A review of the rate of occurrence of cleft lip and palate in Chinese people

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Published data were used to determine the rate of occurrence of cleft lip and/or palate in the Chinese population. The rate of occurrence ranged from 1.33 to 2.23 per 1000 live and stillbirths. Most studies were performed in communities with mixed racial groups, and only three obtained data from multiple sources. Half of the studies included live and still births. The majority of the studies reported pooled figures for subjects with and without other malformations. Furthermore, three different classifications of clefts were used by the authors. Clefts involving the lip and palate had the highest occurrence rate in most studies. Unfortunately, the differences in the rate of occurrence may reflect differences in the technique used for data collection rather than real differences.

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Introduction

A review of the literature seems to indicate that the rate of occurrence of cleft lip and/or palate (CLP) is well established and that the prevalence of CLP is generally considered to be approximately 1 per 1000 (Table 1).[1,2] There is variation, however, between racial groups,[3] with the Mongoloid groups having the highest rate and blacks the lowest.[4] Variation also occurs between reporting centres; undoubtedly, the best figures come from the national register in Denmark, which indicates that the incidence for the period 1976 to 1981, was 1.89 per 1000 births.[5]

Careful analysis of the published literature casts doubt on the validity of the rates of CLP because the epidemiological investigations have tended to be compromised by numerous methodological problems.[6] It has even been said that the problem of incomplete ascertainment has occurred in all of the published studies.[7] The final prevalence figures can be affected by one or several of the following raw data: the design and limitations of the study, the geographic location, the method of reporting, and the population being sampled.[8] Some investigators have attempted to clarify the variables associated with the clefting phenomena. Unfortunately, few variables have remained consistent throughout all of these reports, thus making direct comparisons difficult or invalid. Nevertheless, one of the most consistent findings from these studies is a distinct racial gradient in the incidence of CLP, with higher frequencies being reported for the Mongoloid groups.[9,10]

Although the Chinese belong to a racial group with an apparently higher frequency of CLP, the reported epidemiological data suffer from inherent weaknesses in study design and methodological inadequacies that have apparently been neglected, or were possibly beyond the control of the investigators. It is proposed, with due consideration for these factors, to review the epidemiological data available on the rate of occurrence of CLP in Chinese people.

Rate of occurrence of cleft lip and palate

Published reports are often at variance over the method that should be used to express the occurrence rate of CLP. The rates should be recorded in terms that facilitate prospective, and if possible, retrospective comparisons between study populations.[9] Many investigators have expressed the incidence rate as the number

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per 1000 births, live and/or still[11-17]; however, there are a number of notable exceptions.[10,18,19]

The rate of occurrence ranges from 1.09 to 4.04 per 1000 births. The Washington study by Emanuel and co-workers[13] gives the highest incidence at 4.04 per 1000 live births. This study was performed, however, on a predominantly Caucasian population and hence, the subsample of 1239 Chinese subjects is probably too small to be representative because of the low occurrence rate of CLP. The next highest incidence, of 2.23 per 1000 births is from the Taipei collaborative study, which included both live and stillbirths.[12] The lowest figure that includes both live and stillbirths is the Singapore figure of 1.33 per 1000 births.[10]

The relevant information from 10 published studies is summarised in Table 2. The differences in the figures may not indicate actual differences but reflect variations in the data collection techniques used.[6,20]

Ethnic origins of the samples

Unfortunately, some studies were performed on base populations that contained several ethnic groups, which are well known to have different levels of risks for developing orofacial clefts.[10,13,14,16,17] The authors have then tried, in a variety of ways, to subdivide and present the data according to ethnic group. In a study conducted in British Columbia, the Chinese patients were identified by the surnames and forenames of the children and their parents and the description contained in the clinical records and photographs; all of which were supplemented by correspondence with the various Public Health Units.[14] The investigators indicated that the number of Chinese subjects was probably an underestimate because certain Chinese surnames are identical to Caucasian ones. In the majority of the studies, the method of identifying the ethnic origins of the subjects was not mentioned by the authors.

