Palatal and Dental Arch Morphology in Down Syndrome

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A B S T R A C T

The analysis of palatal vault morphology and maxillary dental arch shape was carried out in the sample of 42 Down syndrome (DS) patients with trisomy 21. The data were compared to those of healthy controls from the same population matched for age and sex. Palatal morphology and upper dental arch shape were studied on hard plaster casts of the patients and controls. No sexual dimorphism in palatal and dental arch shape was observed in DS and controls. Normal palatal shape was more frequent in controls than in DS subjects (52.38% vs. 28.57%; p < 0.05). DS patients displayed significantly higher frequency of shelf-like or »stair palate« (38.1%) than controls (11.9%) (p < 0.02). The younger age group (3–14 year) showed much higher frequency of »stair palate« than controls (26.19% vs. 2.38%; c² = 9.72; p = 0.003). The older group of DS patients did not show increased frequency of such shape of the palatal vault. There was no significant difference in dental arch shape between DS patients and controls. High frequency of shelf-like palate in DS subjects is decreasing by age. The obtained results indicate that palatal vault morphology is subjected to the age related changes. These changes can be attributed to the growth of craniofacial structures and increased tonus of tongue and other orofacial muscles.

Key words: Down syndrome, palatal morphology, dental arch.

Introduction

Down syndrome (DS) or trisomy 21 is associated with specific craniofacial morphology. Medical anthropometry was successfully used in objective evaluation of craniofacial features in DS and some other craniofacial syndromes. It is generally accepted that overall size and development of the craniofacial structures are somewhat deficient in trisomy 21. The midface is particularly underdeveloped. The conclusions on palatal morphology are somewhat controversial. The palate is frequently described as high-arched with narrow palatal vault. Other findings
suggest that the palate in DS subjects is generally smaller. Objective evaluation confirmed that the palatals vault in DS is smaller than in normal subjects. The palatal dimensions in DS are shorter in depth, and lower in height. Numerous minor physical anomalies also show significant correlations with DS and different developmental disorders in children.

The palatal vault in DS in the sagittal plane was found to fit an elliptic paraboloid shape. Early hypotonia in DS and lingual diastasis were recognized as an etiological factor of specific palatal morphology with soft tissue prominence along the palatal surfaces of the maxillary dental arch. Such a type of palate has been described as shelf-like or «stair palate».

It is considered that the hypotonic tongue does not contribute to the remodeling of the palatal vault. Limbrock et al. established that 80–90% of children with DS younger than 3 years of age display lingual diastasis. They assume that the midline prominence of the tongue appears during contractions due to insufficiency of transversal fibres of the genioglossus muscle and insufficiency of the lingual fibrous septum.

Lingual diastasis contributes to the characteristic shape of the palatal vault, which is described as «stair palate». Age changes of palatal shape can lead to a higher frequency of so-called V-shaped palate in children of school age.

The purpose of the present study was to determine the prevalence of individual types of palatal vault and maxillary dental arch in Down syndrome and to compare it with findings for normal controls. The aim was also to identify characteristic changes in palatal shape and age-related changes in these findings.

**Material and methods**

The sample comprised 42 patients with cytogenetically verified diagnosis of trisomy 21 or Down syndrome (16 males and 18 females). Cytogenetic analysis in each subject revealed an extra chromosome 21 or trisomy 21. The age of patients ranged from 3 to 20 years. The control group consisted of 42 normal and healthy young individuals randomly chosen from the Croatian population and matched in age and sex to the patients in the sample (Table 1). Dental study models were obtained from all patients and healthy controls.

<table>
<thead>
<tr>
<th>Age ranges (years)</th>
<th>Down Syndrome (N = 42)</th>
<th>Controls (N = 42)</th>
</tr>
</thead>
<tbody>
<tr>
<td>3–7</td>
<td>8</td>
<td>8</td>
</tr>
<tr>
<td>8–14</td>
<td>14</td>
<td>14</td>
</tr>
<tr>
<td>15–20</td>
<td>20</td>
<td>20</td>
</tr>
<tr>
<td>Total</td>
<td>42</td>
<td>42</td>
</tr>
</tbody>
</table>

*Fig. 1. Classification of the shape of palatal vault: A – normal, B – furrowed, and C – shelf-like or «stair palate» palate.*
The palatal shape and maxillary dental arch form were analysed on the obtained study models. The shape of the palatal vault was classified as: A – normal, B – furrowed, and C – shelf-like or »stair palate« (Figure 1). Maxillary arch form was classified according to Cooper et all. into four categories: A – normal or parabolic, B – ellipsoid, C – U-shaped or hypsiloid and D – pointed or hyperbolic (Figure 2).

