Facial cleft and associated anomalies: incidence among infants at a Jordanian medical centre

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Received: 27/10/05; accepted: 23.01/06

ABSTRACT To estimate incidence of cleft lip and cleft palate among Jordanian infants and to identify associated anomalies, we carried out a prospective, hospital-based study over a 5-year period, January 2000–January 2005. The total number of liveborn infants was 25,440, 60 of whom (2.4/1000) had facial clefts: 20 (33%) with cleft lip, 15 (25%) with cleft palate and 25 (42%) with both. Congenital heart disease was the commonest associated anomaly (47%), followed by skeletal abnormalities (13%). Renal anomalies were found in 10% of cases. The high prevalence of congenital heart disease may justify routine echocardiography screening. The high consanguineous marriage rate may be a contributing factor to the high incidence of cleft anomalies.

Fissures faciales et anomalies associées : incidence chez les nourrissons d’un centre médical jordanien

RÉSUMÉ Afin d’estimer l’incidence des cas de fente labiale et de fente palatine chez les nourrissons jordaniens et de repérer les anomalies associées, nous avons mené une étude prospective en milieu hospitalier sur une période de cinq ans, de janvier 2000 à janvier 2005. Le nombre total d’enfants nés vivants a été de 25,440, dont 60 (2,4/1000) présentaient des fissures faciales : 20 (33 %) une fente labiale, 15 (25 %) une fente palatine et 25 (42 %) une fente labio-palatine. Parmi les anomalies associées recensées, la cardiopathie congénitale était la plus courante (47 %), suivie des anomalies squelettiques (13 %). Des anomalies rénales ont été observées dans 10 % des cas. La prévalence élevée de cardiopathies congénitales peut justifier un dépistage systématique par échocardiographie. Le taux élevé de mariages consanguins peut être un facteur contribuant à l’incidence élevée des anomalies telles que ces fissures.
Introduction

Cleft lip, with or without cleft palate, is a common congenital anomaly that has a strong negative effect on a family expecting to have a normal smiling baby. It results from failure of fusion of the fronto-nasal and maxillary processes. Worldwide, estimated incidence is about 1/1000 births [1]. It is usually of polygenic inheritance (rarely autosomal dominant) and has a higher frequency in some families. It is more common in the pregnancies of older mothers. The risk of recurrence in subsequent pregnancies is about 5%. It may be unilateral (70% on left side) or bilateral.

Cleft palate results from failure of fusion of the palatine processes and the nasal septum. Incidence is estimated to be around 1/2000 births [2]. In 70% of cases it is associated with cleft lip. A variety of clefting abnormalities occur [1–3]: simple cleft of the soft palate; submucous cleft (a bony defect completely covered by mucosa, often there is bifid uvula; and complete cleft of the soft and hard palate, which may involve the alveolar bone. This may be unilateral, bilateral, or involve the midline.

Cleft lip and cleft palate have been associated with maternal anticonvulsant therapy, and can also occur in fetal alcohol syndrome [4].

Descriptive epidemiologic studies have been carried out in many countries; however, no such study has ever been performed in Jordan. We describe a hospital-based study that was conducted at King Hussein Medical Centre to estimate the incidence of cleft lip and cleft palate among Jordanian infants and to identify the commonest associated congenital anomalies.

Methods

This study was carried out at the maternity care unit, a regular unit that performs 300–500 deliveries monthly, in King Hussein Medical Centre in Jordan. The centre is a large public hospital that serves the capital city of Amman and its suburbs. The estimated population of this area is about 1 million.

All liveborn babies, both term and preterm, who were born in the maternal care unit over a 5-year period from January 2000 to January 2005 were included in the survey. Stillborn babies were excluded. The total number of infants with cleft lip, cleft palate or both was determined prospectively.

All mothers of affected babies were interviewed postnatally by the paediatricians in the neonatal unit. A detailed antenatal history, including previous medical illness, drug exposure, antenatal visits, vitamin intake, and ultrasound scans was taken from each mother. Family history was also reviewed. After thorough physical examination, all affected babies had a skeletal survey, 2D echocardiography and renal ultrasound to identify associated anomalies. These were the only anomalies looked for because they are easy to detect and screen for. Dental anomalies were excluded because they were considered part of the cleft anomalies. Central nervous system malformations were also excluded because they need special neuroimaging studies that could not be performed for all affected infants as routine screening tests, although they were done for some infants with multiple anomalies. Chromosomal and genetic studies were not done as screening because they are not available in the centre, and are too expensive to be performed outside.

