Clinical Characteristics of Children with Orofacial Cleft in a Tertiary Centre in Hong Kong

KW Chan, KH Lee, KKY Pang, JW Mou, YH Tam

Abstract

Objective: With the advancement in antenatal screening technique, there is an increase in antenatal diagnosis of orofacial cleft. Updated information on the clinical characteristics and associated anomalies in children with orofacial cleft is essential in the antenatal counseling, subsequent postnatal management and to anticipate resource allocation. Methods: A retrospective review was conducted in a university teaching hospital in Hong Kong for all children with orofacial cleft who underwent primary surgery from 1 January 1996 to 30 June 2011. Results: 274 children (M:F 140:134) with orofacial cleft were included in this study. Fifty-five (20%) children had isolated cleft lip (CL), 91 (33%) children had cleft lip and palate (CLP). One hundred and twenty-three (45%) children had cleft palate (CP) only. Five (2%) children had macrostomia. Among all children with CL and CLP, boy was more commonly affected (66%). In unilateral CL and CLP cases, left side was 3 times more commonly involved. CP was more common for girls (62%). Twenty-eight children were syndromic or had multiple malformations. In this group of children, 26 had isolated CP ($P<0.05$). Pierre Robin syndrome was the commonest associate syndrome (n=14). Ten children had major congenital cardiac disease. Two children had structural brain anomalies. Conclusion: This study provides updated clinical characteristics in children with orofacial cleft in Hong Kong. The associated anomalies were rare in children with CL and CLP. Cardiac and brain anomalies were seen in children with orofacial cleft. Echocardiogram and USG brain is required in selected cases.

Key words Cleft; Lip; Palate

Introduction

Orofacial cleft is the commonest congenital facial anomalies.1 The incidence of orofacial cleft is quoted at 1 per 500 to 1000 live births. A higher incidence is reported in Asia.1,2 The Prince of Wales Hospital is a tertiary referral centre for children with orofacial cleft. In our hospital, a well-established prenatal diagnosis clinic (PDC) under the Fetal Medicine Team has been in operation for more than a decade.3 Our team works in close collaboration with the Fetal Medicine Team. Antenatal counseling will be provided if orofacial cleft is detected by antenatal ultrasonogram (USG). Postnatally, the assessment of any feeding problem and the work up of any associated malformations are required.

Multi-disciplinary approach is required in the management of patient with orofacial cleft. Holistic care is provided by the paediatrician, paediatric; plastic; ear, nose and throat; dental and maxillofacial surgeon, speech therapist, dietitian and nursing staffs. The timing of primary cleft surgery is fixed to the following schedule. Repair of cleft lip is scheduled at 3 months of age and repair of cleft palate is scheduled at 1 year of age. If cleft alveolar is
present, the repair is usually performed at 8-9 year of age. Maxilla osteotomy, dental wiring and rhinoplasty will be performed in early adulthood in selected children with severe maxillary hypoplasia and asymmetry of the nasal profile.

With the introduction of antenatal counseling program, data on the local clinical characteristics and the incidence of associated anomalies in children with orofacial cleft is needed in order to provide more accurate information in antenatal counselling and in postnatal management.

Methods

All children with orofacial cleft who underwent primary surgery in the Prince of Wales Hospital in Hong Kong from 1 January 1996 to 30 June 2011 were reviewed. Children with orofacial cleft referred for secondary lip revision surgery or pharyngoplasty were excluded. The distribution of various types of orofacial clefts, the sex of the patients and any associated malformations or syndrome were studied.

Statistical Methods

Statistical analysis was accomplished using the SPSS program for Windows 15.0 (SPSS, Chicago, Illinois, USA). Chi-square test was used to compare the categorical data. \( P<0.05 \) was considered statistically significant.

Results

Type, Sex and Laterality of the Orofacial Cleft

Over a 15-year period, 274 children with orofacial cleft underwent primary orofacial cleft surgery. Of these, 140 were male and 134 were female. Fifty-five (20%) children had isolated cleft lip (CL), 91 (33%) children had cleft lip and palate (CLP). One hundred and twenty-three (45%) children had cleft palate (CP) only. Five (2%) children had macrostomia (Figure 1).