Statistical tests were used to examine the null hypothesis that the percentage of the specific shape of palatal vault or dental arch per group was equal for DS patients when compared with healthy controls. Differences between DS subjects and the control group were assessed using Chi-square with Yates’ correction or Fisher’s exact test. As the number of subjects was small Fisher’s exact probability test was used.

Results

No statistically significant differences were established between the evaluated traits of males and females, and both groups were evaluated as a whole. The shape of the palatal vault shows significant differences between DS patients and normal controls (Table 2). Normal palatal shape was more frequent in controls (52.38%) than in DS patients (28.57%) ($\chi^2 = 4.94; p = 0.045$). Furrowed type of palate was present in both groups with similar frequencies (33.33% in DS patients versus 35.72% in controls). Shelf-like or »stair palate« was significantly more frequent in DS patients (38.10%) than in controls (11.90%) ($\chi^2 = 7.68; p = 0.011$).

The frequency of shelf-like palate was highest in children with DS aged 3–7 years (75%). The same age controls did not show shelf-like palatal shape at all. DS

**Table 2**

Differences in the Palatal Shape Between Down Syndrome and Controls

<table>
<thead>
<tr>
<th>Palatal shape</th>
<th>Down syndrome (N = 42)</th>
<th>Controls (N = 42)</th>
<th>$\chi^2$ (Fisher)</th>
<th>df</th>
<th>p</th>
</tr>
</thead>
<tbody>
<tr>
<td>1. Normal</td>
<td>12 28.57</td>
<td>22 52.38</td>
<td>4.94</td>
<td>1</td>
<td>0.045 *</td>
</tr>
<tr>
<td>2. Furrowed</td>
<td>14 33.33</td>
<td>15 35.72</td>
<td>0.05</td>
<td>1</td>
<td>n. s.</td>
</tr>
<tr>
<td>3. Shelf-like or »stair palate«</td>
<td>16 38.10</td>
<td>5 11.90</td>
<td>7.68</td>
<td>1</td>
<td>0.011 **</td>
</tr>
</tbody>
</table>

* significantly different to control group p<0.05
** significantly different to control group p<0.02
*** significantly different to control group p<0.01
n.s. = not significant
children aged 8–14 years displayed this type of palate in 35.7% of cases, while patients aged 15–20 years had shelf-like palate in 25% (Table 3). Differences in the frequencies of shelf-like palate between DS patients and controls were highly significant in the age group 3–14 years ($\chi^2 = 9.72; p = 0.003$), but not in the older age group aged 15–20 years (Table 4).

The distribution of individual types of maxillary dental arch was similar in patients and controls (Table 5). There were no significant differences in frequencies of any of the analysed types of dental arch between the two groups.

**Discussion**

The overall skull shape depends on the harmonious development of individual craniofacial units: bones, teeth and muscle functions. Distinctive craniofacial abnormalities are quite characteristic for individuals with DS. Among them a few are common features in all individuals with this syndrome$^{1,5,8,15}$.

Characteristic craniofacial phenotype of DS individuals represents the specific pattern of maldevelopment of the underlying craniofacial skeleton. It primarily relates to the overall reduction in skull size, small midface, reduced maxilla and reduced mandible$^{19–21}$. General hypotonia is one of the characteristic features of

### Table 3

<table>
<thead>
<tr>
<th>AGE (years)</th>
<th>Down syndrome (N = 42)</th>
<th>Controls (N = 42)</th>
<th>$\chi^2$</th>
<th>df</th>
<th>p</th>
</tr>
</thead>
<tbody>
<tr>
<td>3–7</td>
<td>8 6 75.00</td>
<td>8 0 0.00</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>8–14</td>
<td>1 5 35.71</td>
<td>14 1 7.14</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>15–20</td>
<td>20 5 25.00</td>
<td>20 4 20.00</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>TOTAL</td>
<td>42 16 38.09</td>
<td>42 5 11.90</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

$N =$ number of subjects; $n =$ frequency of trait.

