phg foundation
making science work for health

Born healthy:
launching the health needs assessment toolkit for congenital disorders

Congenital Disorders

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The PHG Foundation is the working name of the Foundation for Genomics and Population Health, a charitable organisation (registered in England and Wales, charity no. 1118664; company no. 5823194) which works with partners to achieve better health through the responsible and evidence-based application of biomedical science.

Born healthy - a framework for action

The Born Healthy programme creates a framework and a freely available health needs assessment (HNA) Toolkit to help countries combat the causes and effects of congenital disorders. This brief report provides a summary of our workshop in June where feedback from groups piloting the HNA Toolkit was presented.

A global problem

Congenital disorders (or birth defects) are structural or functional abnormalities that are present from birth. Many middle-income countries are becoming more successful at combating overall rates of perinatal, neonatal and infant mortality yet deaths attributable to congenital disorders have not shown a comparable fall, despite the availability of effective care or preventive interventions for many conditions. In low-income countries, these conditions have been largely ignored in the drive to tackle diarrhoeal diseases, TB, HIV/AIDS and malaria.

Born Healthy is the PHG Foundation's response to the World Health Assembly 2010 resolution which recognises the significance of birth defects as a cause of stillbirths and neonatal mortality.

Why is progress slow?

One of the main constraints on developing services for congenital disorders is lack of evidence of need, particularly when up against other health priorities.

Opening the workshop, Severin von Xylander (WHO) recalled the World Health Assembly resolution that "*recognised the importance of birth defects as a cause of still births and neonatal mortality*" and their exhortation to countries to establish and strengthen monitoring and evaluation systems in order to inform decision-making.

Progress in tackling congenital disorders, he argued, can be achieved where services related to their care and prevention are integrated into the continuum of reproductive, maternal, neonatal and child health, and incorporated into national health plans that have sustained funding.

Echoing the points made by Severin, Chris Howson (from the March of Dimes) told the audience that countries are reporting little progress in assembling the data necessary to build a picture of what effective actions can be taken. Leadership is needed, he urged, to mobilise people, create local champions and build a peer network - all of which will help to create the conditions that will move congenital disorders up the agenda.

The Born Healthy programme creates a framework and a freely available Health Needs Assessment (HNA) Toolkit, to help ministries and others at national and sub-national levels assess health needs in relation to congenital disorders in their own countries, territories or regions.

The HNA Toolkit can aid the preparation of a rational, evidence-based case for the development of effective services for the care and prevention of these conditions. Users are also enabled to select priorities and implement appropriate actions to address identified needs.

We launched the HNA Toolkit and the Born Healthy programme at an international workshop and conference in London on 27-29 June 2011.

If all countries were able to implement interventions as effective as those available in high-income countries, the lives of about 2.5 million children under five could be saved each year.

Aims of the workshop

The HNA Toolkit lead developer Luis Nacul set out the aims of the three day event:

- To assess whether the HNA Toolkit is fit for purpose
- To identify any changes that might be necessary
- Subject to these changes, to approve adoption of the HNA Toolkit
- To consider the potential for using the HNA Toolkit in different countries or regions
- To explore synergies with other relevant international programmes
- To identify ways of creating a Born Healthy 'community of interest' to take the project forward.

Key to the success of the health needs assessment process as a support for strategic planning is a team of major stakeholders whose involvement is crucial to achieving effective change. The team works together to identify action points and assign priorities to those actions.

To aid this process the HNA Toolkit is linked to a specific prioritisation tool which includes identifying milestones and criteria for evaluation.

The HNA Toolkit was found to be a simple and effective tool, enabling users to take a systematic approach to evaluating key features of services and to plan and prioritise cost-effective interventions.

About the HNA Toolkit

The health needs assessment (HNA) Toolkit is a step-by-step guide to enable the user to prioritise and select topics (which may be specific congenital conditions, or services for the care and prevention of congenital disorders, or both) relevant for their country or region, and to answer the questions: Where are we now? Where do we want to be? How do we get there?

