Capacity Building for the Transfer of Genetic Knowledge into Practice and Prevention

Health Needs Assessment for Medical Genetic Services in Middle- and Low-Income Nations

ARNOLD CHRISTIANSON
Division of Human Genetics, National Health Laboratory Service & University of the Witwatersrand, JOHANNESBURG, South Africa.
E-mail: arnold.christianson@nhls.ac.za

RON ZIMMERN.
Public Health Genomics Foundation, Strangeways Research Laboratory, CAMBRIDGE CB1 8RN, UK.
E-mail: ron.zimmern@phgfoundation.org

For the
CAPABILITY Consortium

Introduction

Health care systems are continually confronted by challenges resulting from epidemiological transition, technological advances, changing public expectations and demands, resource limitations and the rising cost of health care[a]. To meet these challenges health care needs assessment (HCNA) was developed, beginning in the 1970s, to inform health care service planning.[1-3]

HCNA provided a rational, epidemiological assisted approach for providing information to plan, introduce and beneficially change health care services to improve the health of populations. Their main objective was to identify programmes, services, interventions, opportunities and resources to improve health care. They were undertaken recognising that resources are always limited and accepting that changing health care services required rationalisation, that is choosing fairly between competing priorities.[1,2]

Time and experience expanded the vision of HCNA beyond the confines of the medical model centred only on health care services. In the 1990s it evolved into Health Needs Assessment (HNA), a systematic, objective epidemiological, qualitative and comparative evidence-based approach to 'commissioning and planning health services' Its aim is to identifying unmet health, including health care, needs in a population and make changes to meet the unmet needs.[8] Health 'gain' is thereby achieved by identifying unmet needs and meeting them, reallocating funds from ineffective and inefficient services to unmet needs and improving poor services.[1,2,4,8,9,12]

Addressing the World Health Organisation's (WHO) Second Expert Committee on Human Genetics in 1963, at a meeting on Human Genetics and Public Health, Dr Candau, Director General, remarked on the development of genetics and 'the need for examination of the present and future significance of human genetics to public health'. Discussing epidemiological or epidemiological transition, he noted that 'with the control of infant mortality and infectious diseases inherited abnormalities are assuming a proportionately greater importance in medical practice'. Considering the future he noted the role of genetics in complex disorders[b], expressing the hope that a fuller understanding of genetics would lead to the clarification of the complex interactions between genetics and environment and their implications for the health of future generations. [5] These comments pertained to industrialised nations.

By 1960 all but two industrialised nations had infant mortality rates less than 40 per 1000 live births, indicating they had largely controlled environmental causes, infection and malnutrition, of infant and childhood mortality and morbidity. Congenital disorders[c] thus gained public health significance as a cause of infant and childhood

[a] Health care encompasses diagnosis, treatment, including rehabilitation and palliative care, counseling, prevention and health education.

[b] Complex disorders develop after birth, some manifesting in childhood but most in mid & later life. They are complex because their aetiology is multifactorial, with the environmental component being mostly postnatal. By comparison with multifactorial congenital malformations they are clinically complex, being systemic & involving different organs & systems. They include a wide range of diseases including common disorders like cancer, cardiovascular disease, diabetes, hypertension, mental disorders and stroke.

[c] Congenital disorders are defined as abnormalities of structure or function, including metabolism, which are present from birth. That is their aetiology is prenatal. The term congenital disorders is

death and disability, prompting the well documented efforts of industrialised nations over the next forty years to control congenital disorders. This included the development of medical genetic services appropriate to their needs and circumstances and underpinned by the continuing acquisition of medical genetic knowledge and technology. When the process began in the early 1960s limited epidemiological data were available and health care needs assessment was a future consideration.[6,7]

The now largely successful campaign of industrialised nations to control congenital disorders has launched the present phase of the development of their medical genetic services, the control of complex disorders.

By the turn of the 21st century, middle- and low-income countries had benefited little from the medical genetic knowledge and technology acquired and applied in industrialised nations. The prevailing consideration was that middle- and low-income countries had an unfinished agenda with infectious diseases and malnutrition and medical genetic services were still a future health need.[6,7]

In 1985 a WHO Advisory Group recognised that many middle- and low-income nations were achieving epidemiological transition by controlling infectious diseases and malnutrition and in time would need to implement and develop medical genetic services, initially like industrialised nations for the care and prevention of congenital disorders. This group explored the relationship between public health and medical genetics and recognised that the model for medical genetic services of industrialised counties would not be applicable to middle-and low-income nations. The challenge was to adapt the general principles of the tertiary care-, individual- and family-based practice of medical genetics in industrialised nations to the public health and primary health care orientated systems of middle- and low-income countries. [8,9]

Cogent approaches and policy for the development of medical genetic services in middle- and low-income nations has been achieved by individuals and groups working independently and collaboratively since 1997. The efforts were informed by the history and progress made previously in industrialised countries. They documented a holistic approach recommending the integration of effective and cost effective interventions and services into public and primary health care woman, reproductive, maternal, and child health for the care and prevention of congenital disorders. These would be supported by paediatric, obstetric, surgical and medical genetic clinical and laboratory diagnostic services in secondary and tertiary health care.[6,7,10,11]

With positive epidemiological transition, some middle- and a few low-income nations have initiated and are developing medical genetic services for the care and prevention of congenital disorders.[6,7] These countries include Argentina, Brazil, Chile, China, Cuba, Egypt, India, Iran, Malaysia, Mexico, Oman, Philippines, Saudi Arabia, South Africa and Thailand. However, recognising that many middle- and low-income nations will not be able to achieve their Millennium Development Goal (4), a two thirds reduction of under-5-year mortality rate of their 1990 status by 2015, the WHO is discussing prioritising these services.[6,13]

---

synonymous, and can be used interchangeably, with the term birth defects. Serious congenital disorders are those that are life threatening or have the potential to cause disability.[6,12]

The documented collective experience of developing medical genetic services in middle- and low-income nations is limited for congenital disorders and virtually non-existent for complex disorders. To assist middle-and low-income nations initiate and develop medical genetic services appropriate to their needs and circumstances it is recommended that they consider undertaking HNA, a suggestion proposed by the WHO in 2000.[11]

Epidemiology of Congenital and Complex Disorders

Congenital disorders are a significant cause of stillbirths and of infant and childhood disease, death and disability, particularly in many middle- and low-income nations (Table 1).

