Voice quality in Down syndrome children treated with rapid maxillary expansion

*cmoura@med.up.pt

Abstract

Down syndrome (DS) is the most common aneuploid chromosomal disorder at birth. Phenotypic characteristics include mental retardation, general hypotonia, pharyngeal hypoplasia, frequently constricted maxillary arch with macroglossia and upper airway obstruction. Generally they have marked language difficulties and a characteristic voice. This prospective study assesses the effects of rapid maxillary expansion (RME) on voice quality in DS children. Acoustic (Praat®4.1 software) and perceptual assessment of the voice were performed in 24 children with DS, aged 4 to 12 years, who had been randomly allocated to RME and non-expanded (NE) groups. Two main evaluations were made; one prior to expansion and the second after two to four weeks of active RME plus a 5 month period of retention. Data between the two groups were compared. Perceptual analysis did not demonstrate any significant modification. RME produced significant changes in F1 frequency of vowel /a/ of DS children’s voices, probably related to alteration of vertical tongue adjustment. It also showed a tendency to decrease F0 dispersion in all studied vowels, illustrating increased stability in F0 production. These results are related to the correction of some DS typical mid-face skeletal deformities such as maxillary bone widening with improvement of upper airway airflow and increased space for the tongue in the oral cavity. The acoustic analysis is a simple, rapid and non-invasive technique which is well accepted by the paediatric population; features that are relevant to its application especially in children with mental retardation.

1. Introduction

Down syndrome (DS) is the most common aneuploid chromosomal disorder, with a prevalence of 1/770 [1]. Phenotypic characteristics include mental retardation, general hypotonia, maxillary hypoplasia and enlarged tongue, narrowing of the pharynx and short neck [1]. These factors may result in specific voice characteristics with particular acoustic alterations. The majority of DS individuals have some difficulties with oral communication, and their vocal quality has been described as hoarse, breathy, rough and nasal; the possible anatomical and physiological bases for such a description remain ill defined [2].

Rapid maxillary expansion (RME) is an orthodontic procedure used to correct narrow transverse maxillary diameter, and leads to a widening of the arch perimeter providing more space for alignment of crowded teeth, with a further benefit of enlargement the apical bone and this includes the nasal width [3]. RME improves nasal patency and concomitantly confers more space for the tongue in the oral cavity [3,4]. Because of the close relationship between the structures and the function they perform, several functions related to the oro-facial muscles may also improve. Assessment of paediatric voice quality has proven to be problematic [5]. Fiberoptic endoscopy is often difficult and rushed in the uncooperative child, and stroboscopic examination is technically challenging in any young patient. Computer-assisted voice analysis provides objective acoustic measurements and it is well tolerated by children [5]. Acoustic characteristics are different over the world, depending on the language used. Perceptual evaluation of the voice is essentially based on subjective assessment by the examiner, according to international standard protocols, and should be considered as a complementary evaluation [6].

The main objective of this study was to assess the evolution of vocal quality, including objective acoustic measures and perceptual evaluation, in a group of DS children submitted to RME compared with an age matched control group.

To our knowledge this is the first such study performed in DS children

2. Patients and Methods

Recruitment of DS children was done by mail to the main organizations working with this group in Portugal. Two centres for clinical observation were prepared, one in the south (Lisboa) and the other in the north (Porto). A paediatric-dentist and an otolaryngologist examined each of the 106 children included in this study. The inclusion criteria were: 1) cytogenetic diagnosis of 21 trisomy, 2) age between 4 and 12 years, 3) persistent nasal obstruction and/or repeated upper respiratory infections, 4) presence of lateral cross bite and/or signs of maxillary compression, 5) adequate cooperation, 6) availability to frequent follow-up journeys for treatment and monitoring and 7) informed agreement from their legal representatives. Approval was obtained from the research ethics committees of all the institutions involved. Of the 106 children included in this study and then divided into sub-groups according to age ([4; 7]; [7; 10] and [10; 13]); children from each of these sub-groups were randomly placed into two groups (fig.1): RME (or expanded) and no specific treatment group (non-expanded (NE)), using a random digits table. None of these children were submitted to any other otolaryngologic or orthodontic surgery during the study period. All the DS children included in this study resided at home, were integrated in general schools and showed no evidence of significant hearing loss. To obtain voice samples children were instructed to vocalize and sustain each of the five main Portuguese vowels (/a/,[e],[i],[o],[u]), for at least 5 seconds, in a flat tone, at their most comfortable pitch and at a constant intensity. Each vowel was produced at least 5 times. They also named a group of figures, presented on cards, applying the main phonetic sounds in Portuguese. Recordings were made in a quiet room with a high quality microphone (Sony®-V420) using a constant mouth to microphone distance of 10 cm, and a 45° of axis positioning. Voice input was recorded using a digital audiotape recorder (Sony®TCD-D3 DAT), sampled at 44.100 Hz, with 16-bit resolution and stored on an IBM-compatible computer disk [6]. The middle stable portion of the sample was then extracted [7].
The intra-oral device was applied to the 13 children in the RME group (fig. 1).