Boys were more commonly affected in children with CL and CLP (M:F; 97:49). In unilateral cases, left side was more commonly involved (L:R; 94:31). There is no significant difference on the sex and the laterality of cleft in unilateral CL and CLP \( (P>0.05) \). Bilateral isolated cleft lip was rare and only 1 child was affected. One child had midline cleft lip and palate. In contrary, girls were more commonly had CP (M:F; 27:64). Five children had macrostomia (M:F; 2:3) (Figure 2).

Associated Malformations

Twenty children were syndromic. Fourteen children had Pierre Robin syndrome. Two children had Van der Woude syndrome. One child had velocardiofacial syndrome. One child had kabuki syndrome, 1 child had Crouzon syndrome and 1 had Apert syndrome (Table 1).

Beside the 20 children with a well known syndrome, 8 children had multiple malformations. A total of 10% (28/274) of children with orofacial cleft was either syndromic or had multiple malformations.

Regarding the relationship between the type of orofacial cleft and the 28 syndromic children or children who had multiple malformations, majority of children had isolated CP \( (n=26) \) \( (P<0.05) \). Two had CLP. None of the child with isolated CL had multiple malformations.

Congenital heart disease (CHD) was diagnosed in 10 children by echocardiogram. Four children had ventricular septal defect (VSD), 3 children had tetralogy of Fallot (TOF), 2 children had atrial septal defect (ASD), and 1 had pulmonary stenosis (PS). Seven out of the 10 children required cardiac surgery. In addition, 7 children had other associated anomalies. In this group of children, 4 children had CLP, 6 had isolated CP (Table 2) \( (P>0.05) \).

USG or magnetic resonance imaging (MRI) brain was performed in children with dysmorphism or suspect midline defect. Only two children had structural brain anomalies. Partial agenesis of corpus callosum was detected in a dysmorphic child. Anterior encephalocele was detected in another child who had midline cleft lip and palate.

Figure 1  Distribution of cleft type.
Table 1  Associated malformation and the incidence

<table>
<thead>
<tr>
<th>Type of associated anomalies</th>
<th>Number</th>
<th>Type of cleft CLP</th>
<th>CP</th>
</tr>
</thead>
<tbody>
<tr>
<td>Syndromic group</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Pierre Robin</td>
<td>14</td>
<td>0</td>
<td>14</td>
</tr>
<tr>
<td>Van der Woude</td>
<td>2</td>
<td>1</td>
<td>1</td>
</tr>
<tr>
<td>Velocardiofacial</td>
<td>1</td>
<td>0</td>
<td>1</td>
</tr>
<tr>
<td>Kabuki</td>
<td>1</td>
<td>0</td>
<td>1</td>
</tr>
<tr>
<td>Crouzon</td>
<td>1</td>
<td>0</td>
<td>1</td>
</tr>
<tr>
<td>Apert</td>
<td>1</td>
<td>0</td>
<td>1</td>
</tr>
<tr>
<td>Multiple group</td>
<td>8</td>
<td>1</td>
<td>7</td>
</tr>
</tbody>
</table>

CLP: cleft lip and palate; CP: cleft palate

Table 2  Distribution of the major congenital heart disease

<table>
<thead>
<tr>
<th>Type of congenital heart disease</th>
<th>Type of cleft CLP</th>
<th>CP</th>
</tr>
</thead>
<tbody>
<tr>
<td>Ventricular septal defect</td>
<td>1</td>
<td>3</td>
</tr>
<tr>
<td>Tetralogy of fallot</td>
<td>2</td>
<td>1</td>
</tr>
<tr>
<td>Atrial septal defect</td>
<td>1</td>
<td>1</td>
</tr>
<tr>
<td>Pulmonary stenosis</td>
<td>0</td>
<td>1</td>
</tr>
</tbody>
</table>