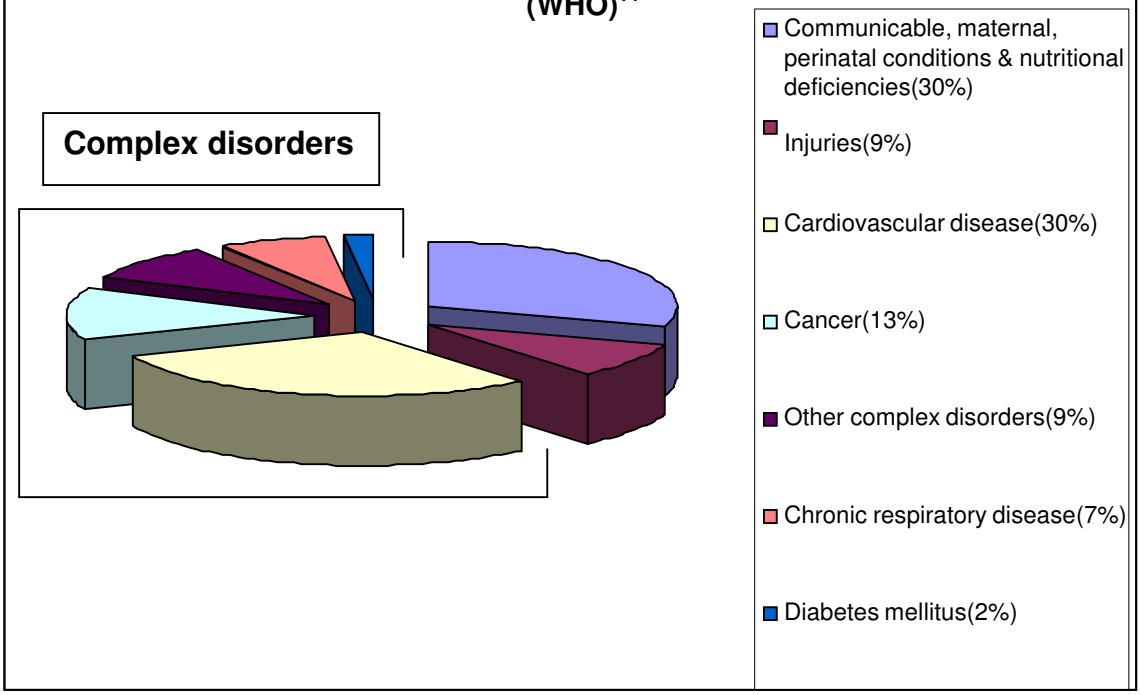
Table 1. GLOBAL GENETIC CONGENITAL DISORDERS[6].

	Low-income countries	Middle-income countries	High-income countries	Total
Annual total birth defects (millions)	4.75 60%	2.64 34%	0.49 6%	7.88
Annual early deaths of birth defects (millions)	2.38 72%	0.79 24%	0.14 4%	3.3
Annual under 5 deaths UNICEF '01 (millions)	8.8 79%	1.8 16%	0.6 4%	11

An estimated 7.88 million children are born each year with a serious congenital disorder of genetic origin. Birth prevalence of these disorders ranges from 40 per 1,000 live births in high-income countries to a maximum of 82 per 1,000 live births in low-income countries, with a global average of 60 per 1000 live births. The majority, 94%, of children with serious genetic congenital disorders are born in middle- and low-income nations, were facilities for their care is limited. A minimum estimated 3.3 million children with serious genetic congenital disorders die annually, with 95% of the deaths occurring in middle- and low-income nations. Global data on the epidemiology of congenital disorders due to teratogens are limited, but it is estimated that at least a million children are affected annually. Teratogens cause between five and ten percent of congenital disorders in industrialised countries, and 10%-15% in middle- and low-income nations. Reasons for the difference include increased frequencies of intrauterine infection, maternal malnutrition, lack of environmental protection policies, poorly regulated access to medication, poverty and lack of health care.[6]

Positive demographic and epidemiological transition increases longevity and thus the number of people reaching middle and old age. With this, as previously witnessed in industrialised nations, the burden of complex disorders is emerging as a public health challenge in middle-income nations. In 2005 an estimated 58 million people died, 35 million (61%) from cardiovascular disease, stroke, cancer and other complex disorders (Figure 1). Only a fifth of these deaths were in industrialised countries, the remaining 80 percent occurring in middle- and low-income nations.[14] Thus medical genetic services to assist in the care and prevention of complex disorders are being developed in industrialised countries and are a future reality for middle- and low-income nations.

FIGURE 1. ESTIMATED GLOBAL CAUSES OF 58 MILLION DEATHS IN 2005 (WHO)[14]



Health Needs Assessment

HNA is a systematic method for identifying unmet health, including health care, needs in a population and make changes to meet these unmet needs (Box 1). Health 'gain' is thereby achieved by identifying unmet needs and meeting them, reallocating funds from ineffective and inefficient services to unmet needs and improving poor services[1,2,4]

The process by which health needs assessment is carried out involves a combination of steps that are not necessarily linear, as comparisons between steps, and adjustments, will be necessary.[4] However all the components need to be completed and included before final analysis (Figure 3).

