Craniofacial characteristics in Saudi Down’s syndrome

Reema Al-Shawaf, Wafa Al-Faleh *

Department of Oral Medicine and Diagnostic Sciences, College of Dentistry, King Saud University, Saudi Arabia

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Abstract

Down’s syndrome is a genetic disorder that results from a chromosomal abnormality due to trisomy of all or a large part of chromosome 21. In Saudi Arabia the incidence has been reported as 1 in 554 live births, therefore, the aim of this study was to study the craniofacial characteristics in the Saudi Down’s syndrome patients in comparison with those in normal subjects. Clinical and radiographic manifestations were studied in 30 Down’s syndrome patients and 30 control subjects. The prevalence was determined and compared to the findings of healthy controls using the appropriate statistical descriptive analysis. Among the Down’s syndrome group several craniofacial manifestations were observed including brachycephaly, depressed nose, slanting of the eyes, epicanthic folds and strabismus. Oral manifestations included incompetent lips, macroglossia, fissured tongue and a high arched narrow palate. Thinning of the cranial vault and delayed closure of the sutures were observed radiographically in 16 (59.3%) and 10 (37%), respectively. Absent frontal air sinuses were found in 86.2%. The findings of this study showed a significantly high prevalence of facial and cranial characteristics among Saudi Down’s syndrome patients when compared to normal controls of the same population.

1. Introduction

Down’s syndrome is a genetic disorder that results from a chromosomal abnormality due to trisomy of all or a large part of chromosome 21. It is the most recognized congenital, autosomal anomaly associated with delayed physical and mental development (Regezi and Sciubba, 1999).

Down’s syndrome patients present with numerous characteristic physical findings (Desai, 1997). Among the most characteristic findings, are the craniofacial manifestations. Aberrations in both craniofacial structures have been identified (Regezi and Sciubba, 1999; Shafer et al., 1983).

The facial manifestations reported in Down’s syndrome include, depressed nasal bridge, slanting eyes with epicanthic folds, ocular hypotelorism and strabismus. Oral aberrations include incompetent lips, macroglossia, fissured tongue and a narrow high arched palate (Desai, 1997; Gorlin et al., 2001; Regezi and Sciubba, 1999).
Craniofacial radiographic findings in Down’s syndrome include brachycephaly, thinning of the calvarium, defective ossification along the sutures, delayed closure of sutures, absent or poorly developed air sinuses, decreased intraorbital distance and small rudimentary nasal bones (Gorlin et al., 2001; Newton and Potts, 1971; Regezi and Sciubba, 1999; Spitzer and Robinson, 1955; Taybi and Lachman, 1996). These classically described features vary significantly between individuals despite the widely recognized typical characteristics of Down’s syndrome (Levinson et al., 1955).

In Saudi Arabia, there is a relatively high incidence of Down’s syndrome. The incidence has been reported as 1 in 554 live births (Niazi et al., 1995). Therefore, the aim of this study is to study the craniofacial manifestations in the Saudi Down’s syndrome patients in comparison with those in normal subjects.

2. Subjects and methods

The study sample consists of two groups, the first group comprised thirty Saudi Down’s syndrome patients of both genders at an age range from 12 to 24 years. Different hospitals, Down’s syndrome care centers and schools in Riyadh city were contacted to collect the sample. Families were contacted and the study was explained to them. Those who agreed to participate in the study were included. All of the patients are proven to be affected by the genetic abnormality based on a chromosomal analysis by a Karyotype test. This was found in the medical files or school files of each individual.

The control group included thirty Saudi normal subjects of both genders with matching age ranges. The controls were selected by explaining the study to the mothers in the King Khalid University Hospital waiting area. Those who agreed to participate in the study were contacted and included as controls, in order to compare the rate of prevalence of similar clinical and radiographic craniofacial aberrations between the two groups. The study protocol was reviewed and approved by the Ethical Committee and Research Center at the College of Dentistry.

