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Dental occlusion and arch size and shape in karyotype 46,XY females

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SUMMARY The dental arch dimensions and occlusion of five Finnish individuals with complete testicular feminization were compared with their first-degree relatives and population female and male controls. The women with complete testicular feminization tended to have larger maxillary arch dimensions in all three spatial planes than the female and male controls, and larger mandibular arch dimensions in the transversal plane than the female controls.

The results also suggested that the height/width ratio in the maxillary arch and width/length ratio in the mandibular arch would be greater in these females than in population females. Both the molar and canine sagittal relationships were more mesial than in the female and male population controls.

As the phenotype in these 46,XY females is due to insensitivity to, or lack of androgens, it is suggested that the presence of the Y chromosome in these females leads to arch dimensions falling between those of normal females and males. This follows the same general dimensional pattern observed in their adult stature.

Introduction

The literature contains descriptions of phenotypically female 46,XY individuals and suggests various causes for their sex reversion. Dewhurst (1971) recognizes three groups of 46,XY females, depending on the mode of sex reversion: testicular failure, transsexualism or androgen insensitivity. The true or complete testicular feminization syndrome (TFS) or complete androgen insensitivity syndrome (CAIS) which affected eight of the individuals in the present series, involves normal androgen synthesis and Mullerian duct inhibition, but androgen action is deficient. The clinical signs are bilateral testes, female external genitalia, undergoing of puberty with breast development, blind-ended vagina and no Mullerian derivatives. The locus for the human cytosol androgen receptor is X-linked. The basic endocrine defect seems to be end-organ insensitivity to androgens, and the failure of androgen action plus testicular oestrogens could provide an explanation for the development of secondary female sex characteristics.

The stature of 46,XY androgen-insensitive females has been found to be above average for females and above that expected for their female midparental height. This supports the concept of a stature-determining role for Y-chromosomal genes independent of their critical function in testicular determination (Quigley *et al.,* 1992).

Similar results have been reported after anthropometric studies on the same group of Finnish 46,XY women with complete testicular feminization (CTF) as in this report. Most of their body dimensions were found to be larger than those of female controls, but they were generally shorter than those of male controls. In terms of head dimensions, 46,XY females show larger values for the circumference and length of the head and face than female controls (Varrela *et al.,* 1984).

Patients with incomplete testicular feminization syndrome (ITFS) are classified into two types: Type I ITFS and Type II ITFS. Swyer's syndrome is the XY form of 'pure or complete gonadal dysgenesis' or 'pure testicular dysgenesis' (Glenn, 1976). 46,XY individuals with partial gonadal dysgenesis may display asymmetric gonadal development, with a streak gonad on one side and a testis or dysgenetic testis on the other (Fechner *et al.,* 1993).

In earlier studies on oral cavities of these females (complete form of testicular feminization) the permanent teeth were found to be larger in the labio-lingual dimension than the teeth of normal females as well as of their first-grade female relatives, and seemed to be more similar in size to those of normal men (Alvesalo and Varrela, 1980). The enamel of the maxillary central permanent incisors had similar mesio-distal thickness in the CTF females as in the population control males and females, while the dentine was of approximately the same thickness in the CTF females and control males and clearly thicker than in control females (Alvesalo, 1985).

Subjects and methods

Eight Finnish 46,XY females aged 18–42 years at the time of dental examination (mean age 25.4) with complete testicular feminization were examined. The controls were eight first-degree female relatives aged 6–43 years (mean age 22.7), 45 normal females aged 9–56 years (mean age 24.6) and 46 normal males aged 9–43 years (mean age 26.3), taken from the same 'Kvantti Study' series of individuals with sex chromosomal abnormalities and normal control subjects as the 46,XY females. The statistical comparisons, however, were made between those five 46,XY females, who had their first-degree relatives examined. Two of the four individuals not diagnosed as having complete testicular feminization probably had incomplete testicular feminization and defective androgen action. The third probably had Swyer's syndrome and the fourth partial gonadal dysgenesis, with little or no androgen secretion.