BOX 1. DEFINITIONS

A *health need* is a population's ability to benefit from an intervention or service. It is a function of both the prevalence of the problem under consideration and the effectiveness of the intervention(s) or services- health care, social, legal or policy development- available for the health need. The intervention or services may be one or combination of the above components. Problems and disorders which have no effective remedies, no matter how common, have no need, whilst low prevalence problems and disorders with available effective interventions are conceptualised as being problems of lesser need.[8,9] However, as a system develops and improves and health needs for common problems are met, less common problems with available effective interventions can become priorities.[1,2]

Health care needs are those that can benefit from health care interventions, whilst *health needs* additionally include changes to social & environmental factors that influence health, including socioeconomic status, education, pollution, diet, employment and behaviour. The former, health care needs, are more explicit than the latter being more open to measurement and intervention.[1,2]

Needs must be distinguished from *demands*. *Demands* are what patients want and ask for, but may not be regarded as being a need once the relevant analysis is carried out. Supply is the health care provided. Needs, demands and supply exist in an overlapping relationship (Figure 2). Assessing need is by comparison to assessing demand and supply difficult, but it should not be replaced by either or both.[1,2]

Figure 2. RELATIONSHIP BETWEEN NEEDS, DEMANDS & SUPPLY[15]

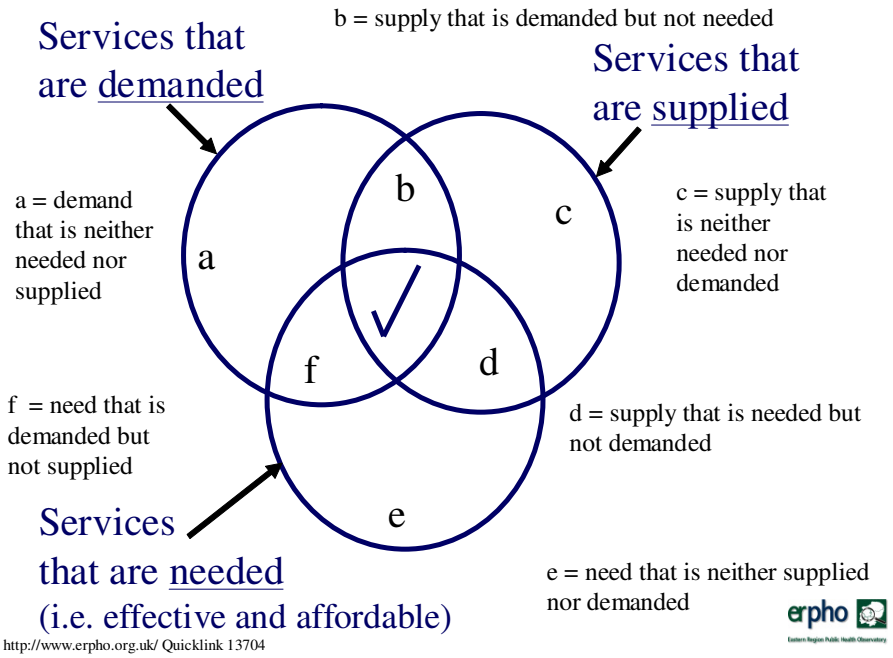
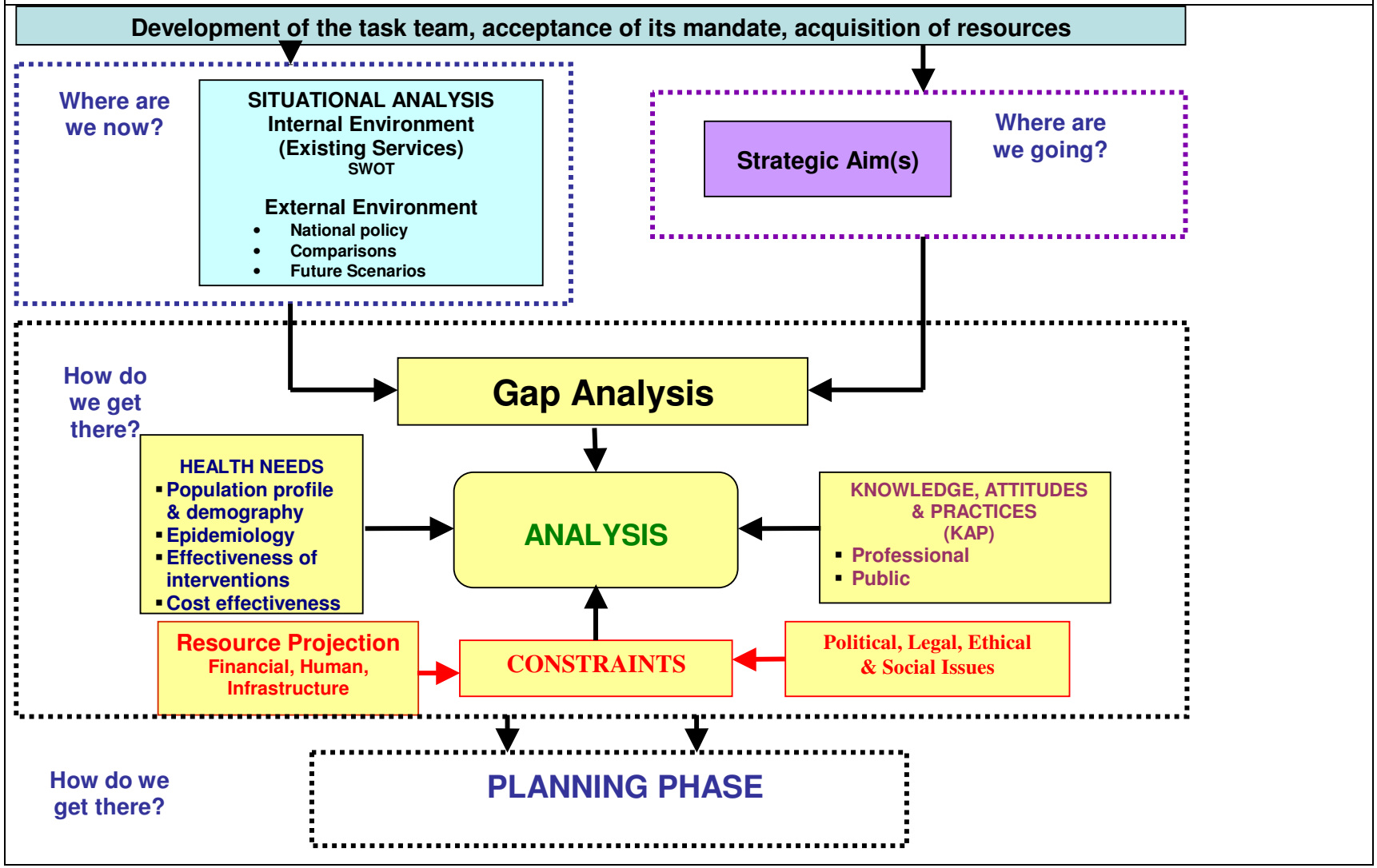


