, the palate is often found to be narrow, but approximately normal in height (Shapiro *et al.*, 1967; Laine *et al.*, 1985). The presence of lateral palatine ridges is thought to relate to a long-standing lack of tongue thrust into the palatal vault (Hanson *et al.*, 1976; Gorlin *et al.*, 2001).

Turner syndrome (TS) is a sex chromosome disorder which is characterized by total or partial loss of one X chromosome or chromosomal mosaicism (Gelehter *et al.*, 1998). In TS, a so-called high-arched palate is present in approximately 35 per cent of subjects (Gorlin *et al.*, 2001). The width of the palate of TS females is reported to be reduced with lateral palatine ridges commonly present (Laine *et al.*, 1985; Szilágyi *et al.*, 2000; López *et al.*, 2002). A somewhat increased frequency for a cleft palate in TS subjects has been suggested (Gorlin *et al.*, 2001; López *et al.*, 2002). The skeletal features of TS are thought to arise from the haploinsufficiency of noninactivated genes on the X chromosome and from the unspecific effects of aneuploidy on cell proliferation (Ogata and Matsuo, 1995; Gelehter *et al.*, 1998; Haverkamp *et al.*, 1999). Sex chromosomes are suggested to influence palatal width as this increases with the number of sex chromosomes (Laine *et al.*, 1985, Laine and Alvesalo, 1993a). Furthermore, the short stature homeobox-containing osteogenic gene, which is at least partly responsible for statural growth reduction in TS (Rao *et al.*, 1997; Clement-Jones *et al.*, 2000), is suggested to be involved in the development of the characteristic palatal form in TS (Ross *et al.*, 2001; Rappold *et al.*, 2002). Some researchers support the concept that the craniofacial features of TS are largely determined by a compressive effect of distended lymphatics and lymphoedema on developing skeletal tissues (Ogata *et al.*, 2002).

The palatal form of TS subjects is described as narrow, but normal in height, and lateral palatine ridges are commonly present. However, data on the rest position of the tongue and on the relationship between tongue position, palatal dimensions, and lateral palatine ridges in TS subjects are limited. Therefore, the aim of the present study was to assess whether the tongue position is abnormal in TS by comparing the tongue position between TS subjects and their female family members. The significance of the tongue position and palatal dimensions for the presence of lateral palatine ridges was assessed by comparing the tongue position and palatal dimensions in TS subjects with and without the presence of palatine ridges. The other aim of the study was to assess the maternal contribution to their TS daughters' palatal dimensions and therefore the association of palatal height and width between mothers and their TS daughters was determined.

Subjects and methods

Study subjects

The study population consisted of 71 females with TS [mean age 18.3 years (SD 7.5)], 27 of their biological mothers [mean age 37.2 years (SD 7.5)], and 23 of their sisters [mean age 17.4 years (SD 7.9)]. The karyotype of all TS subjects was 45,X, while the mothers and sisters were normal. The X chromosome monosomy of the TS subjects was established for medical reasons. All the subjects were part of the Kvantti Project of patients with sex chromosome abnormalities, the data which have been collected since the early 1970s. Approximately 35 per cent of the TS subjects had received oestrogen treatment and small dose growth hormone substitutes, the impact of which on the craniofacial growth of the TS subjects can be considered insignificant (Park *et al.*, 1983; Rongen-Westerlaken *et al.*, 1992; Hass *et al.*, 2001).

Measurements of tongue position and palatal dimensions and evaluation of the presence of lateral palatine ridges

The position of the tongue in the oral cavity was assessed on lateral cephalograms as the distance of the tongue surface from the palatal plane (T-PL). The measurements were made at the distal end of the upper first molars (Figure 1), because identification of the tongue surface is difficult in the middle and anterior area of the oral cavity. Lateral cephalograms in which the contrast of the tongue surface could not be identified were excluded, as well as those of subjects with an edentulous maxillary and/or mandibular molar area. The lateral cephalograms of 71 TS subjects, 13 mothers, and 23 sisters were included. A combined group of 36 mothers and sisters was used as the control for evaluation of tongue position in the TS subjects.