CLP: cleft lip and palate; CP: cleft palate

Discussion

This is an updated review of the clinical characteristics of orofacial cleft in Hong Kong. The first report on the distribution of orofacial cleft in Caucasian was CL:CLP:CP-1:2:1. More recent studies in Asia and Africa showed similar distribution. In contrary, a high proportion of CP (45%) was noticed in our study (Figure 1). This distribution was also noticed in more recent European studies. The different in distribution may be racially and environmentally determined. Besides, different studies may have different recruitment criteria. Some studies recruited both stillbirth and live birth. Some studies excluded syndromic children. Another reason that could explain the increase in prevalence of CP in recent western studies and in Hong Kong may be related to the antenatal screening and subsequent termination of pregnancy. The reported rate of antenatally detected CL or CLP ranged from 17.5-58%. However, the antenatally detected rate of isolated CP ranged only from 0-6.5%. The reported incidence of terminations of pregnancy caused by diagnosis of a facial cleft ranged from 0-92%. If the rate of terminations of pregnancy caused by the diagnosis of a facial cleft increased, the prevalence of CP will be increased because isolated cleft palate is rarely detected antenatally.

Most published series showed left side involvement in two-third of cases in children with CL and CLP. The distribution of the laterality of unilateral cleft in our series was similar to other series. Left side was more commonly affected for CL and CLP. Sixty-nine percent of isolated cleft lip was on the left side. Sixty-three percent of children with CLP had left side involvement (Figure 2).

The sex distribution of orofacial cleft was also similar to those published series. There were more boys with CL and CLP (Ratio with girls 2:1). On the other hand, CP was more frequently occurs in girls (Ratio with girls 3:2).

Orofacial cleft had a well known association with other congenital malformations, especially in children with isolated CP. Cleft children can be classified as
syndromic or multiple group. In the syndromic group, a well known syndrome can be identified. In this study, 20 children had a well known syndrome associated with orofacial cleft. In the multiple group, the children had dysmorphism and multiple malformations but no well-defined syndrome can be identified. Eight children in this study belonged to this group. The overall prevalence of syndromic and multiple group in children with isolated CP was 17%. On the other hand, the prevalence in children with CLP and CL was only 2.2% and 0% respectively (Table 1). In case if fetus with CL or CLP is detected antenatally, this local updated information can be provided during antenatal counseling and may allay the fears of parent.

The prevalence of CHD in patient with orofacial cleft ranged from 1.3 to 27%. A high prevalence may be related to the higher incidence of associated syndrome in the study population. Consanguinity was a well known risk of associated malformations or CHD. Difference in the prevalence may also be related to the study design. The definition of CHD was different in different studies. Chen underwent echocardiogram for all children with CP. The prevalence was 11.1% and half of the children had ASD. Whether the ASD had spontaneously resolved or had any clinical significant was not reported. In another study, a high incidence of mitral valve prolapse (MVP) was reported. Inclusion of MVP as a CHD may be debatable. In our center, the prevalence of CHD was 3.4%. The prevalence was 24% in syndromic or multiple group. Some centers advocated performing routine echocardiogram in order to detect the presence of CHD. It may lead to detect of clinically incidental CHD. Screening for CHD by echocardiogram in syndromic patient or in children with murmur may be a more resource-friendly approach.

The use of routine USG brain in children with orofacial cleft is also debatable. In our centre, only 2 children had structural brain anomalies detected by USG or MRI. One child had midline cleft. Midline cleft had a well known association with structural brain anomalies such as holoprosencephaly. He had anterior encephalocele and subsequent underwent repair. Another child with partial agenesis of corpus callosum had multiple malformations. Recently MRI study of the brain in children with orofacial cleft showed different proportion in grey and white matter. Further studies are needed to understand the importance of this finding and any potential clinical significance. The use of USG brain is advised in syndromic patient or children with midline cleft.

In conclusion, this study provides updated clinical information on children with orofacial cleft. The associated syndromic anomalies were rare in children with CL and CLP. Echocardiogram and USG brain is necessary in selected cases. This study also provides updated information useful in parental counseling and to anticipate resources allocation.

References