FIGURE 3. SCHEMATIC APPROACH TO HEALTH NEEDS ASSESSMENT



All projects have a beginning. This would be the proposal from the relevant authorities or organisation to undertake a HNA. The mandate for this would then be accepted by those tasked with leading the project, whose first responsibilities would be to choose a representative team with the necessary skills to undertake the HNA and ensure adequate resources are available to complete the task.[4] The task team should include appropriate professionals, including senior health managers, policy makers and planners, and public representatives. Its first undertaking would be to consider and clearly establish the strategic aim(s) of the HNA, ensuring compliance with their mandate. Doing this begs the question, "Where are we going?"

As HNA is about making changes that benefit the health of a defined population it is necessary to consider and understand the current situation in the country as it pertains to health. In essence doing a situational analysis to determine 'Where are we now?'

Establishing the internal environment of the population comprises obtaining an objective profile of existing health, including health care, services relevant to the topic under review. A baseline knowledge of existing services allows one to understand the current situation in order to consider what to change to and how. [2,16] Putting these services through a SWOT (strength, weaknesses, opportunities, threats) analysis will enhance the review.

The external environment is an appraisal of those issues that have moulded the internal environment to the present and which can be anticipated to impact the future, particularly changes that might be proposed to benefit health. National laws, policies, regulations and guidelines are relevant to the past, present and future of health and health care and need to be carefully considered. Comparing and contrasting services offered (the comparative approach) in different places is a powerful tool that informs what services are available, how they are meeting health and health care needs elsewhere, and if differences exist between them and the local existing service levels. They help identify deficiencies in provision of services. Finally an understanding of current and future knowledge and technology is necessary to anticipate future trends in the development of services that can benefit health in the given scenario. [2,4]

Comparing the current situation, the internal and external environment, and the strategic aims will enable an estimation of the 'gap' between them and is important in leading into the next phase, the collection of information for analysis, which helps chart the way forward, i.e. 'How do we get there?'

Essential for HNA is the determination of the health needs of the population. These are determined initially on consideration of the population's profile, demography and the epidemiology of the disorders under consideration-incidence (birth prevalence for congenital disorders), prevalence, mortality and morbidity, including subcategories of severity, which may require different

interventions or services. This will quantify the number of individuals that could benefit from a particular intervention. It is then crucial to establish the effectiveness[d] of available interventions or services for the disorders or health issues under consideration. No matter how common a problem, if there is no effective intervention there can be no benefit and thus no health need. Having established effectiveness of interventions it is then necessary to try and determine their cost-effectiveness, or the cost per unit benefit. [2,4,16]

To augment the assessment of existing services and obtain further insight into health needs the knowledge, attitudes and practices (KAP) of professionals and the public should be sought. This involves what has previously been referred to as the 'corporate approach'- the structured collection of knowledge, opinions, attitudes, and practices of stakeholders including health professionals (doctors, nurses, managers, policy makers, planners, service providers, experts) and lay people (patients, their families, the public, politicians and the press). This will be augmented by the professional and public representatives on the HNA task team. Obtaining and assessing public and professional knowledge, attitudes and practices may be subjective but used appropriately the information acquired is sensitive to local circumstances and embraces a large amount of knowledge and experience of individuals that would otherwise be ignored. In isolation it may emphasise demands rather than needs, it is open to personal, political and financial agendas but it is essential to democratise the process and assists justify decisions taken.[2,4,16]

The last data necessary before analysis is information on resources, human, infrastructural and financial, available to ensure the proposed health interventions and services, or changes thereof, can be achieved and are sustainable. Availability of resources, amongst other issues- political, ethical, legal and social- may comprise constraints that require careful consideration in the future development of health services.

The information gathered is now available for analysis. Its purpose is to formalise those disorders and factors that impact health and the effective, and if possible cost-effective, interventions available for them that will assure health benefit. This enables prioritisation considered by some, including health economists, to be a fundamental part of HNA. The process of prioritisation should be transparent, robust and logical and different models are available for consideration. Financial constraints, amongst others, are always present in health care and thus choices will always be a part of the initiation and development of health, and particularly health care, services.[1,4]

It should now be possible to formulate a rational plan to accomplish the project's strategic aim(s). A clear set of objectives compatible with the strategic aim(s), analysis and prioritisation should be listed with interventions or services