The height and width of the palate were measured at the level of the upper canines, first and second premolars, and first molars on dental casts. The palatal width was measured using a sliding calliper. The distance between the most prominent points of each tooth pair at the gingival margin was measured (Figure 2a). A palatometer manufactured at the Dental Institute, University of Oulu, was used for height measurements (Figure 3). The measurements were made perpendicular to the midline of the palate at the level of the most prominent points of each tooth pair separately on the right and left sides. The mean values of the right and left side measurements were used for further calculations (Figure 2b). The palatal index (PI) was calculated at the level of the canines, first and second premolars, and first molars using the formula $PI = h/w \times 100$, where h is the palatal height measurement and w the palatal width measurement. The PI values express the form of the palate



Figure 1 Tongue position. T-PL, the perpendicular distance of the tongue surface from the palatal plane at the distal end of the upper first molar.



Figure 2 Palatal (A) width (W) and (B) height (H) measurements. W3, palatal width at the level of the canines; W4, palatal width at the level of the first premolars; W5, palatal width at the level of the second premolars; W6, palatal width at the level of the first molars. H3, palatal height at the level of the canines; H4, palatal height at the level of the first premolars; H5, palatal height at the level of the second premolars; H6, palatal height at the level of the first molars.

as the relative palatal height (Ashley-Montagu, 1934). The palatal dimensions were obtained from 71 TS females and 26 mothers. The comparison of the PI values was made between 71 TS subjects and 26 mothers. The association of palatal dimensions between TS females and their mothers was evaluated using 26 biological 'mother–TS daughter' pairs. The cephalometric T-PL and palatal height and width dimensions were measured twice by one author (MRP), with at least a 2 week interval between the measurements. Intraclass correlation coefficients ≥ 0.9 for the measurements indicated a satisfactory level of intrainvestigator reliability.



Figure 3 Palatometer. A dental cast is placed on a movable base and positioned so that the pointer of the meter can be set at the gingival margin of the tooth for calibration to zero. The dental cast is then moved transversally so that the pointer of the meter can be set at the midline of the palate and the palatal height read on the digital display.

The presence of lateral palatine ridges was evaluated visually from dental casts. Three groups were formed: group 0 (n = 26) with no lateral palatine ridges, group 1 (n = 20) with small lateral palatine ridges, and group 2 (n = 25) with prominent lateral palatine ridges. Examples of the palatal form in each group are shown in Figure 4. Subjects with an easily recognizable palatal abnormality with remarkable crests were classified into a group of prominent lateral palatine ridges (Figure 4C,F). The group with small palatine ridges included subjects with smaller crests (Figure 4B,E). The 0-group consisted of palatal vaults without crest formation (Figure 4A,D).

Statistical analyses

The statistical significance of the differences in tongue position between TS subjects and their mothers and sisters as a control group, and differences in PI values between TS subjects and mothers were assessed by an independent sample's *t*-test. The relationship between the presence of lateral palatine ridges, tongue position, and palatal dimensions was studied using one-way ANOVA and the Bonferroni *post hoc* multiple comparison test. The association of palatal dimensions between TS daughters and their mothers was assessed with partial correlation analysis controlling for the effect of age.

Results

Tongue position in subjects with TS

The distance of the tongue from the palatal plane was significantly larger in TS subjects compared with the controls, indicating a lower tongue position in TS subjects (Table 1).



Figure 4 Lateral palatine ridges. A, dental cast demonstrating a normal palate without lateral palatine ridges, group 0. B, dental cast showing a palatal vault with small lateral palatine ridges, group 1. C, dental casts of palatal vaults with prominent lateral palatine ridges of two different degrees, group 2. D, transverse section of a dental cast at the level of the first upper molars demonstrating a palatal vault without lateral palatine ridges, group 0. E, transverse section of a dental cast at the level of upper first molars demonstrating a palatal vault with small lateral palatine ridges, group 1. F, transverse section of a dental cast at the level of the upper first molars demonstrating a palatal vault with prominent lateral palatine ridges, group 2.

Table 1 Tongue position (T-PL, the distance of the tongue surface from the palatal plane measured at the distal end of the upper first molar) in Turner syndrome (TS) and control subjects.