Both groups were seen in the dental clinics at the College of Dentistry, King Saud University. All of the patients were subjected to a detailed case history, thorough clinical examination was carried out by the first authors. Panoramic, lateral cephalometric and posteroanterior radiograph were taken unless recent radiographs were available. The radiographic interpretation was carried out by the two authors. Consents were obtained from patient’s guardian for each patient prior to the examination procedures.

2.1. Facial manifestations

Examinations of the head, nose, eyes, mouth, tongue and palate were performed and recorded either as apparently normal or abnormal as observed from the clinical examination. Facial and intraoral photographs were obtained when possible. Abnormalities have been recorded in a specially prepared examination form.

Facial characteristics

Head: Brachycephalic: Established clinically as reduced anteroposterior dimension with increased skull width with flattening of the occipit.

Nose: Flat or depressed nasal bridge: Established clinically when the upper, bony part of the nose is lower than average. The nose appears flat, broad and short. The nasal tip tends to be rounded and poorly defined. There tends to be less projection than desired.

Eyes:

- Slanting: Established clinically when an imaginary line drawn from the inner corner to the outer corner is slanted upwards.
- Presence of epicanthic fold: Established clinically when a skin fold of the upper eyelid (from the nose to the inner side of the eyebrow) covers the inner corner (medial canthus) of the eye.
- Strabismus: Established clinically when the eyes do not align in the same direction.

2.2. Oral manifestations

Mouth: Incompetent lips (open mouth): Established clinically when upper and lower lips do not come in contact at rest.

Tongue:

- Macroglossia: Established clinically as an abnormally enlarged tongue that cannot be contained within the oral cavity and has crenated lateral borders.
- Fissured: Established clinically as the presence of one or more fissures on the dorsum surface of the tongue.
- Palate: Narrow high arched palate: Established clinically when the palate is narrow and constructed with a high vault.
- Cleft palate: Established from medical history of the patients as well as intraoral examination.

2.3. Radiographic examination

Panoramic radiographs, lateral and posteroanterior cephalometric views were performed for both groups, unless recent radiographs were available. The radiographs were done at the College of Dentistry, King Saud University. The Panoramic and skull radiographs were taken using a panoramic machine OP 100 with a tube voltage of 57–85 KV and a tube current of 2–16 mA with a minimum total filtration of 2.5 mmAl, using cranex intensifying screen (HI plus regular speed) and Kodak X-OMAT RP pan DF 75 under strict radiation protection measures. All the radiographs were processed with an automatic processer according to the manufacturer’s instructions. The radiographs were carefully interpreted using a light viewer under room dim light conditions for the presence of the following features:

- Thinning of the cranial vault: Evaluated subjectively when there was a decreased distance in diploe space (space between inner and outer cortical plates) of the cranial vault.
- Delayed closure of the sutures: This was evaluated subjectively when the cranial sutures appeared wide or patent compared to the control.
Absent or hypoplastic sinuses: Both the maxillary and frontal air sinuses were considered to be absent when the shadow of the sinus could not be identified on the radiographs and hypoplastic when it appeared to be smaller than usual.

Descriptive analysis for continuous variables was reported as mean values, range and standard deviation. Categorical variables were summarized as frequencies and percentages. The comparisons between Down’s syndrome patients and control group were performed using Pearson’s chi-square test with statistical significance set at \( P < 0.05 \). Statistical package of the Social Sciences (SPSS) was used for all the statistical analyses.

3. Results

The study sample consisted of two groups. There were 30 patients in both groups. In each group there were 14 males and 16 females with an age range from 12 to 24 years. In the Down’s syndrome group, the mean age was 15.93 SD (3.290), whereas, in the control group the mean age was 14.73 SD (3.016) years.