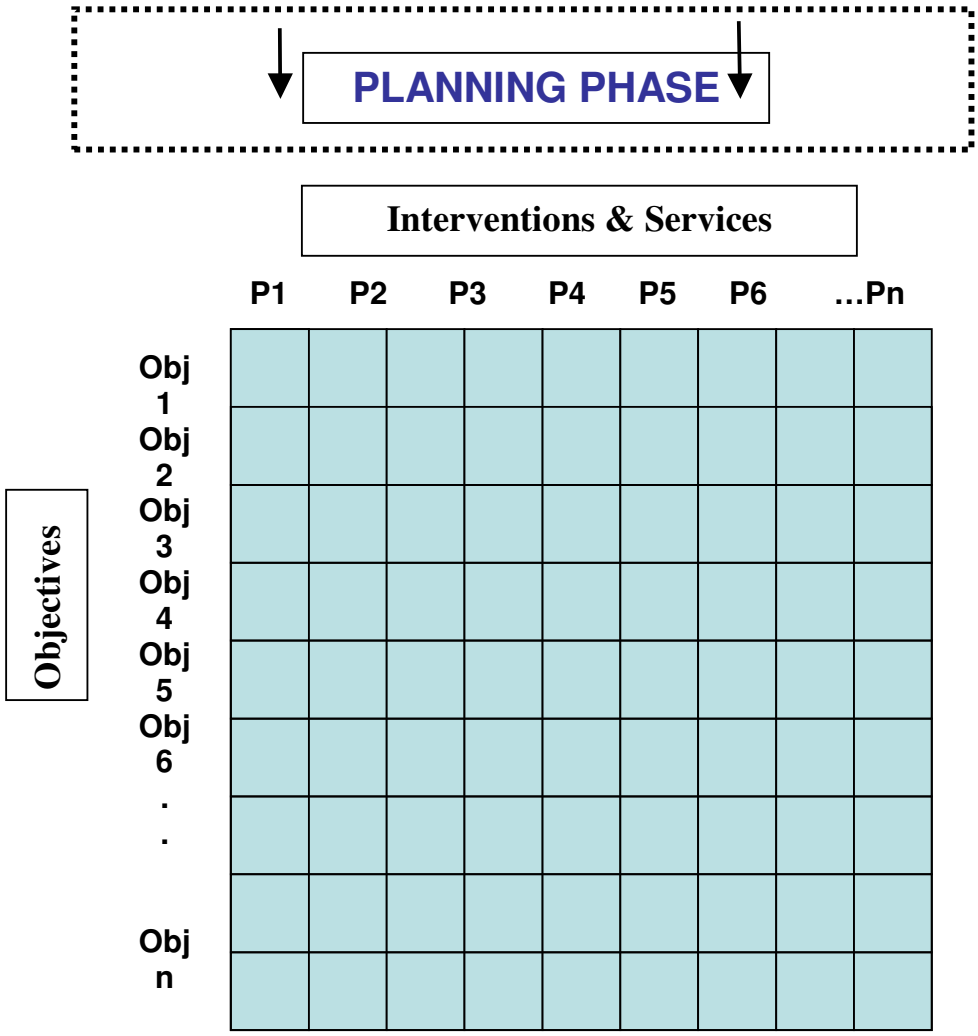
[d] Effectiveness is the degree to which possible improvements in health are actually attained.[17]

necessary to achieve them. These can be placed in matrices, a simple example of which is shown in Figure 4. Obviously certain of the interventions and services will be part of achieving more than one objective and visa versa.[3,4,16]

Setting the objectives and outlaying the strategies to achieve them also requires delegation of the interventions and services to those responsible for accomplishing them, ensuring that they are adequately trained and equipped to undertake the task and setting timelines for their achievement.

It is necessary in the planning phase to establish an audit process to evaluate both the ongoing progress and the outcome of the HNA. Indicators for both these processes should be set.[4] Piloting the plan, or parts thereof, may be considered before full implementation.

Figure 4. SCHEMATIC APPROACH TO HEALTH NEEDS ASSESSMENT (2)



Numerous challenges face those tasked with undertaking HNA but if successfully overcome they strengthen the health care system under consideration. The challenges include[1,4,16]:

- The involvement of professionals from different disciplines. This includes not only medical practitioners from different fields, but members of different professions including nursing, psychology, social welfare, senior health managers, policy makers and planners. In addition these professions have to work with public representatives. In this situation territorial imperatives can lead to disagreement and conflict and inhibit collaboration and information sharing.
- The variety of stakeholders participating can result in a diversity of definitions for the issues under review. A common language with simple, clear definitions of the issues needs to be established and used.
- Health professionals used to considering the benefit of individual patients in their daily work may have difficulty in adapting to the concept of developing interventions and services for a population.
- Accessing the wide array of data needed for an HNA can be difficult and time consuming.
- Critical to ensuring success of HNA is ensuring that whatever is planned has the acceptance of all stakeholders, particularly those that will ultimately have to implement and administrate the changes to the system. Managers are crucial in translating HNA findings into effective action.

Successful accomplishment of an HNA however also has benefits:

- The involvement of professions from different disciplines and background allows them to expand their vision, work as part of a team and develop partnerships and networks.
- In the process professional skills and experience are developed.
- Community involvement improves communication with the public and their understanding of issues involved. The democratisation of the process also improves the chances of public acceptance of the changes proposed and therefore success.
- The ultimate benefit is health gain and better use of resources.

Health Needs Assessment for Medial Genetic Services in Middle- and Low-Income Nations

The initial development of medical genetic services in industrialised nations occurred without the benefit of HNA as both it and many of the components from which it is derived, epidemiology and evidence for the effectiveness of interventions, were not available. However, examples of HNA for medical genetic services are now available, an example being the Oregon's Strategic Plan for Genetics and Public Health.[18]

Forty years later, despite HNA being formalised, and several middle-income nations having initiated medical genetic services, the use of HNA in developing these services has been limited. Potential barriers to its current use in the field of congenital disorders in middle-income countries are numerous. These include limited knowledge and understanding amongst many involved in developing medical genetic services of the process of health needs assessment; a supposed lack of epidemiological data; insufficient comprehension of the scope and availability of effective interventions for congenital disorders; and a perceived limitation of resources (human and financial) to deal and come to terms with the issues.

HNA should be a practical tool, available to most practitioners, not requiring great skill and needing only basic numeracy and common sense.[2,16] On this basis HNA for medical genetic services for congenital and complex disorders should be achievable in all middle- and low-income countries. Issues involved are discussed below.

The approach to HNA delineated above could be used with the following considerations.

1. Strategic Aim(s)

Each task team undertaking a HNA would need to develop its strategic aim(s) in accordance with its mandate and the circumstances of their country. Thus the strategic aim(s) would need to be developed with consideration of the country's internal and external environment.

2. Situational Analysis

2.1. Internal Environment

Universal medical genetic services, including clinical and diagnostic laboratory services, are limited in middle- and low-income countries. Several middle- and some low-income nations have components of medical genetic services for congenital disorders which although not universal are being expanded. All countries have women, reproductive, maternal and child health services that with appropriate orientation can be mobilised for the care and prevention of congenital disorders. These include family planning, antenatal care, infant and child health (paediatrics), surgery, especially paediatric surgery, and neuro-developmental therapies. The information on the resources, human, technological, and financial, clinical and laboratory, that are available to be coordinated and developed to undertake the development of medical genetic services should be available from different sources in each country.