### Table 4

<table>
<thead>
<tr>
<th>Age (years)</th>
<th>Down syndrome (N = 42)</th>
<th>Controls (N = 42)</th>
<th>$\chi^2$ (Fisher)</th>
<th>df</th>
<th>p</th>
</tr>
</thead>
<tbody>
<tr>
<td>3–14</td>
<td>11 26.19</td>
<td>1 2.38</td>
<td>9.72</td>
<td>1</td>
<td>0.003 ***</td>
</tr>
<tr>
<td>15–20</td>
<td>5 11.90</td>
<td>4 9.52</td>
<td>0.12</td>
<td>1</td>
<td>n. s.</td>
</tr>
<tr>
<td>Total</td>
<td>16 38.09</td>
<td>5 11.90</td>
<td>7.68</td>
<td>1</td>
<td>0.011 **</td>
</tr>
</tbody>
</table>

### Table 5

<table>
<thead>
<tr>
<th>Dental arch shape</th>
<th>Down syndrome (N = 42)</th>
<th>Controls (N = 42)</th>
<th>$\chi^2$ (Yates)</th>
</tr>
</thead>
<tbody>
<tr>
<td>1. Normal (parabolic)</td>
<td>17 40.48</td>
<td>20 47.62</td>
<td>$\chi^2 = 0.969$</td>
</tr>
<tr>
<td>2. Ellipsoid</td>
<td>2 4.76</td>
<td>1 2.38</td>
<td>df = 3</td>
</tr>
<tr>
<td>3. U-shaped or hypsiloid</td>
<td>8 19.05</td>
<td>9 21.43</td>
<td>p = 0.809 n.s.</td>
</tr>
<tr>
<td>4. Pointed or hyperbolic</td>
<td>15 35.71</td>
<td>12 28.57</td>
<td></td>
</tr>
</tbody>
</table>
individuals with DS. It is particularly expressed during the early age of postnatal development. Hypotonia affects the orofacial muscles and tongue which has an important influence on the shaping of palatal morphology\(^6\),\(^{15}\),\(^{17}\).

The results obtained in the present study indicate that individuals with DS display characteristic palatal morphology that can be attributed to the specific chromosomal constitution. Normal palatal shape was significantly more frequent in healthy controls than in DS patients (52.38\% vs. 28.57\%; \(p < 0.05\)). Furrowed type of palate was present in both groups with similar frequencies (33.33\% in DS patients versus 35.72\% in controls). Shelf-like or »stair palate« was significantly more frequent in DS patients (38.1\%) than in controls (11.9\%) (\(p < 0.02\)). The frequency of shelf-like palate was the highest in the youngest age group of patients (Table 3). In age group 3–7 years 75\% of DS subjects displayed this type of palate, and none of the controls. Shelf-like palate was less frequent in age group 8–14 years (35.7\%), and was lowest in age group 15–20 years (25\%). DS patients aged 3–14 years displayed significantly higher frequency of shelf-like palate than controls (26.19\% versus 2.38\%; \(\chi^2 = 9.72; p = 0.003\)). In the older group of subjects there were no significant differences in the frequency of stair palate between the two groups.

The shape of the palate in DS has been often described as high »gothic« or »V« shaped. The present study failed to demonstrate high frequency of so called »gothic« palate with high and narrow vault. The shape of the maxillary dental arch in DS patients does not show significant differences compared to healthy controls. It seems that this feature is not considerably affected in persons with DS (Table 5).

Panchón-Ruiz et al.\(^{12}\) examined palatal vault morphology in 57 Spanish patients with DS aged 18 to 36 years. They were unable to establish sexual dimorphism in palatal dimensions in DS and normal controls. Palatal dimensions in DS subjects were significantly reduced. Specific palatal morphology was attributed to the palate in DS patients. The shape of the palatal vault in the sagittal direction in DS fits an elliptic paraboloid shape. Linear dimensions of the palate in DS and controls did not show significant sexual differences.

Characteristic craniofacial phenotype of DS individuals shows the specific pattern of maldevelopment of the underlying craniofacial skeleton. It primarily relates to overall reduction in skull size, small midface, reduced maxilla and reduced mandible\(^{19–21}\). Previous studies showed that palatal size and shape are significantly affected in DS\(^5\),\(^{10–12}\).

Richtsmeier et al.\(^{21}\) studied the mechanisms by which segmental trisomy 16 in mice disrupts craniofacial development. This type of aneuploidy represents the genetic model of DS. They established that specific craniofacial components are affected disproportionately in this animal model for DS. The face of trisomic mice is reduced in size and the entire mandible is smaller as in humans with DS. Specific skeletal malformations in the animal model corresponded to the craniofacial dysmorphogenesis in DS.

The observations on the animal experimental model for DS confirmed disproportional growth of specific craniofacial components in this type of aneuploidy, leading to the disruptive development in DS individuals. It is primarily expressed as facial size reduction and brachycephaly. For the final shaping of the skull, the function of craniofacial muscles also play an important role\(^6\),\(^{15,17,21}\).