<table>
<thead>
<tr>
<th>Table 1</th>
<th>Facial manifestations in 30 Down’s syndrome patients.</th>
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<tbody>
<tr>
<td></td>
<td>Frequency (%)</td>
</tr>
<tr>
<td>Brachycephaly</td>
<td>16 (53.3)</td>
</tr>
<tr>
<td>Depressed nose</td>
<td>28 (93.3)</td>
</tr>
<tr>
<td>Slanting of eyes</td>
<td>28 (93.3)</td>
</tr>
<tr>
<td>Epicanthic fold</td>
<td>20 (66.7)</td>
</tr>
<tr>
<td>Strabismus</td>
<td>10 (33.3)</td>
</tr>
<tr>
<td>Incompetent lips</td>
<td>30 (100)</td>
</tr>
<tr>
<td>Macroglossia</td>
<td>18 (60)</td>
</tr>
<tr>
<td>Fissured tongue</td>
<td>21 (70)</td>
</tr>
<tr>
<td>Narrow high arched palate</td>
<td>17 (56.7)</td>
</tr>
<tr>
<td>Cleft palate</td>
<td>0 (0)</td>
</tr>
</tbody>
</table>

Figure 1  Clinical photograph of a 19-year old Down’s syndrome male, showing: (A) the abnormal shape of the head depressed nasal bridge, as well as the incompetent lips, (B) macroglossia and fissured tongue in the same patient and (C) strabismus of the eyes.
3.1. Orofacial manifestations in Down’s syndrome

Table 1 summarizes the most common facial manifestations presented in Down’s syndrome patients. None of the cases included in this study had cleft palate. Fig. 1 represents various clinical facial manifestations in Down’s syndrome group included in this study, whereas Fig. 2 shows the deep palatal arch.

Thinning of the cranial vault and delayed closure of the sutures could be assessed in 27 radiographs. They were found in 16 (59.3%) and 10 (37%) cases, respectively. Hypoplastic maxillary sinuses were not a common finding in this group as it was found in two cases (6.9%). Frontal air sinuses agenesis was very common in Down’s syndrome patients, it was found in 25 (86.2%) cases, whereas, hypoplastic frontal air sinuses were uncommon in this group, it was recorded in only one patient (3.4%) Fig. 3.

Using the Pearson’s chi-square test, there was a significant difference between the Down’s syndrome group and control regarding the thinning of the cranial vault, delayed closure of the sutures and in the absent or hypoplastic frontal air sinuses. Table 2 summarizes the frequencies, percentages and $P$ value of the chi-square test for the craniofacial radiographic manifestations in both groups.

4. Discussion

In this study, Down’s syndrome in particular was chosen to investigate as in Saudi Arabia there is a relatively high incidence of Down’s syndrome with a reported incidence of 1 in every 554 live births. A trend toward an increased incidence of Down’s syndrome with advanced maternal age has been found (Niazi et al., 1995). Moreover, the life expectancy of these patients has increased significantly over the past few years (Horbelt, 2007; Kumar et al., 1997). With advanced medical care and facilities, Down’s syndrome patients are living longer and are being more socially involved than before. With this decreased mortality rate and increased incidence, the overall result is a growing population of Down’s syndrome individuals in Saudi Arabia.

The results of the present study revealed that, brachycephaly was observed in more than half of the Down’s syndrome group. This observation is lower than that reported by Levinson et al. (1955) who studied the variability of clinical features in 50 Down’s syndrome subjects up to 17 years of age and found that 82% of the patients presented with brachycephaly with flat occipit. On the other hand, the reported incidence in this study, of depressed nose, slanting eyes and epicanthic folds was comparable to the findings found by Cohen and Winer (1965) and Levinson et al. (1955). Levinson et al. (1955) found that 14% of his study samples were affected by strabismus. However, the strabismus observed in the present study was higher than that reported by Levinson et al.
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Table 2 Radiographic manifestations of the craniofacial anomalies in both groups.