Medical genetic services for complex disorders, in particular cancer genetic services, are a recent development in industrialised countries. No similar universally available services are presently known to be available in middle-or low-income nations.

2.2. External Environment

2.2.1. National policies

National laws, policies and guidelines are a matter of record and should be easily accessible.

2.2.2. Comparisons

Comparing and contrasting services offered in different places may initially serve a different function in developing medical genetic services in middle- and low-income nations as compared to industrialised countries. Rather than just comparing services between similar countries with well developed services to identify service gaps, as in industrialised nations, the comparison in middle- and low-income nations would be between industrialised nation services and existing middle- or low-income nation's services to evaluate what might be possible to implement in local circumstances to close the gap between the two. Comparisons of some components of medical genetic services between middle- and low-income nations with similar levels of service delivery are to a limited extent now possible and hopefully will become increasingly available in the future. In addition the existing national policy frameworks of different countries could be studied and compared to obtain direction from different national approaches to issues.

Differing from high-income countries, middle- and low-income nations will have to develop a strong base of clinical services in primary care. There is a little documented experience on how to achieve this and thus only limited scope for comparison. Comparison may however be undertaken by a corporate approach, obtaining advice from experienced colleagues in other countries.

2.2.3. Future scenarios

Genome based technology is advancing rapidly. Countries in a state of transition are also changing across many different parameters. Political, economic and social change will provide a backdrop and context for health services that will differ greatly, even in ten years, from existing conditions. It will be necessary to anticipate the range of scenarios that might affect the provision of all health services, but particularly clinical and laboratory medical genetic services when planning the development of services for the care and prevention of congenital and, in time, complex disorders.

3. Population profile and demography

People with congenital disorders are born into populations the profile of which, ethnic, religious, cultural, and socioeconomic, can influence birth prevalence and demography. Many middle- and low-income nations have populations that are heterogeneous. Determining their profile and demography is therefore essential.

4. Epidemiology

The equivalent of incidence for congenital disorders is birth prevalence, the number of individuals affected by a disorder (e.g. Down syndrome) or group of disorders (e.g. congenital heart defects or all congenital disorders) per 1000 live births. Population prevalence refers to the number of individuals affected by a disorder or group of disorders in a defined population at a given time. It is measured per 1000, 10 000 or 100 000 people, depending on the number of affected individuals.

Severe congenital disorders are a common cause of infant and childhood death, particularly in middle- and low-income countries. The difference between birth prevalence and population prevalence for individuals affected by a disorder or group of disorders, at a given time and defined population, is thus an indication of the mortality associated with the disorder.[6] Care services for individuals with a congenital disorder, if effective, will reduce the difference between birth and population prevalence, allowing the difference to be used in audit as an outcome indicator. This would, however, require a congenital disorder surveillance system with on-going follow-up by a registry. Decreasing birth prevalence can be used as an outcome indicator for prevention programmes.

Most middle- and low-income countries have limited empiric data on the epidemiology of congenital disorders. However, those middle-income countries presently involved in developing medical genetic services have, to varying degrees empiric data that can assist them in determining health need for congenital disorders.

The acquisition of epidemiological data is costly and time consuming. Its lack or deficiency in middle- and low-income countries need however not result in avoidance of the process of HNA or the implementation of medical genetic services on a less objective basis. Recently, modelled global congenital disorder birth prevalence data, presented on a country by country basis, has been published in the form of the Modell Birth Defects Database.[6]

Based on the understanding that most genetic congenital disorders have similar birth prevalence globally, the work of Baird et al in British Columbia and Czeizel with the Hungarian Congenital Malformation Registry was used to obtain baseline congenital disorder estimates. These were then adjusted to account for

circumstances known to alter the birth prevalence of congenital disorders in particular countries or regions of the world (Box 2).[6]

Box 2. Factors responsible for affecting birth prevalence of congenital disorders.[6]

Malaria. Carriers of haemoglobin disorders (sickle cell anaemia & thalassaemia) and glucose-6-phosphate dehydrogenase deficiency genes have a survival advantage in avoiding the lethal effects of malaria. Therefore, they selectively live to reproductive age in malaria endemic regions having children and passing on the gene. Over generations the frequencies of these genes in these populations have increased, simultaneously raising the birth prevalence of the disorders.

Migration. People moving to different countries and regions embed their single gene defects into the populations they enter. E.g. sickle cell anaemia spread from Africa to the Americas and the Caribbean by the slave trade, and porphyria to South Africa with the emigration of the Dutch for trading purposes in the 17th century.

Migration includes urbanisation - the movement of people from rural, traditional lifestyles to towns and cities. This puts them at risk of common disorders with a genetic predisposition and exposure to teratogens, particularly alcohol, increasing the likelihood of fetal alcohol syndrome.

Parental consanguineous marriage. This social custom, involving the marriage of cousins or uncles and nieces, is accepted by at least 20 percent of the world's population. It increases the birth prevalence of autosomal recessive genetic/congenital disorders, almost doubling the risk of neonatal and childhood death, mental retardation and serious genetic/congenital disorders.

Advanced maternal age. Advanced maternal age (>35 years old) is associated with an increased birth prevalence of chromosomal trisomies, particularly Down syndrome. In middle- and low-income countries, a high percentage of women conceive over the age of 35 years without the availability of community education and universally available and accessible family planning services, medical genetic screening, prenatal diagnosis and associated services. The birth prevalence of chromosomal abnormalities is therefore high in these countries.

Personal Poverty. Reduced socio-economic circumstances are associated with an increased birth prevalence of genetic/congenital disorders. Poverty means many mothers are malnourished, before and during pregnancy, and are at greater risk of exposure to environmental teratogens such as alcohol and maternal infection.