		TS subjects $(n = 71)$	Controls $(n = 36)$	P value
T-PL/mm 10.9 ± 4.0 7.6 ± 3.4 <0.	PL/mm	10.9 ± 4.0	7.6 ± 3.4	< 0.001

The values are means \pm SD.

PI values for TS subjects and mothers

The TS subjects had higher PI values than their mothers at the level of all measured tooth pairs (Table 2). The mothers' PI values, measured at the level of the second premolars and first molars, were within the normal limits of 39–43 per cent reported by Knott and Johnson (1970).

Significance of tongue position and palatal dimensions on the presence of lateral palatine ridges

There was a statistically significant difference (P < 0.05) in palatal width at the level of the first molars and a strong trend for a difference in palatal width at the level of the second

premolars between the TS subjects with different lateral palatine ridges (Table 3). In the Bonferroni *post hoc* analysis, TS subjects with prominent palatal ridges had significantly narrower palates compared with TS subjects with normal palates. Tongue position was not statistically significantly different between the TS subjects classified according to the manifestation of lateral palatine ridges (Table 3).

Association of palatal height and width measurements between TS females and their mothers

None of the palatal dimensions had a statistically significant association between TS females and their biological mothers; however, a strong trend (P=0.07) for an association of palatal width at the level of the first premolars between mothers and TS daughters was observed (Table 4).

Discussion

In the present TS sample, in which approximately one-third of the subjects had prominent lateral palatine ridges and another third normal palates without palatine ridges, the

Table 2 Palatal index (PI) using the formula $PI = h/w \times 100$, *h* the height and *w* the width of Turner syndrome (TS) subjects and mothers.

PI	TS (<i>n</i> = 71)	Mothers $(n = 26)$	P value
Canine	16.3 ± 8.0	10.6 ± 5.7	<0.01
First premolar	41.3 ± 10.1	32.9 ± 7.9	<0.01
Second premolar	49.6 ± 9.0	42.1 ± 8.8	<0.01
First molar	48.7 ± 8.4	40.9 ± 7.6	<0.01

The values are means \pm SD.

Table 3 Tongue position (T-PL, the distance of the tongue surface from the palatal plane at the distal end of the upper first molar) and palatal height and width in Turner syndrome (TS) subjects for the occurrence of lateral palatine ridges.

	Group 0 (<i>n</i> = 26)	Group 1 (<i>n</i> = 20)	Group 2 (<i>n</i> = 25)	P value
Tongue position				
T-PL	10.3 ± 4.6	10.4 ± 4.5	11.9 ± 4.0	0.30
Palatal height				
Canine	3.4 ± 1.5	4.1 ± 2.0	3.6 ± 1.8	0.28
First premolar	9.6 ± 1.8	10.5 ± 2.4	10.1 ± 2.2	0.34
Second premolar	14.0 ± 2.1	14.5 ± 2.2	13.8 ± 2.0	0.43
First molar	15.1 ± 2.8	15.2 ± 2.4	14.3 ± 2.6	0.59
Palatal width				
Canine	22.6 ± 2.2	22.1 ± 1.9	21.7 ± 2.2	0.44
First premolar	25.4 ± 2.0	24.5 ± 1.4	23.9 ± 3.1	0.31
Second premolar	29.7 ± 2.4	28.8 ± 1.9	27.7 ± 3.4	0.05
First molar	31.5 ± 2.2	31.0 ± 2.2	29.5 ± 3.1	0.02

Group 0, TS subjects with normal palates; group 1, TS subjects with small lateral palatine ridges; group 2, TS subjects with prominent lateral palatine ridges. The values are means \pm SD.

tongue position was found to be low. The presence of prominent lateral palatine ridges was related to the reduced posterior palatal width. There was a trend for an association in palatal width measurements in the premolar area between TS females and their mothers.

The results of the present study show that the tongue position of females with TS is lower than the tongue position of normal females. A low rest position of the tongue increases the relative pressure of the cheeks on the upper dental arch which causes narrowing. The lower dental arch, on the other hand, becomes wider because of the increased lingual pressure. Characteristic malocclusions, such as a lateral crossbite, distal molar occlusion with increased overjet and a tendency to an anterior open bite, result from this (Proffit and Fields, 2000). These malocclusions are reported to be common among females with TS (Laine et al., 1986; Midtbø and Halse, 1996; Szilágyi et al., 2000). The characteristic narrow palatal shape in TS was seen in the form of high PI values for the present TS subjects, while the PIs of the mothers were within normal values (Knott and Johnson, 1970).

