EDITORIAL

CLEFT LIP AND/OR PALATE: SEARCHING FOR AETIOLOGICAL FACTORS

Cleft lip and/or palate (CLP) are among the most common congenital malformations and compared to many other anomalies, CLP are easily diagnosed and described(1). These defects account for approximately 65% of all congenital craniofacial anomalies, such that approximately one baby in every 500 live births shows these clefting defects in one form or another(2). For the last three decades clefts have been the focus of numerous studies by both epidemiologists and geneticists(3). Epidemiological and family studies have suggested that both environmental and genetic factors play a role in CLP aetiology. However, which genetic and environmental factors are important for CLP is still largely an enigma(1). Currently, the strongest evidence implicates a primary gene on 6p and a role for transforming growth factor alpha (TGFA) as a modifier of the clefting status while other genes involved may include RARA, BCL3 and genes on 4_q4. However, the role of interactions with the environment, especially common exposures such as tobacco and alcohol, still require additional large epidemiological studies to be performed in concert with molecular analyses.

Based on the current developments in CLP epidemiological and genetic-epidemiological research, some areas seem very promising for 21st century research (large-scale inclusion of genetic information) while other problems (delineation of non-syndromic cases and the lack of incidence data) probably will face CLP researchers for many years to come(1). Besides these fundamental problems, CLP researchers have realised the need for co-ordinated worldwide studies of more homogenous populations.

Although CLP has been widely investigated elsewhere, there is still a dearth of information on the black populations of Africa(5). In this issue of the Journal Orkar *et al*(6) have presented some baseline data regarding the pattern of occurrence of CLP in an African population that could add value to the cumulative worldwide information on this subject. It must be emphasized, however, that future trends in research on this subject must focus not only, on basic epidemiological characteristics but also on in-depth studies of genetic and environmental risk factors. Thus, an international collaboration increases the potential for aetiological heterogeneity but, on the other hand, also increases the power to detect aetiological factors common to most settings(1). The human face provides the first recognisable image of the new-born infant experiences and thereafter a major focus for comfort and identification(4). Hence, any anomaly of the face including CLP certainly gives rise to considerable morbidity which is psychologically traumatic to the parent; and infact the community at large.

Notably, people who have CLP experience cosmetic, speech, hearing and dental problems. All these are disabilities that demand expensive and long-term management modalities. From the point of view of health economies, the optimal management of individuals with clefts of the lip and palate is, undoubtedly, a major burden at all levels of healthcare. Apart from the primary surgical procedures to reconstruct the defects, other follow up protocols may include revisions, pre-operative orthodontic treatment, oronasal fistula closure, bone grafting of the alveolar clefts and orthognathic surgeries. Furthermore, the role of the Ear, Nose and Throat specialists and speech therapists in the overall care of these patients must at all times be appreciated. Evidently the cost of optimal management of CLP cases among the economically less endowed populations has to be significantly prohibitive. Thus greater proportions of those affected throughout the diverse world populations can hardly afford even the basic desired treatment. Research efforts on this subject must, therefore focus on the aetiological factors that could be preventable. Currently it is gratifying to note that researchers on the genetic pathways of craniofacial deformities have concentrated on the areas of interventional prevention.

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