Level of health care. The birth prevalence of genetic/congenital disorders is influenced by the national level of available and accessible health care, especially services for woman,

reproductive (including family planning) and maternal health. These health care services are capable of preventing Down syndrome, common single gene defects, neural tube defects and congenital malformations, fetal alcohol syndrome, congenital syphilis, iodine deficiency disorder and congenital rubella syndrome.

The availability and accessibility of neonatal (newborn) and child health care services influences the infant and childhood mortality (death) rates, including those for infants and children with genetic/congenital disorders, thus affecting the population prevalence of genetic/congenital disorders.

The Modell Birth Defects Database and the recommendations stemming from its analysis were endorsed by an Expert Committee of the WHO's Human Genetic Programme which met in May 2006.[6,12] Using the Database it is considered possible to establish the common genetic congenital disorders in a country.

Common genetic congenital disorders include the haemoglobin disorders and glucose-6-phosphate dehydrogenase deficiency in tropical countries where malaria was endemic and consanguinity commonly practised. Down syndrome, neural tube defects and congenital heart defects are common in most middle-and low-income nations and occulocutaneous albinism is prevalent in sub-Saharan Africa. In countries where consanguinity is high inborn errors of metabolism are also frequent.[6]

Non-genetic congenital disorders consequent on abnormalities of the fetal environmental, particularly teratogens, account for 10-15% of the total birth prevalence of congenital disorders in middle- and low-income countries. Disorders of particular significance, depending on the country, are congenital syphilis, congenital iodine deficiency disorder, fetal alcohol syndrome and congenital rubella syndrome (CRS). Data on the first two may be available in each country. On average the annual birth prevalence of CRS can be estimated as approximately 0.58/1000 live births, if immunisation programmes are absent. Only one middle- or low-income country, South Africa, has accurate data on the population prevalence of FAS. If suspected as a problem, epidemiological studies need to be done, and FAS may then be considered as a future priority, depending on the outcome of this work. [6,9]

It is proposed that epidemiological data from the Modell Birth Defects Database, supplemented by national empiric data, can be used with information on the effectiveness of relevant interventions and services to develop a country's initial health need profile for the care and prevention of congenital disorders. The birth prevalence figures in the Database should be used as 'expected' birth prevalence estimates.[6] This would be a starting point. HNA should be a continuing and repetitive process to enable on-going health improvement and it is a prerequisite of that process to establish evaluation and monitoring systems- including birth defect surveillance systems and registries- to ensure empiric

epidemiological data will be obtained for future development of comprehensive medical genetic services.[1,4]

5. Effectiveness and Cost effectiveness

A range of effective interventions and services for the care and prevention of common congenital disorders that are applicable in middle- and low-income nations has been documented.[6,7,9-11] Many others that may not presently have priority but are available for when the health need presents itself.

The costs of care for haemoglobin disorders, thalassaemia and sickle cell disorder, have been calculated for middle- or low-income countries.[10,19] The principles associated with the development of these costs are applicable to other disorders. Cost-effectiveness of these interventions, in the context of middle-and low-income nations, has still to be confronted.

However, in this regard, a simple set of principles derived from Iran's experience with establishing services for care and prevention of thalassaemia, apply. The cost of care for some congenital disorders, especially those requiring lifelong medical treatment (e.g. thalassaemia, haemophilia), can be high and thus not cost-effective in isolation. For chronic conditions, like many of the congenital disorders, care that saves lives prolongs it and the cost of that care continues through that prolong life. Consequently the national cumulative cost of care increases incrementally year on year as more people with the disorder are born and live. In circumstances of finite resources this can eventually affect the sustainability of the service. In this situation the only way to achieve sustainability is to successfully implement comprehensive prevention services. This is in line with the aims of medical genetic services in which patient care and prevention are not alternatives, but are complementary and inseparable aspects of the service for individuals with a congenital disorder, their families and the community. When combined with effective prevention the overall service for care and prevention of a particular disorder can be cost- effective.[6,9,20]

Surgery is an important and cost-effective form of treatment for many congenital malformations. Examples of surgery that can save lives or significantly reduce morbidity includes that for congenital heart defects, cleft lip and palate, clubfoot, gut and urogenital abnormalities.[6,9] Preventive interventions that in isolation are effective and are considered cost-effective include family planning (contraception services), the iodinisation of salt to prevent iodine deficiency disorder and fortification of a staple food with folic acid to prevent neural tube defects.[6]

6. Knowledge, Attitudes and Practices of Professionals and the Public

KAP assessment as discussed above has disadvantages in that it is open to political, financial and individual agendas but is essential in that it democratises

the process. It also has the potential to realise a large amount of knowledge and expertise.

Professional opinion needs to be obtained from 2 groups. Medical professionals include medical geneticists, obstetricians, paediatricians, surgeons, public and primary health care practitioners, genetic counsellors, nursing staff and speech, occupational and physiotherapists. Their involvement has the potential to make available large amounts of knowledge and accumulated expertise that would be significant in the initial phases of medical genetic service development when empiric data may be limited. Invaluable assistance is also available through consultation with experts in the development of medical genetic services from other countries.

The involvement of senior health policy makers, planners and managers is necessary as whatever is decided upon has to be grounded in, amongst other issues, that which is politically and managerially possible for the health care system. To ignore them is to invite failure.

Obtaining public opinion may be an involved process, and should include not only consumers but also the community at large. Consumers can be accessed through patient/parent support groups, if these are available. The public can be accessed with a range of tools including citizen's juries, user consultation panels, focus groups, questionnaire surveys, opinion surveys of standing panels and particularly in middle- and low-income nations community appraisals.[21,22]

7. Constraints

Constraints, financial, social, legal, political and possibly others are a feature of all health systems and therefore a 'filter' that all the components of HNA have to negotiate before analysis and prioritisation can lead to effective planning.

8. Planning.

Health service planning is the ultimate goal of HNA, using the information obtained from the analysis and then prioritisation to set objectives with rational interventions and services to meet the strategic aims. Below in an example of a set of objectives, interventions and services that might be derived for the development of medical genetic services for the care and prevention of congenital disorders.