Lateral palatine ridges are a non-specific feature of a variety of disorders in which they are related to the lack of tongue thrust into the palatal vault (Hanson et al., 1976; Gorlin et al., 2001). The long-lasting interference in tongue position may result from neuromotor dysfunction of the facial muscles, a functionally restricted tongue, or from the primary palatal malformation (Gorlin et al., 2001). The neuromotor status of the subjects was not evaluated in the present study. It has been reported that females with TS have good verbal skills; however, their motor skills are poor and the deficit becomes even clearer when spatial accuracy of movements are demanded (Nijhuis-van der Sanden et al., 2002; Ross et al., 2004). Therefore, the possible contribution of neuromotor deficits to the low tongue position and the lack of the moulding effect of the tongue on the palatal vault cannot be excluded. However, in the present investigation,

Table 4 The association of palatal height and width measurements between Turner syndrome (TS) subjects and their biological mothers (n = 26).

	TS daughters and mothers		
Palatal height	Canines $(n = 26)$	-0.11 (0.83)	
-	First premolars $(n = 22)$	0.56 (0.24)	
	Second premolars $(n = 19)$	-0.52(0.29)	
	First molars $(n = 21)$	-0.29(0.58)	
Palatal width	Canines $(n = 22)$	0.46 (0.36)	
	First premolars $(n = 19)$	0.78 (0.07)	
	Second premolars $(n = 14)$	0.67 (0.15)	
	First molars $(n = 15)$	-0.65 (0.16)	

The values are correlation coefficients, *P* values in parenthesis. The number in parenthesis following the name of the tooth shows the number of tooth pairs which were compared between TS daughters and their mothers.

prominent lateral palatine ridges were associated with reduced palatal width rather than with differences in tongue position.

Palatal width might be influenced by sex chromosomes, because palatal width increases with the number of sex chromosomes from 45,X to 46,XX and from 46,XY to 47,XYY (Laine *et al.*, 1985; Laine and Alvesalo, 1993a). However, 47,XXY males have narrower palates than normal 46,XY males (Laine and Alvesalo, 1993b) indicating that the role of sex chromosomes in palatal width development is not straightforward. In family studies, parent–offspring regression and correlation is an important part of the estimation of heritability (Falconer and Mackay, 1996). There are very few studies concerning the correlation of palatal dimensions between parents and their children (Bowden and Goose, 1968).

The common problem when measuring the palatal width between tooth pairs arises from the inaccuracy caused by forward drifting of posterior teeth because of malalignment and missing teeth. This made exact positioning of the reference points and the comparison of measurements between individuals difficult. In the present study, the maternal contribution to the TS daughters' palatal height and width was assessed by evaluating the association of the palatal dimensions between TS daughters and their biological mothers. Somewhat higher correlation coefficients in width measurements in the premolar area (r > 0.65) were found in the present study compared with previously published data of palatal width correlations between normal family members (r < 0.30; Bowden and Goose, 1968). Growth of the palate is reported to be more stable in the premolar than in the molar area, as the PI measured between the premolars does not increase but stays about the same from the primary through the mixed to the permanent dentition (Howell, 1981). In studies of the dental arch form in 'normal' families, some maternal effects have been reported for arch form (Hu et al., 1991) and the position of the second premolars and first molars has been suggested to have stronger genetic than environmental control compared with the position of the teeth in the anterior segments of the dental arches (Hu et al., 1992). The relative stability of height and width development of the palate and the possible maternal effects on the width of the palate might be seen in the tendency for an association of the palatal width dimensions in the premolar area between mothers and their daughters with X chromosome monosomy.

Conclusions

Tongue position in the present subjects with TS was low. The prominent lateral palatine ridges, which were present in approximately one-third of the TS subjects, were related to the narrowness of the posterior palate. There was a trend for a maternal contribution in palatal width in the premolar area between females with TS and their mothers.

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