Table 1. The incidence of cleft lip and palate in different racial groups

Racial group	Clefts per 1000
Blacks	0.5
Caucasians	1.0
Japanese	2.34
American Indians	3.63

Source: Gorlin RJ, et al. Williams & Wilkins, 1971.[4]

Studies that were conducted in Sichuan province, China,[19] Taiwan,[12] and in Hong Kong[10,15,18] almost exclusively investigated Chinese populations. In Taiwan, the collaborative study confined the sample to births to Chinese parents. However, in the remaining studies the ethnic origin of the population under investigation was not specified, but simply assumed to be Chinese.

Base population

When reporting the occurrence of CLP, it is essential to define precisely the population in which the malformation was measured; that is, whether the estimates are based on all conceptions, all births, or all live births. The rates should be presented separately for live births and embryonic and foetal deaths.[21] Inclusion of stillbirths tends to elevate the incidence over and above that derived from only live births.[1,10]

Unfortunately, five of the studies reviewed in Table 2 include live and stillbirths in the study populations; however, one[12] did present separate figures for live births and stillbirths as recommended by Hook.[21] The other five studies only included live births. Therefore, when making comparisons between the figures from studies on the incidence rates of CLP, attention must be paid to the criteria of the base population.

The existing evidence suggests that clefts that are associated with other bodily malformations and syndromes should be considered to be epidemiologically different from clefts without associated malformations because the pooling of all CLP cases, with and without other malformations, adversely influences research into the aetiology of oral clefts and consequently, the accuracy of genetic counselling.[1] Only a minority of the studies involving Chinese populations have included separate figures for clefts occurring as a single anomaly and those occurring in association with other malformations[10,12,14] while the majority of studies have inappropriately reported pooled figures for incidence.

Source of the sample

The determination of the true prevalence of CLP in a given population requires that a truly representative sample of an appropriate size is identified and selected for investigation. Because of the relative rarity of CLP, a large sample size is required, which is probably why most of the investigators have not studied population-based samples. Only three of the studies have presented data from multiple sources of ascertainment[13,14,19] and most of the studies rely on hospital records. Although convenient, this approach can introduce a selection bias

Table 2. Studies of the incidence of cleft lip, cleft palate, and cleft lip and palate combined in Chinese populations from different parts of the world

Investigator	Period	Location	Source	No. of births	No. of clefts	Base population	Clefts per 1000
Wei and Chen (1965)	1955-62	Taiwan	Hospital births	14 834	28	L,* S†	1.92
Stevenson et al (1966)	na	Hong Kong	Hospital births	9876	16	L,S	1.62
Stevenson et al (1966)		Kuala Lumpur		8625	15	L,S	1.74
Stevenson et al (1966		Singapore		31 503	42	L,S	1.33
Emanuel et al (1972)	1965-68	Taiwan	Hospital births	25 517	37	L,S	2.23
Emanuel et al (1973)	1956-65	Washington	Multiple sources	1239	5	L	4.04
Lowry and Trimble (1977)	1952-71	British Columbia	Hospital births	12 430	22	L	1.76
Paterson (1977)	1955-76	Hong Kong	Hospital records	-	1800	L	1.09
Leung (1980)	1977-79	Hong Kong	Hospital births	73 464	137	L	1.86‡
Tan (1988)	1986-87	Singapore	Hospital births	18 589	28	L	2.04
Boo et al (1990)	1986-87	Kuala Lumpur	Hospital births	12 115	23	L,S	1.90
WHO (1991)	1985-88	Sichuan	Multiple sources	65 000 (annually)	553	L,S	1.89

*L live births; †S stillbirths; ‡ calculated from original data

because the surviving patients with the more severe types of cleft and those with associated congenital malformations can be included in artificially large proportions,[22] thus distorting the final prevalence figures.