OBJECTIVES	INTERVENTIONS & SERVICES
1. **To recognise, diagnose, treat, manage and rehabilitate patients with genetic/congenital disorders**	1. **To establish and provide educational programmes for the general population**
2. **To counsel and support patients with genetic/congenital disorders and their families**	2. **To establish and provide training programmes for health professionals**
3. **To promote the reduction of maternal age and knowledge of the impact of consanguinity on genetic/congenital disorders**	3. **To establish and provide services for the management of patients with genetic/congenital disorders including counselling and support for them and their families**
4. **To improve maternal nutrition**	4. **To establish and provide obstetric services including specific programmes for the screening of maternal infections, fetal abnormalities and the detection of rhesus incompatibility**
5. **To reduce maternal exposure to infections, environmental teratogens, drugs and alcohol**	5. **To establish and provide appropriate services for the screening of inherited and heritable disorders in the neonate**
6. **To reduce obstetric causes of genetic/congenital disorders**	6. **To establish and provide molecular, biochemical and cytogenetic laboratory services**
7. **To enable parents to exercise reproductive choices that are consistent with their wishes and in the context of the culture in which they live**	7. **To establish a programme for the promotion of maternal health**

Conclusion

HNA is a systematic approach for planning and providing cogent health services in a setting of finite resources, epidemiological transition, technological advance and changing public expectations. All of these circumstances apply to medical genetic services in middle- and low-income nations, even more so than in industrialised countries. However, middle- and low-income countries seeking to develop medical genetic services are not, to the best of our knowledge, using formal HNA to inform their efforts. It is proposed that further assistance can be made available to middle- and low-income nations for their efforts to undertake HNA by developing a HNA 'toolkit'. This process has begun and financing to pilot the toolkit in different countries is being sought.

Significant challenges face the development of medical genetic services in middle- and low-income nations. Because of their stage in epidemiological

transition their major medical genetic health needs will presently involve mainly congenital disorders. HNA can assist these countries determine and prioritise their medical genetic health needs and establish well organised and structured medical genetic services, both clinical and laboratory, appropriate to their needs and resources.

Successful implementation of a health service plan derived by HNA, as described above, should not be seen as an endpoint. It will change the internal environment of the population, whilst epidemiological transition, technological advances, changing public expectations and demands, resource limitations and the rising cost of health care remain continuing influences on the ever changing external environment. If HNA is to have a purpose the successful achievement of its strategic aim(s) should be seen as the first step in an on-going and cyclical process of HNA for continuing health gain.

REFERENCES

1. Wright J, Williams R, Wilkinson JR. Development and importance of health needs assessment. BMJ 1998; 316: 1319-1313.
2. Stevens A, Mant J, Raftery J, Simpson S. Health Care Needs Assessment. The epidemiological based needs assessment reviews (Volume 1) 2nd edition. Radcliffe Publishing, UK.2004.
3. Smith P, Haggard S. Planning for a single speciality in a Health District. J Operational Research Society 1982; 33: 29-39.
4. Health needs assessment: A practical guide. National Institute for Health & Clinical Excellence. 2005. http://www.nice.org.uk/media/150/35/Health_Needs_Assessment_A_Practical_Guide.pdf
5. WHO.Human genetics & public health. Technical Report Series No. 282. Geneva, Switzerland: World Health Organization, Geneva, Switzerland. 1964.
6. Christianson AL, Howson CP, Modell B. March of Dimes Global Report on Birth Defects: the hidden toll of dying and disabled children. March of Dimes Birth Defects Foundation, White Plains, New York: March of Dimes. 2006.
7. WHO/ World Alliance for the Prevention of Birth Defects. Services for the prevention and management of genetic disorders and birth defects in developing countries. Geneva, Switzerland: World Health Organization, 1999.
8. WHO. Community approaches to the control of hereditary diseases. Report of an advisory group. Geneva, Switzerland: World Health Organization, 1985.

9. Christianson AL, Modell BM. Medical genetics in developing countries. Annual Reviews of Genomics and Human Genetics 2004; 5: 219-265. [Available on-line at http://www.annualreviews.org]
10. Alwan A, Modell B. 1997. Community control of genetic and congenital disorders. Eastern Mediterranean Regional Office Technical Publication Series 24, 1997. WHO: Regional Office of the Eastern Mediterranean, Alexandria, Egypt.
11. WHO. Primary health care approaches for prevention and control of congenital and genetic disorders. Geneva, Switzerland: World Health Organization, 2000.
12. WHO & March of Dimes. Management of birth defects and haemoglobin disorders. Geneva, Switzerland: World Health Organization, Geneva, Switzerland. 2006.
13. WHO. Birth Defects. Report by the Secretariat for the 125th session of the Executive Board. Geneva, Switzerland: World Health Organization, 2009. [www.who.int/gb/ebwha/pdf_files/EB125_7-en.pdf] (Accessed 11th June 2009.)
14. WHO, Global Alliance Against Chronic Respiratory Disease. The burden of chronic diseases. WHO, Geneva, Switzerland. 2005
15. Eastern Region Public Health Observatories. What is Health Needs Assessment. http://www.erpho.org.uk/viewResource.aspx?id=13704 (Accessed 31 January 2009)
16. Stevens A, Gillam S. Needs assessment: from theory to practice. BMJ 1998; 316: 1448-1452.
17. Donnabedian A. An introduction to quality assurance in health care. Oxford University Press Inc. New York. 2003.
18. Silvey K, Newell A. Oregon's Strategic Plan for Genetics and Public Health. Oregon Department of Human Services. 2002.
19. WHO. Guidelines for the control of haemoglobin disorders. WHO, Geneva, Switzerland. 1994.
20. Samavat A, Modell BM. Iranian national thalassaemia screening programme. BMJ 2004; 329: 1134-1137.
21. Jordan J, Dowswell T, Harrison S, Lilford R, Mort M. Whose priority? Listening to users and the public. BMJ 1998; 316: 1668-1670.
22. Wright J, Walley J. Assessing health needs in developing countries. BMJ 1998; 316: 1819-1823.