The occlusal variables examined comprised a modified Angle's classification of the molar occlusion, the canine relationship, overjet and overbite and a metric analysis of the size and shape of the dental arches. The occlusion and the size of the maxilla and mandible were recorded by examining and measuring hard stone casts poured in dental alginate impressions. Angle's classification (Proffit, 1986) and Björk *et al.*'s (1964) method was used as a basis for determining the sagittal occlusion of the permanent molars on both the right and left side of the face. By combining and modifying the above methods, a classification of occlusions, to an accuracy of half a cusp, was achieved. The occlusion of the molars was classified as either neutral, distal or mesial. The sagittal relationship of the canines was recorded on both sides of the jaws in both the permanent and mixed dentitions. Horizontal overjet and vertical overbite were measured in millimetres, only for fully erupted teeth. The widths of the maxilla and mandible were measured between the first permanent molars, the first and second premolars, the permanent canines, the first and second deciduous molars and the deciduous canines, taking the extreme palatal points at the gingival level (Figure 1). The sagittal lengths were measured at the incisal point, along a line perpendicular to a plane between the mesial surfaces of the first molars, the measurement ending at this plane (Figure 2). Other sagittal lengths were measured from the incisal point along a line perpendicular to a plane between the extreme palatal points of the deciduous and permanent canines and molars and the premolars at the gingival level.

The height of the palate was measured from the extreme palatal points at the level of the deciduous and permanent molars, the deciduous and permanent canines and the premolars (Figure 3). The method used for measuring the width and length of the maxilla and mandible was a slightly modified version of that of Laine and Alvesalo (1986). Sliding digital callipers (VIS, MAUa-E, Fabrik für feinmechanische Erzeugnisse 'General Swierczewski', 01–234, Warsaw, Poland) with an accuracy of 1/100 mm and a steel pipe of 3 mm diameter with a sliding steel pin inside were used to perform the measurements. The arch dimensions of the CTF females were compared with their first-degree relatives and with the population control male and female means. The distribution of the molar and canine relationships was tested with a contingency table (Abacus Concepts, Stat View II, Macintosh). If a tooth was absent, the analysis automatically left out the corresponding variable.

A series of 70 double determinations were performed on a control population of males and

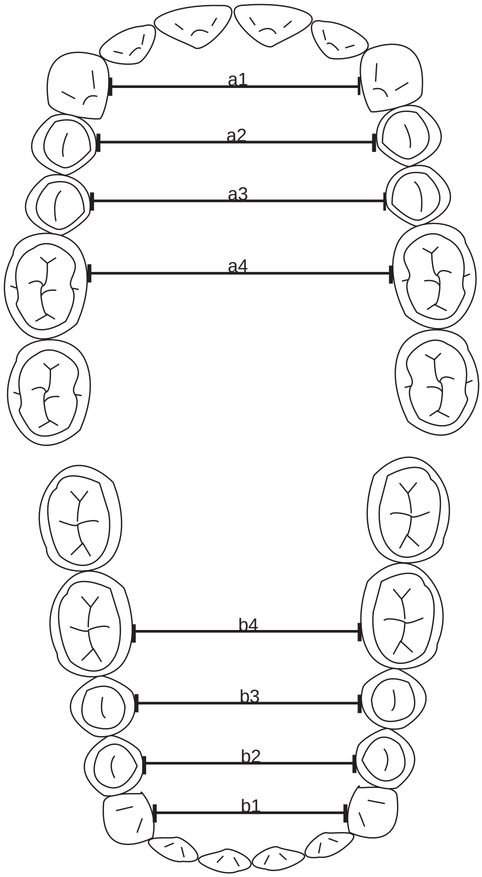

Figure 1 Arch widths. Maxillary arch width at the most palatal points between: C (a1), P1 (a2), P2 (a3) and M1 (a4). Mandibular arch width at the most lingual points between: C (b1), P1 (b2), P2 (b3) and M1 (b4).

females from another series in order to evaluate the repeatability of the occlusal recordings. The results of the comparisons of class variables were expressed as percentages of the same values in the first and second determinations, with the following results: sagittal molar relationship 95 per cent, sagittal canine relationship 83 per cent, in most cases of scissors bite, vertical open bite and crossbite 100 per cent, except for a few teeth with 97 and 99 per cent. The intra-observer method error of the linear measurements was analysed by the method suggested by Bland and Altman (1986). The standard deviation of the differences between the repeated measurements was calculated and 95 per cent of the differences were expected to lie in the limits of –2 SD to +2