Only extra chromosome 21 does not cause the specific type of abnormal palatal growth. Shapiro\(^{22}\) considers that it rather leads to the disruption of develop-
men-tal homeostasis and generally dis-
turbed growth in DS. The abnormalities of palatal development were also ob-
erved in individuals with an extra X and Y chromosome\textsuperscript{23,24}. Abnormalities in pal-
tal shape were established in 45,XX women and 47,XXY men. It suggests that abnormal palatal morphology is not spe-
cific for a particular chromo-
somal constitution\textsuperscript{25–28}. The theory on the increased develop-
mental instability caused by ad-
ditional chromosomal material is used to explain abnormal development in DS\textsuperscript{29}.

The atypical muscle pattern and lin-
gual hypotonia or diastasis is particularly expressed during the first three years of life. The prominence on the dorsal surfa-
ceanthe medial line of the tongue during sagittal contraction is due to insufficiency of the lingual transversal muscle fibers as well as insufficiency of the fibrous lingual structures\textsuperscript{15}. The distal surface of the tongue influences the shape of the palatal vault. In normal subjects the button of the palate is round-shaped. The lingual diastasis in DS patients produces a shelf-like palatal shape. Pronounced lingual diastasis during the first few years of life leads to the characteristic shape of the palatal vault, which is usually described as a shelf-like or »stair« palate\textsuperscript{12,15,17,30}. Limbrock et al.\textsuperscript{15} suggests that hypotonia of the tongue plays an important role in the hard palate morphology. It seems that both generally caused disturbed morpho-
genesis and highly expressed tongue hypotonia play an important role in the final shaping of the hard palate and maxillary dental arch in patients with Down's syn-
drome.

So called »stair palate« is often found in DS children during the early years of life. This term denotes a specific palatal form, which is characterised by step-like transition of the striking palatal promi-
nence\textsuperscript{15,30}. Fisher-Brandies\textsuperscript{31} describes this type of palate in DS subjects as »U« or »W« palatal form and observed it in 20% of children with DS.

The high frequency of »stair« palate in the younger group of patients in this stu-
dy gradually decreased with age. The ob-
served age-related changes in palatal morphology show a trend towards gradual changing and reshaping of the palatal vault by muscle function. Farkas et al.\textsuperscript{32} also observed significant age-related chan-
ges of some craniofacial measurements in DS patients.

Conclusions

It could be concluded that the shape of the palatal vault in DS patients shows characteristic morphology. A specific type of shelf-like palate is dominant during the early years of growth and develop-
ment. During the late period of growth significant changes in palatal morpholo-
gy can be observed. These changes can be attributed to the increased tonus and function of the tongue and orofacial mus-
culature, which leads to the reshaping of the palatal vault in DS patients.

\textbf{REFERENCES}

MORFOLOGIJA NEPCA I ZUBNOG LUKA KOD DOWNOVOG SINDROMA

S A Ž E T A K

Analiza morfologije nepčanog svoda i zubnog luka provedena je u uzorku od 42 pacijenta s Downovim sindromom (DS), kromosomske konstitucije s trisomijom 21. Podaci su uspoređeni s onima za zdravu kontrolnu skupinu iste populacije, odgovarajuće dobi i spola. Morfologija nepca i gornjeg zubnog luka proučavane su na tvrdim sadrenim modelima pacijenata i kontrolne skupine. U pacijenata s Downovim sindromom i kontrolne skupine nije utvrđen spolni dimorfizam za oblik nepca i zubnog luka. Normalan oblik nepca bio je značajno učestaliji u zdravih osoba nego u DS osoba (52.38% prema 28.57%; p < 0.05). DS pacijenti su pokazali značajno višu učestalost stepenastog nepca (38.1%) nego zdrave osobe (11.9%) (p < 0.02). Mlađa dobna skupina (3—14 godina) pokazivala je višu frekvenciju stepenastog nepca nego kontrola (26.19% prema 2.38%; χ² = 9.72; p = 0.003). Starija grupa pacijenata s DS nije pokazivala povećanu učestalost toga tipa nepčanog svoda. Nisu nađene značajne razlike u zubnom luku između pacijenata s DS i kontrolne skupine. Visoka učestalost stepenastog nepca kod pacijenata s DS smanjivala se s porastom dobi. Dobiveni rezultati ukazuju da je morfologija nepčanog svoda podložna promjenama vezanim za dob. Te se promjene mogu pripisati rastu kraniofacijalnih struktura i povećanom tonusu jezika i ostalih orofacijalnih mišića.