Figure 1: Study profile. DS - Down syndrome, RME - Rapid maxillary expansion group, NE - Non-expanded group, T0 - time immediately before the intra-oral device application, End of AE - end of active expansion (approximately two to four weeks after T0), T1 - time after the consolidation period, almost 6 months after T0.

The other children with DS were assigned to the NE group. RME was accomplished with an appliance designed for each individual, usually fixed with orthodontic banding to the posterior teeth. The two maxillary bones were separated at the midline suture by mean of a screw mechanism located in the midline of the appliance (fig. 2). Activation rates of the order of 0.3 to 0.5 mm per day permitted painless separation at the midline of the appliance (fig. 2). Activation rates of the order of 0.3 to 0.5 mm per day permitted painless separation at the midline of the appliance (fig. 2). The retention period consisted of two to four weeks of active expansion and 5 months of retention; the appliance was then removed (T1). The retention period consisted of the maintenance in situ of the intra-oral appliance after maximum expansion. Registers of sampled vowels were analysed using Praat®-doing phonetic by computer, version 4.0.47. To avoid as far as possible any interference with the control of speech prosody and articulation, acoustic analysis of voice relied on a sustained vowel task. The parameters calculated by Praat® displayed numerically and graphically were classified into 1 of 7 groups of measurements: 1) fundamental frequency (F0): average F0, median F0, minimum F0, maximum F0; 2) intensity (I): average I, Maximum I; 3) frequency perturbation: absolute jitter (Jitta), jitter % (Jitt), relative average perturbation jitter (jitter rap), pitch period perturbation quotient (jitter ppq5); 4) amplitude perturbation: shimmer dB, Shimmer %, amplitude perturbation quotient (shimmer apq5); 5) noise evaluation: harmonic to noise ratio (HNR), spectral tilt (energy); 7) formants (F) and bandwidths (Bw), 1 to 3. For formants analysis, Praat® applies a Gaussian-like window and computes the LPC coefficients with the algorithm by Burg for each analysis window [8]. The formant frequency values from the computer analyses were visually matched to those of the spectrogram. Because the various pitch and amplitude perturbation measures exhibit high correlation among each other, only some of the jitter and shimmer parameters were considered for further analysis. The perceptual analysis was done by a panel formed for two expert speech therapists, using a modified 4-point scale of the GRBAS scale proposed by Hirano. 1989. The scale adapted to Portuguese as the RASAT scale (Rouquidão (grade), Aspereza (roughness), Soprosidade (breathy), Astenia (asthenic), Tensão (strained)), was used [9].

Figure 2. RME device: a - before expansion, b – at maximum expansion.

The scale varies from 0, normal, to 3, severe. Nasal resonance was also assessed. For the sake of auditory-perceptual analysis, the same acoustic samples were blindly randomised. Measurements in RME and control groups were obtained before the intra-oral application of the device (T0) and after the treatment period (T1). At each time several measurements were recorded. Four children, three from the control group and one from the experimental group dropped out before the trial was finished (fig. 1). Descriptive statistics were obtained for data at T0 and T1 for both the RME and control groups. Within each of the treatment groups, for the different variables under study, the effect of time was analysed using the non-parametric Wilcoxon test for pairwise comparisons. Evolution of different variables from T0 to T1 was computed and differences between treatment groups were analysed using the nonparametric Mann-Whitney test for unpaired comparisons. For overall comparison of both groups, an ANOVA with repeated measures at two factors, vowel and time, was used for the different variables under study. All statistical analysis was performed using SPSS® version 12.0 for Windows.