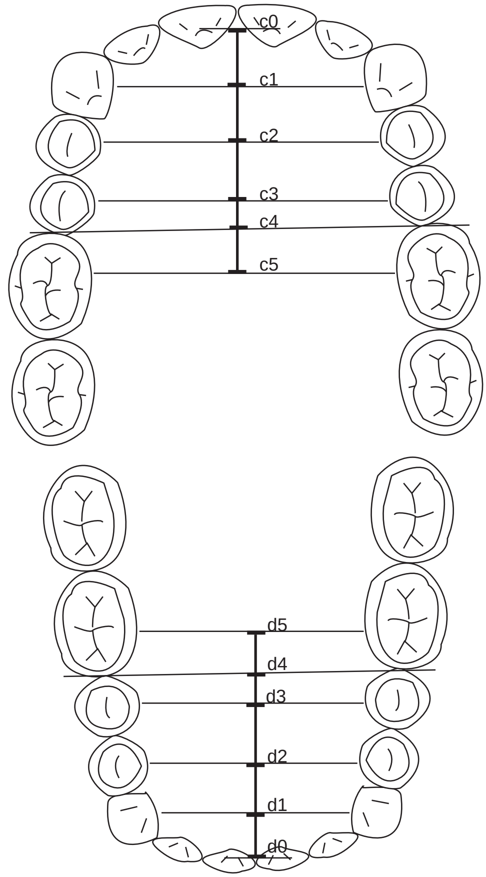

Figure 2 Arch lengths. Maxillary arch length from the incisal point (c0) to a line drawn between the most palatal points of: C (c1), P1 (c2), P2 (c3), the contact points between P2 and M1 (c4), M1 (c5). Mandibular arch length from the incisal point (d0) to a line drawn between the most lingual points of: C (d1), P1 (d2), P2 (d3), the contact points between P2 and M1 (d4), M1 (d5).

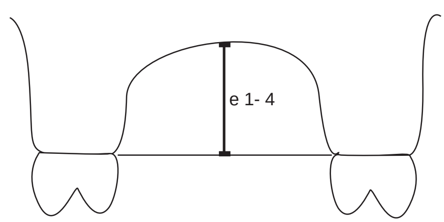

Figure 3 Palatal height. Palatal height from the palatal midline to a line drawn between the most palatal points at the gingival margins of: C (e1), P1 (e2), P2 (e3), M1 (e4).

SD. The error of measurement given in –2 SD to +2 SD ranged from 0.0 to 1.66 (mean limit 0.79), with the greatest error in the c4–c0 dimension, thus it was estimated not to be significant.

Results

The occlusal variables were compared between the 46,XY females with CTF (n = 5), who had their first-degree female relatives examined (n = 5), and the mean of population male and female controls.

Table 1 shows the ratios of every CTF individual having smaller values than the mean of population male and female controls, smaller values than her first-degree female relative and larger mean values than those of the control females.

It was observed that the women with CTF tended to have a generally broader maxillary arch, a broader mandibular arch, a higher palatal vault, a greater palatal height/maxillary width ratio, a longer maxillary arch and a greater incisal overbite than the female population control subjects (Table 1). The values of the individual CTF showed a strong tendency to exceed the mean of control males and females for the maxillary width and length and for the anterior widths of the mandible and the anterior maxillary height to width ratios. The women with CTF had a trend towards a smaller mandibular width/length ratio than the male controls, but larger than in the female controls (Table 1). The 46,XY females showed a trend towards a more mesial molar and canine relationship than the female population control subjects.

Comparison of the women with CTF with their first-degree female relatives showed basically the same trends concerning arch dimensions, arch form, and molar and canine relationships as when comparing them with the normal female controls (Table 1).

Discussion

This is to our knowledge the first report on arch dimensions, arch form and dental occlusion in 46,XY females.

The results from tooth crown studies have shown that the Y chromosome affects both dentine and enamel growth, which in the former case is probably due to proliferative activity, and in the latter to the secretory activity of the highly differentiated cells, i.e. ameloblasts (Alvesalo and Tammisalo 1981, Alvesalo, 1985, Alvesalo *et al.,* 1987, 1991). Assuming pleiotropy (i.e. a gene having more than one phenotypic effect), the larger dental arch dimensions in the 46,XY females may then be the consequence of the Y chromosome genes affecting proliferative and appositional growth in the craniofacial complex, independently of androgen action. This finding of larger size is in agreement with those of Varrela *et al.*, (1984) of larger body dimensions, head circumference, and length of head and face in women with CTF compared with normal women.

The present results are also in agreement with the findings by Laine and Alvesalo (1993), who stated that an increased number of Y chromosomes, as in the male XYY, involves transversally wider and sagittally longer maxillary arches than are found in normal men. These men also present with more mesial occlusion compared with controls (Laine *et al.,* 1992).

Alvesalo and Varrela (1980) also found the permanent teeth of 46,XY females to be similar in size to those of normal men.