<table>
<thead>
<tr>
<th>Down’s syndrome group</th>
<th>Control group</th>
<th>P value for both groups*</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Males frequency (%) within gender</td>
<td>Females frequency (%) within gender</td>
</tr>
<tr>
<td>Thinning of the cranial vault***</td>
<td>7 (58.3)</td>
<td>9 (60)</td>
</tr>
<tr>
<td>Delayed closure of the cranial sutures ***</td>
<td>6 (50)</td>
<td>4 (26.7)</td>
</tr>
<tr>
<td>Absent maxillary sinuses</td>
<td>0 (0)</td>
<td>0 (0)</td>
</tr>
<tr>
<td>Hypoplastic maxillary sinuses</td>
<td>1 (7.7)</td>
<td>1 (6.2)</td>
</tr>
<tr>
<td>Absent frontal air sinuses</td>
<td>10 (76.9)</td>
<td>15 (93.8)</td>
</tr>
<tr>
<td>Hypoplastic frontal air sinuses</td>
<td>1 (7.7)</td>
<td>0 (0)</td>
</tr>
</tbody>
</table>

* P value for chi-square test.
*** Percentages calculated out of 27.

(1955) and slightly less than that reported by Cohen and Winer (1965).

With regard to oral manifestations in Down’s syndrome patients, incompetent lips were observed in all of the cases in the present study. This finding is higher than that reported in the literature (Asokan et al., 2008; Levinson et al., 1955; Smith et al., 1976). The mouth is held open because of the relatively narrow nasopharynx and unusually enlarged tonsils and adenoids. A similar explanation for the open mouth and tongue protrusion, related to the need to provide an airway. Also, the mouth is open as secondary to the protrusion of the enlarged tongue (Smith et al., 1976).

This condition may lead to mouth breathing, drooling, chapped lower lip and angular cheilitis. Mouth breathing leads to chronic periodontitis and respiratory tract infections (Desai, 1997; Horbelt, 2007). In addition, chronic mouth breathing may lead to decrease in saliva and dryness of the mouth. This reduces the natural cleansing that occurs in the oral cavity and may contribute to the development of caries (Pilcher, 1998).

In this study, clinical examination of the tongue revealed that macroglossia is found in a high percentage of the Down’s syndrome group which is the same as the finding reported by (Asokan et al., 2008; Gullikson, 1973; Smith, 2001). However, macroglossia was found in only 30% in the study of Levinson et al. (1955). Cohen and Winer (1965) studied the dental characteristics in 123 Down’s syndrome patients with an age range from 3 to 30 years, they found that macroglossia was found in 11.3% which is lower than the prevalence reported in this study. This could be due to the difference in the sample size.

Controversy exists between authors regarding whether macroglossia is true or relative. Some authors have speculated that macroglossia is relative due to the small oral cavity. According to some authors, macroglossia is caused by inadequate lymphatic drainage (Desai, 1997; Horbelt, 2007; Smith et al., 1976). The pressure of the enlarged tongue against the mandible and teeth can produce a crenate lateral border to the tongue, open bite, spacing of teeth and mandibular prognathism (Cohen and Winer, 1965; Levinson et al., 1955). This could be due to the small sample size of this study in comparison to their studies. Aging may contribute to the development of fissured tongue (Neville et al., 2009). It has been observed that with age, the tongue in people with Down’s syndrome tend to develop cracks and fissures (Pilcher, 1998). In 1953 Oster cited from (Cohen and Cohen, 1971) found fissured tongue in patients at all ages but noted an increase in frequency with age. Therefore, it is possible that the percentage of fissured tongues in the present study is higher than that reported in previous studies due to the difference in the age range which included 12–24 years, whereas the previous studies included younger patients in which the fissures possibly may not have developed yet.

Fissured tongue can be attributed with evidence to that the condition may be either a polygenic trait or an autosomal dominant. Moreover, aging and local factors may contribute to its development (Neville et al., 2009). These fissures can become impacted with food and cause halitosis. This can be controlled by regular brushing of the dorsal surface of the tongue (Desai, 1997; Neville et al., 2009).