3. Results:

The voice profiles of RME DS children were compared with the data of the NE group. The independent variables of height, weight, and body mass index obtained a low correlation with any of the acoustic variables. Comparing the different variables under study between T0 and T1 no significant differences between RME and NE group were found, except for the F1 frequency of the vowel /a/ that raised significantly (p<0.01) in the RME group (Tab. 1). This difference is maintained when comparing the evolution (T1-T0) of F1 frequency between the two groups. Evaluating the relationship between F1 and F2 frequencies of the vowel /a/ before (T0) and after RME (T1) among the two groups, the RME group tends to approach to values of the general population contrary to the NE group [10, 11] (Fig. 3). The full data are available at http://lfp.esi.fe.up.pt/publicacoes.html.en#artigos. The effect of vowel and time identity were entered into the overall repeated measures ANOVA and the relationships between vowels /a/ vs. /u/ and /a/ (to evaluate the effects of tongue height) and /a/ vs. /u/ (to assess the positioning of the tongue body along the horizontal plane) were also assessed, showing that the two groups were not statistically different. Regarding speech therapy assessed parameters, both groups exhibited the
same grade of differences between T0 and T1 in all items and the
analysis of evolution (T1-T0) between the two groups was
also not significant.

Table 1. Acoustic analysis datar mean and standard deviation
(SD) of evaluations at each of the experimental stages.

<table>
<thead>
<tr>
<th>Acoustic parameter</th>
<th>Vowel</th>
<th>RME</th>
<th>T0</th>
<th>T1</th>
<th>T0-&gt;T1 (RME)</th>
<th>T0-&gt;T1 (NE)</th>
</tr>
</thead>
<tbody>
<tr>
<td>F0 (Hz)</td>
<td>a</td>
<td>223.9</td>
<td>± 45.7</td>
<td>217.7</td>
<td>253.2</td>
<td>223.8</td>
</tr>
<tr>
<td>F0 (Hz)</td>
<td>e</td>
<td>793.7</td>
<td>± 47.7</td>
<td>253.2</td>
<td>233.7</td>
<td>233.7</td>
</tr>
<tr>
<td>F0 (Hz)</td>
<td>i</td>
<td>502.0</td>
<td>± 37</td>
<td>253.2</td>
<td>223.8</td>
<td>223.8</td>
</tr>
<tr>
<td>F0 (Hz)</td>
<td>o</td>
<td>± 49.8</td>
<td>± 44.9</td>
<td>223.8</td>
<td>223.8</td>
<td>223.8</td>
</tr>
<tr>
<td>F0 (Hz)</td>
<td>u</td>
<td>± 48.1</td>
<td>± 48.1</td>
<td>223.8</td>
<td>223.8</td>
<td>223.8</td>
</tr>
<tr>
<td>Formants F1 (Hz)</td>
<td>F1 (Hz)</td>
<td>± 45.7</td>
<td>± 45.7</td>
<td>217.7</td>
<td>253.2</td>
<td>223.8</td>
</tr>
<tr>
<td>Formants F2 (Hz)</td>
<td>F2 (Hz)</td>
<td>± 47.7</td>
<td>± 47.7</td>
<td>253.2</td>
<td>233.7</td>
<td>233.7</td>
</tr>
<tr>
<td>Formants F3 (Hz)</td>
<td>F3 (Hz)</td>
<td>± 37</td>
<td>± 44.9</td>
<td>223.8</td>
<td>223.8</td>
<td>223.8</td>
</tr>
<tr>
<td>Formants F4 (Hz)</td>
<td>F4 (Hz)</td>
<td>± 48.1</td>
<td>± 48.1</td>
<td>223.8</td>
<td>223.8</td>
<td>223.8</td>
</tr>
</tbody>
</table>

4. Discussion

The voice quality of DS individuals has been described as
husky and monotonous, raucous and low-pitched and in
comparison with the general population DS children exhibit
more breathiness, roughness and nasality [2, 12]. The present
study was designed to gain a more complete understanding of
both the acoustic bases and the underlying anatomical and
physiological bases of the differences that set many of DS
children’s voices apart from nonretarded counterparts. DS
children constitute a difficult group, particularly regarding
good collaboration.

![Figure 3. Comparison of sustained [a] vowel characteristics before (T0) and after rapid maxillary expansion (T1), in 2
groups of children with DS (RME-group with intervention; NE-group without intervention). Ref 1: Portuguese vowel /a/
formant frequency (F1 and F2) [10]; Ref 2: Brazilian Portuguese vowel /a/ formant frequency (F1 and F2) [11].

It should be noted when interpreting the results of this study that the sample size is small because of the restrictive
selection criteria and the difficulty of continuing in a
prospective clinical investigation over a long period. Furthermore, mean results showed a large individual
variability.

The greatest movement from RME is inferior and anterior; the
separated palatine bones widen the maxilla and there is often
some slaying of the pterygoid process of the sphenoid bone;
the lateral walls of the nasal airflow incline outwards, taking
with them the inferior turbinate, and enlarging the airway [4,
13]. All these skeletal changes are correlated to an
enlargement of the vocal tract, mainly noted at the nasal and
oral cavity, with, possibly, better accommodation of the
characteristically large tongue found in DS.