The effects on the molar and canine occlusions and dental arch dimensions may be caused by increased growth in the maxillary sutures, increased intramembranous apposition, and increased growth at the condyles. It is generally believed that increased condylar cartilage proliferation might cause a mesial molar occlusion, which in turn demands dentoalveolar compensation in both the maxillary and mandibular arches, and may secondarily result in a longer maxillary arch. This feature was observed in the women with CTF. It may also be possible that the alveolar arches in the CTF females are enlarged in order to accommodate larger teeth.

Normal males and females seem to have mandibular arches of about the same length, while normal men and those with CTF have broader maxillary and mandibular arches than population control women.

Due to the small sample size and missing values, no statistically significant differences

Table 1 Arch dimensions (in mm) and dimensional ratios of the women with complete testicular feminization (CTF), their relatives and controls of both sexes

	CTF		Female relatives		Control females		Control males		CTF < $\overline{X3}$ (ratio)	CTF < female rel. (ratio)	CTF $\overline{X}$ > control females $\overline{X}$
	Mean	(*n*/SD)	Mean	(*n*/SD)	Mean	(*n*/SD)	Mean	(*n*/SD)			
Age	24.85	(5/9.68)	24.85	(5/10.60)	25.87	(43/7.98)	28.02	(41/6.66)			
Maxillary width											
a4	34.5	(2/0.71)	33.0	(4/2.71)	32.68	(31/2.55)	34.56	(27/2.62)	0/2	1/2	
a3	32.0	(4/2.0)	29.33	(3/4.04)	31.12	(26/2.57)	32.72	(29/2.4)	1/4	0/2	
a2	27.25	(4/1.71)	29.5	(4/5.45)	26.63	(35/2.34)	28.25	(32/1.97)	2/4	1/4	
a1	25.0	(4/1.83)	24.0	(4/0.82)	22.61	(38/3.89)	24.65	(37/1.44)	1/4	0/3	4/4
Mandibular width											
b4	31.0	(3/1.73)	30.5	(4/3.11)	31.16	(25/2.62)	33.25	(24/2.17)	3/3	1/3	
b3	30.25	(4/3.40)	30.0	(5/4.18)	29.61	(31/3.04)	30.59	(34/2.93)	3/4	3/4	
b2	26.0	(5/2.45)	25.6	(5/3.91)	25.0	(41/2.06)	26.68	(40/1.87)	1/5	1/5	
b1	18.8	(5/1.64)	17.2	(5/2.39)	17.84	(43/1.4)	18.63	(41/1.56)	1/5	1/5	3/4
Maxillary length											
c4–c0	25.25	(4/1.26)	24.0	(5/3.67)	23.64	(33/2.32)	23.73	(33/2.38)	0/4	1/4	
c5–c0	30.5	(2/2.12)	28.0	(4/1.63)	28.69	(29/2.09)	28.48	(27/2.29)	0/2	0/2	
c3–c0	22.75	(4/0.96)	23.0	(4/2.83)	21.69	(26/2.09)	21.48	(29/1.99)	0/4	1/3	
c2–c0	15.75	(4/0.96)	15.33	(3/0.58)	14.79	(33/1.67)	14.69	(32/1.97)	0/4	1/3	
c1–c0	9.0	(4/0.82)	9.4	(5/1.14)	8.44	(36/1.34)	8.35	(37/1.6)	1/4	2/4	5/5
Mandibular length											
d4–d0	21.0	(4/2.16)	20.6	(5/1.52)	20.74	(34/1.69)	20.18	(33/2.2)	2/4	3/4	
d5–d0	24.67	(3/2.08)	24.0	(4/0.82)	24.0	(25/1.58)	23.63	(24/2.45)	1/3	1/3	
d3–d0	17.25	(4/2.22)	17.4	(5/1.14)	17.77	(31/1.69)	17.24	(34/2.36)	2/4	1/4	
d2–d0	11.0	(5/1.87)	10.4	(5/1.52)	11.17	(41/1.36)	10.75	(40/1.86)	3/5	2/5	
d1–d0	5.2	(5/1.79)	6.0	(5/1.0)	5.84	(43/1.07)	5.73	(41/1.27)	3/5	2/5	2/5
Palatal height											
e4	16.0	(2/1.41)	14.25	(4/2.75)	14.42	(31/2.33)	16.19	(26/1.67)	1/2	0/2	
e3	14.0	(4/2.16)	12.0	(4/3.74)	12.81	(26/1.58)	13.82	(28/2.26)	2/4	0/3	
e2	7.75	(4/1.26)	6.33	(3/2.08)	7.57	(35/1.87)	6.78	(32/2.71)	1/4	1/3	
e1	1.0	(4/0.0)	0.8	(5/0.45)	0.53	(38/0.65)	0.76	(37/0.76)	4/4	0/4	4/4
Maxillary height/width											
e4/a4	0.46	(2/0.05)	0.43	(4/0.07)	0.44	(31/0.09)	0.47	(26/0.07)	1/2	0/2	
e3/a3	0.44	(4/0.1)	0.36	(3/0.09)	0.41	(26/0.07)	0.43	(28/0.09)	2/4	1/2	
e2/a2	0.29	(4/0.06)	0.24	(3/0.08)	0.29	(35/0.08)	0.24	(32/0.10)	1/4	1/3	
e1/a1	0.04	(4/0.00)	0.03	(4/0.02)	0.02	(38/0.03)	0.03	(37/0.03)	0/4	0/3	3/4
Maxillary width/length											
a4/c5–c0	1.13	(2/0.10)	1.19	(4/0.16)	1.14	(29/0.11)	1.22	(27/0.13)	1/2	1/3	
a3/c3–c0	1.41	(4/0.08)	1.36	(3/0.25)	1.45	(26/0.17)	1.54	(29/0.18)	3/4	0/2	
a2/c2–c0	1.73	(4/0.06)	1.77	(3/0.23)	1.83	(33/0.25)	1.96	(32/0.28)	4/4	2/3	
a1/c1–c0	2.79	(4/0.24)	2.69	(4/0.34)	2.75	(36/0.61)	3.08	(37/0.74)	2/4	2/3	
a3/c5–c0	1.08	(2/0.08)	1.03	(3/0.18)	1.06	(23/0.09)	1.15	(23/0.1)	1/2	0/1	
a2/c5–c0	0.94	(2/0.04)	0.97	(3/0.16)	0.92	(27/0.11)	1.0	(25/0.1)	1/2	2/2	
a1/c5–c0	0.87	(2/0.08)	0.86	(4/0.07)	0.81	(29/0.08)	0.88	(27/0.09)	1/2	1/2	4/7