In the literature, the reported prevalence of high arched palate in Down’s syndrome ranges from 33.3% to 74% (Gullikson, 1973; Levinson et al., 1955; McMillan and Kashgarian, 1961). The results of this study fall within this range. Some authors have explained this as a result of midface hypoplasia (Desai, 1997). Others speculated that in actuality the vault is of normal height but the sides of the hard palate are abnormally thick (Horbelt, 2007; Pilcher, 1998). However, in those studies as well as the present study this manifestation was evaluated subjectively based on pure clinical observations.

The constricted narrow palate may create less space in the oral cavity for the tongue, affecting both speech and mastication (Pilcher, 1998). In addition, also “V” shaped high vault palates may show soft palate insufficiency and reduce the retention of maxillary dentures (Desai, 1997).

In general, the oro-facial manifestations observed in this study are in accordance with those reported in previous studies. However, the differences noted in the prevalence of reported manifestations in this study and those in previous studies regarding some of the features may reflect the variability observed among the Down’s syndrome population observed by Levinson et al. (1955), who found that the clinical stigmata observed among the Down’s syndrome patients are variable for all the characteristic features of this syndrome.

The findings of the present study regarding craniofacial radiographic manifestations: are consistent with those re-
ported by other investigators (Clift, 1922; Spitzer, 1967; Spitzer and Quilliam, 1958; Spitzer et al., 1961; Spitzer and Robinson, 1955).

In this study, thinning of the calvarium was observed radiographically in Down’s syndrome patients. This is in agreement with the findings of Spitzer et al.

Delayed closure of the sutures was not a common finding in Down’s syndrome patients. Spitzer and Quilliam (1958) and Spitzer and Robinson (1955) also observed this radiographic finding among their study sample. The delayed closure of the sutures observed in this study could be attributed to underdeveloped brain in Down’s syndrome patients. Thinning of the calvarium along with the wide sutures may make the Down’s syndrome patients more susceptible to fracture in case of a traumatic injury.

A significantly high prevalence of congenitally absent frontal air sinuses was observed among the Down’s syndrome group in the present study. This finding is supported by others (Frostad et al., 1971; Spitzer and Quilliam, 1958; Spitzer et al., 1961; Spitzer and Robinson, 1955) despite the differences in the age range of these studies. However, hypoplastic frontal air sinuses were not considered a common finding in this study. Similar findings were reported by Spitzer and Robinson (1955). However, Spitzer and Quilliam (1958) reported hypoplastic frontal air sinuses in 60% out of the 20 Down’s syndrome patients ranged from 4 to 15 years. Frontal air sinuses normally develop at the age of 6 years in normal individuals. This may explain the high prevalence reported by Spitzer and Quilliam (1958) since there is a possibility that frontal air sinuses which were considered to be hypoplastic are actually in the early stage of the development and have not yet reached their maximum size.

In this study, agenesis of the maxillary sinus was not observed. This is in contrast to the findings of Spitzer and Robinson (1955) who reported this finding in only 3.6%. Therefore, congenital absence of maxillary sinuses is not considered a common finding in Down’s syndrome. However, in the present study, hypoplastic maxillary sinuses were found in only 3.4% of the Down’s syndrome group. This finding is in agreement with Spitzer and Robinson (1955). However, it is in contrast with the findings of Spitzer and Quilliam (1958) who reported this finding in 45% of their sample. This may be attributed to the younger age group included in their study, since lateral growth of the maxillary sinuses usually ceases by the fifteenth year of age.

In the present study, no significant differences were found between males and females in the Down’s syndrome group in contrast to some of the studies.

In conclusion, there is a significantly high prevalence of facial and cranial characteristics among Saudi Down’s syndrome patients when compared to normal controls of the same population.

References