Classifications of cleft type

As with any epidemiological investigation, spurious differences in the reported rates, in this instance, for orofacial clefts may be attributable to the use of different diagnostic criteria[23,24] or inadequacies in the classification systems employed for the cleft phenotypes. The subsequent grouping of different cleft types complicates comparative analysis.[25] Most of the studies have employed the classification suggested by Fogh-Andersen,[26] in which subjects are divided into three main groups, that is, isolated cleft lip, combined cleft lip and palate, and isolated cleft palate. In the other studies there have been various classifications, for example, patients have been classified into cleft lip or cleft palate, which have then been subdivided.[15,19] In the study by Leung,[15] the subjects with clefts involving both the lip and palate were counted in the cleft lip group as well as the cleft palate group, whereas in the study in Sichuan,[19] cleft lip was used to include those clefts involving the alveolar process and the palate. Another approach has been to divide the clefts into those of the primary palate, clefts of the primary and

secondary palate, and clefts of the secondary palate only.[17] This approach was adapted from a classification proposed by Kernahan and Stark.[27] In the study by Wei and Chen[11] there were no subdivisions according to cleft type. These variations create difficulties when trying to compare the rates of CLP occurrences in different populations residing in different regions.

In spite of different methods of managing the data for the different types of clefts, it is generally accepted that cleft lip with cleft palate has the highest incidence. However, some studies have found cleft lip to be more common than cleft palate[10,12,18] while in the majority of studies, cleft palate has been found to have a higher prevalence than cleft lip alone.[10,13-16] Unfortunately, the studies conducted in Washington (United States),[13] British Columbia (Canada),[14] Singapore,[10] and Malaysia[10] were based on mixed racial groups, which compromises the quality of the final data.

Sex ratio according to cleft type

One of the variables associated with cleft type that is consistent throughout almost all of the published studies is the incidence of CLP according to gender. Males are more commonly born with cleft lip, or a combination of cleft lip and palate, while females are more frequently affected by isolated clefts of the palate.[5,9,18]

Most studies have demonstrated a predominance for the male to be affected by cleft lip alone,[10,13,14,18] however, the opposite was observed in the study by Emanuel and co-workers.[12] Although four studies indicate that it is more common for males than females to have cleft lip and palate,[10,13,14,18] two studies fail to confirm this pattern.[9,11] In most studies, cleft palate occurs more often in females than males,[10,13,18] although there have been some exceptions. Cleft palate was more common in males in one study[12] and the incidence for males and females was similar in another study.[14] Unfortunately, in some instances, when the subjects have been subdivided according to cleft type, the numbers have become too small to accurately indicate whether there is any real variation between gender.

Strategies in future studies

The fact that CLP is a congenital craniofacial malformation that can easily be diagnosed immediately postpartum led to the establishment of a centralised registration system in Denmark. This system has for many years proved to be a successful means of gathering data for the rather homogeneous population of that country.[5] This principle could be adopted in Hong Kong, making it necessary for hospitals to forward data on CLP to a central register. To ensure the validity of the data, however, registration would have to be compulsory.

Alternatively, some of the inadequacies of the published data could, in the future, be avoided if multiple sources of ascertainment from population-based samples are used for the determination of incidence statistics. Strategies should also be developed to estimate the bias and the completeness of ascertainment and to make appropriate corrections to the data.[6] Furthermore, another more easily achieved approach, that satisfies the requirements of basic epidemiology, population genetics, and genetic epidemiology, is the sharing of clinical data from different centres.[9]

Conclusion

In formulating these conclusions, it must be remembered that they have been drawn from data gathered from studies with different diagnostic criteria, sources, and sample sizes. In addition, some investigators included data for stillbirths and clefts associated with other malformations and syndromes, and the subjects came from different populations from different geographic locations.

It appears from the published literature that higher quality descriptive epidemiological data for the full spectrum of orofacial clefts in the Chinese population, which fulfil the requirements of basic epidemiology and population genetics are still not available. However, bearing in mind the limitations, the currently available data has shown that the incidence of CLP in the Chinese population ranges from 1.33 to 2.23 per 1000 live and stillbirths, and that cleft lip with cleft palate appears to be the most common cleft type.

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