Table 1 *continued*

	CTF		Female relatives		Control females		Control males		CTF < $\overline{X3}$ (ratio)	CTF < female rel. (ratio)	CTF $\overline{X}$ > control females $\overline{X}$
	Mean	(*n*/SD)	Mean	(*n*/SD)	Mean	(*n*/SD)	Mean				
Mandibular width/length											
b4/d5–d0	1.26	(3/0.07)	1.27	(4/0.16)	1.30	(25/0.12)	1.43	(24/0.20)	3/3	2/3	
b3/d3–d0	1.78	(4/0.31)	1.72	(5/0.18)	1.67	(31/0.17)	1.81	(34/0.34)	2/4	1/4	
b2/d2–d0	2.44	(5/0.55)	2.5	(5/0.46)	2.26	(41/0.26)	2.58	(40/0.64)	2/5	3/5	
b1/d1–d0	4.09	(5/1.78)	2.97	(5/0.88)	3.14	(43/0.6)	3.61	(41/2.09)	2/5	0/5	
b3/d5–d0	1.17	(3/0.09)	1.18	(4/0.01)	1.19	(22/0.10)	1.29	(24/0.21)	2/3	1/3	
b2/d5–d0	1.0	(3/0.11)	1.0	(4/0.09)	1.02	(25/0.08)	1.14	(24/0.15)	2/3	1/3	
b1/d5–d0	0.75	(3/0.11)	0.68	(4/0.04)	0.74	(25/0.06)	0.79	(24/0.1)	2/3	0/3	4/7
Overjet and overbite											
OJ	3.0	(4/1.83)	3.2	(5/1.92)	3.49	(37/1.61)	3.11	(37/1.24)	2/4	2/4	
OB	3.5	(4/2.38)	2.0	(5/1.22)	2.97	(37/1.28)	2.81	(37/1.47)	2/4	0/4	1/2

$\overline{X3}$: mean of control females and control males.
CTF < $\overline{X3}$ (ratio): how many times the value of the CTF < $\overline{X3}$/comparison.
CTF < female relatives (ratio): how many times the value of the CTF < subjects' female relative/comparison.
CTF $\overline{X}$ > control females $\overline{X}$ (ratio): in how many variables the mean of the CTFs > mean of control females/variables.

could be shown with a non-parametric test, which otherwise would have been appropriate failing to meet the requirements of normality.

Conclusions

As the phenotype in these females with complete testicular feminization is due to insensitivity to, or lack of proper androgens, it is suggested that the presence of the Y chromosome in these females leads to arch dimensions falling between those of normal females and males. This follows the same general dimensional pattern observed in their adult stature.

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