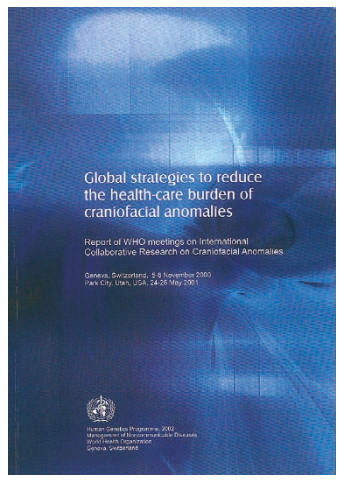
Global strategies to reduce the healthcare burden of craniofacial anomalies



Peter Mossey reviews the latest report by the World Health Organisation on the state of science in the field of craniofacial anomalies.

This publication provides a report of two consensus meetings held by international experts under the auspices of the World Health Organisation (WHO). The first meeting held in Geneva between 5th and 8th November 2000 included concurrent workshops on research concerning the genetic basis of CFA, gene-environment interactions, and the treatment of CFA. The second meeting held in Utah between 26th and 28th May 2001 considered the prevention of CFA. This is a comprehensive review of the state of the science in this field, available on the internet at www.who.org, and the following is a summary of the findings.

Craniofacial Anomalies (CFA) affect a substantial proportion of the global society. Cleft lip and/or palate, for example, occurs in approximately 1 per 500-700 births. Thus, each year there are about 220,000 new cases and the prevalence at birth varies considerably across geographic areas or ethnic groupings. The costs incurred from CFA in terms of morbidity, health care, emotional disturbance, and social and employment exclusion, are considerable for affected individuals, their families and society.

Current research regarding CFA falls into three related spheres - aetiology, prevention, and treatment. Unfortunately, much of this research is being conducted independently, with little evidence of a coherent global strategy. The goal of this initiative is to reduce duplication of effort and achieve broader coverage of priority research needs by bringing together international researchers through collaborative partnerships, and to develop global consensus on CFA research directions and common research protocols.

Research on the genetic basis of CFA has benefited dramatically over the last decade from the development of recombinant DNA technology. In over 50 craniofacial syndromes, the genes involved have either been mapped to a chromosome location or actively isolated and their structure identified. This achievement, however, represents only a fraction of the total number of craniofacial syndromes defined. The pathogenesis of the most common forms of CFA (non-syndromic) is especially challenging because they appear to arise from complex polygenic interactions with environmental factors. A co-ordinated international approach to this work will not only provide an effective means of sharing data, samples and resources, but a research strategy which takes into consideration geographic and ethnic variation in the incidence of CFA may be a key to understanding their aetiology and thereby developing strategies for primary prevention.

Research that may lead to the prevention of CFA has been based, primarily, on isolated case control studies in Europe, the USA and Latin America. As yet, these projects have occurred independently of each other, and consistent evidence about potential interventions such as dietary supplementation in the periconceptional period have yet to emerge. International standardization of research protocols, consensus on preventive interventions suitable for clinical trials, and the performance of trials in an international framework, will enhance the validity, consistency and generalisability of the evidence.

The treatment of CFA has, so far, escaped the rigours of contemporary health technology assessment, and great confusion surrounds the optimal management for even the most common conditions. For each of the many subgroups of CFA, the attainment of homogeneous samples of adequate size for randomised trials and long-term follow-up represents a formidable challenge. Multisite cooperation is a prerequisite. In the developing world, the costs of rehabilitation and problems of access put treatment beyond the reach of vast numbers of affected individuals.

The main objectives of this initiative are to develop an international network for consensus building, the planning and protocol development for international collaborative, multi-disciplinary studies of methods of treatment and prevention of CFA, supported by the creation of a directory of CFA research resources and publicly-accessible internet-based research database.

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The message that comes through loud and clear is that the ultimate humanitarian and scientific research objective in craniofacial anomalies birth defects is primary prevention, and if we aspire to this the days of isolated research effort are over, and global collaboration is necessary. It also recognises that until a viable method of primary prevention is identified, research is urgently required on surgical protocols and systems of delivering care in different geographic and economic circumstances.

Peter Mossey,
Professor of Craniofacial Development
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