Multiple malformation was deemed to be present if more than 2 organ systems were affected.

Results

The total number of liveborn babies during the study period was 25 440. Sixty were
born with facial clefts (2.4/1000); 20 of these (33%) had isolated cleft lip [left-sided in 15 (75%) and right-sided in 5 (25%)]. All 20 were full term babies. Male to female ratio was 3:1. Skeletal survey and renal ultrasound were normal in all, but 5 (25%) had congenital heart disease, detected using 2D echocardiogram (3 ventricular septal defect; 2 atrial septal defect).

Fifteen infants (25%) had isolated mid-line cleft palate; all were full term males; all had normal skeletal survey and renal ultrasound, but 8 (53%) had congenital heart disease (5 ventricular septal defect; 2 atrial septal defect; and 1 single ventricle).

Twenty five babies (42%) had combined cleft lip and cleft palate, with male to female ratio 3:1. Five babies (20%) had birth weight < 2500 g. Fifteen (60%) had cardiac defects and some of these also had other associated defects (Table 1). Three infants had multiple malformation syndromes: EEC syndrome [ectrodactyly, ectodermal dysplasia, and cleft lip or palate]; pseudothalidomide syndrome; and amniotic constriction band sequence.

On reviewing the maternal history of the 60 infants born with facial clefts, we found:

- all mothers multigravida
- age range was 25–38 years
- consanguineous marriage in 50 cases (83%)
- none of the previously born siblings were affected

<table>
<thead>
<tr>
<th>Facial cleft</th>
<th>No.</th>
<th>CHD</th>
<th>Skeletal</th>
<th>Renal</th>
</tr>
</thead>
<tbody>
<tr>
<td>Cleft lip</td>
<td>20</td>
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<td>0</td>
</tr>
<tr>
<td>Cleft palate</td>
<td>15</td>
<td>8</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td>Cleft lip &amp; palate</td>
<td>25</td>
<td>15</td>
<td>8</td>
<td>3</td>
</tr>
</tbody>
</table>

CHD = congenital heart disease.

Discussion

Overall incidence of facial clefts was around 2.4/1000 live births. The frequency of associated congenital defects was 47%; other studies have reported a frequency that varied from 7% to 65% depending on the type of study population or whether it was hospital based [5–7]. The high frequency in our study could be because this was a hospital-based, rather than population-based, study and also because of the high rate of consanguineous marriage in our society. Consanguineous marriage is common in Arab communities, and this may lead to a higher incidence of congenital malformations in offspring, as reported in a number of other studies [8,9]. The 3 main types of cleft, isolated cleft lip, isolated cleft palate, and complete cleft lip and palate, were almost equally distributed in the group of infants reported here; however, the prevalence of associated congenital defects was not evenly distributed among these 3 groups. Of the infants with cleft lip, only 25% had other defects whereas 53% of those with cleft palate and 60% of those with bilateral cleft lip and cleft palate had other defects. A more extensive cleft seems to be associated with a higher risk of other congenital defects. This is similar to what has been reported in other studies [6,7].
Isolated cleft lip or palate was not associated with impaired fetal growth or prematurity. In contrast, 20% of babies with combined cleft lip and palate had birth weight < 2500 g.

There are divergent reports in the literature as to which congenital malformations are most common in cleft babies. Stark, in a review of 100 infants, found clubfoot to be the most common malformation [10]. Other investigators have reported an increased number of malformations in the head region, the extremities, the genital region or the central nervous system [9,11]. The most common single congenital anomaly in our study was congenital heart disease; it occurred in about 50% of all affected babies. Anomalies of the extremities and skeletal system were mainly associated with cleft lip and palate. Renal anomalies were relatively rare and associated with more severe clefts.

**Conclusion**

The high prevalence of malformations in cleft infants emphasizes the need for a thorough investigation of these infants; routine echocardiography screening for cardiac malformations may need to be considered. Close cooperation between the plastic surgeon, the orthodontist and the paediatrician is important to comprehensively cover all aspects of these often complicated cases. Consanguineous marriage should be discouraged in our society as it is implicated in the higher incidence of congenital malformations, although more formal studies are needed to investigate this issue.

**References**