Those variations in shape and dimension of the vocal tract
should cause modifications of the acoustic and perceptual
assessments, mainly at the formant frequencies [14]. In this
study children from the RME group usually showed increased
formant frequency from T0 to T1 (Tab.1). The degree of
vowel opening associated with the lowering of the mandible
and of the tongue has a direct relation to F1 frequency, which
increases with the opening of the mouth [15]. This effect may
explain the significant increase in the F1 frequency of the
vowel /a/ in the RME group, when comparing T0 to T1,
presumably because the increased space in the oral cavity
allowed a better vertical tongue adjustment. This was not
apparent in the NE group. It should be emphasized that this
difference persisted when comparing the evolution (T1-T0) of
F1 frequency of the vowel /a/ in the RME group, when comparing T0 to T1, presumably because the increased space in the oral cavity
allowed a better vertical tongue adjustment. This was not
apparent in the NE group. It should be emphasized that this
difference persisted when comparing the evolution (T1-T0) of
F1 frequency of the vowel /a/ between the two groups.

However, there were no significant differences regarding F2
frequency and the corresponding horizontal movement of the
tongue in the RME group, nor when comparing the evolution
(T1-T0) of F2 frequency between the two groups.

Nevertheless, F2 frequency of the vowel /a/ was raised in a
larger proportion from T0 to T1 in the RME group than in the

---

**Note:** The text contains tables, figures, and statistical data. The tables and figures are not included in the natural text representation, as indicated by the placeholders and the nature of the content. The natural text focuses on the discussion of the results and the implications of the study, as well as the methodological considerations.
NE, probably due to a higher degree of freedom of the pharynx since it is slightly augmented by the horizontal dislocation of the tongue [15]. The conjunction of F1 and F2 frequencies evolution of vowel /a/ is well expressed in Fig.3, with the RME group demonstrating a tendency to approach to values of general population [10, 11]. On the other hand, F3 showed a general tendency to rise in the RME group, with the exception of the vowel /u/ that showed a slight reduction, (opposing the aleatory modification of the NE group).

It was expected that the relationship between the height (F1) and the place (F2) of vowels, studied using /a/ vs. /i/ and /u/ and /l/ vs. /u/ would reveal any differences between the evolutions of the two studied groups. But physiological adjustments considered for formant production are not simple and direct; they are the result of interaction of several complex mechanisms. It can also be argued that even the DS children have the psycholinguistic ability to adapt to slow and progressive changes, such as those occurring during RME.

Concerning F0 frequency, the NE group showed a reduction in all vowels from T0 to T1; the RME children achieved reductions, on vowels /a/ and /l/ and /u/, and almost no modifications (negligible rises) on vowels /e/ and /o/ (Tab.1). Vocal disorders with neurological bases typically have a large variability of F0 frequency [12].

However, the dispersion of F0 decreased in all studied vowels between different observation times, only in the RME group illustrating a gain of stability in F0 production (Tab.1). Possibly this is related to the larynx connection to the tongue through the hyoid bone [16]. An improved anatomical and physiological relation between the structures may adjust neuromuscular and aerodynamic control. Nevertheless this result was not expressed in jitter values which did not demonstrate any tendency to improve, in both groups.

In relation to shimmer measures the NE group showed a slight propensity to improve, which was not observed in the RME group. However the data showed no significant changes.

The noise evaluation measures did not demonstrate any significant tendency toward change in either group. Regarding the assessment by the speech therapists, change in the articulation pattern and general voice quality should be expected in both groups as a function of maturation. The evolution of both groups resembled. Probably this is because the perceptual assessment is less reliable and objective than the acoustic analysis [6].

The long term effects of RME on voice characteristics of DS children remains to be confirmed.

In conclusion, RME provides significant changes in the F1 frequency of the voice of Down syndrome children, probably related to changes of vertical tongue adjustment. It also shows a tendency to decreases the dispersion of F0 in all studied vowels, illustrating a gain of stability in F0 production. The computer acoustic analysis, using Praat® software, is an objective system with reproducible results. It is a simple, rapid and non-invasive technique which is well accepted by children as young as 4 years. These attractive features are relevant to its application in paediatric populations, especially those with mental retardation.

5. References


6. Acknowledgements:

We would like to express our gratitude to David Stevenson for his valuable assistance in the preparation of the manuscript.