The HNA Toolkit guides the user through a systematic assessment of existing health policies, services and interventions, and a process for identifying gaps and areas for improvement. It provides background information on a range of congenital disorders and services and, for each one, evidence of effectiveness of preventive options. There is a linked 'tool and calculator' for the user to generate estimates of the burden of disease and the potential for reducing that burden by appropriate interventions.

The calculator spreadsheets are populated with indicative demographic and epidemiological data for a range of countries but these data can be replaced or supplemented by local data where available.

The HNA Toolkit was developed over two years by the PHG Foundation with support from the Centre for Health Informatics and Multi-professional Education (CHIME) at University College London. Epidemiological data used in the Toolkit were derived from the Modell Database of Constitutional Congenital Disorders (MGDB), developed by Professor Bernadette Modell and colleagues from CHIME.

Evaluating the HNA Toolkit materials

A half-day workshop was devoted to evaluating the materials for nine HNA Toolkit topics:

- Chromosomal disorders
- Neural tube defects
- Orofacial clefts
- Teratogens
- Congenital syphilis
- Preconception care and screening
- Prenatal care and screening
- Newborn care and screening, and
- Medical genetics services.

In general the materials were considered to be of high quality and fit for purpose. Suggested changes are being incorporated into the updated chapters, the first seven of which went live on the Born Healthy website in December 2011.

A substantially revised version of the chapter on medical genetics services will extend its scope to include the full range of services involved in the care and prevention of congenital disorders, which are included in the responsibilities of many genetics services worldwide.

Not only does it enable health needs to be identified and actions prioritised for specific conditions and services, but it brings together a wide range of stakeholders to forge new alliances, engage governments and policy makers, and create change.

Reports from the pilots

During 2010, a prototype HNA Toolkit was piloted by three countries in Latin America: Argentina, Uruguay and Brazil. Brazil and Argentina ran two pilots. The workshop was an opportunity for feedback on the HNA Toolkit.

Piloting the HNA Toolkit

All three countries who piloted the HNA Toolkit have well-established genetics services, newborn screening programmes, birth defects registries, and universal access to prenatal care. They have a growing middle class, increasing life expectancy and decreasing infant mortality. However there continues to be a high degree of socioeconomic inequality, with low levels of education among the poorest people, and health services that are uneven in accessibility and quality, especially in rural areas.

The workshop was an opportunity to hear from the pilot groups how they used the HNA Toolkit, what they learned about the process and about congenital disorders in their region.

Each group selected a number of topics from the HNA Toolkit, basing their choice on their experience of needs in the region in which they work.

A powerful and systematic tool that effectively demonstrates the impact of congenital disorders and acts as a catalyst to bring people together.

Mariela Larrandaburu, Ministry of Public Health, Uruguay

In Uruguay, the pilot focused on newborn screening (NBS) and medical genetics services.

Conclusions from the HNA exercise were that the newborn screening programme is generally strong, but could be improved by:

- Developing a standardised protocol for newborn physical examination
- Establishing an epidemiological surveillance programme linked to the NBS programme
- Strengthening the national registration system for birth defects
- Legislating to mandate welfare benefits for affected infants identified by screening
- Evaluating the implications of expanding the screening panel for metabolic diseases.

The assessment of genetics services showed that these services were very weak in the public sector, were essentially unregulated, and were entirely concentrated in Montevideo, the national capital.

The team identified as weaknesses the HNA Toolkit's lack of local and regional data and the time-consuming nature of the HNA process.

Overall, the team from Uruguay found that the HNA Toolkit is a powerful, systematic tool to effectively demonstrate the impact of congenital disorders and acts as a catalyst for bringing people together from across disciplines and regions to address shared problems.

Maria Teresa Sanseverino, Hospital de Clinicas de Porto Alegre, Brazil

The first pilot team in Brazil considered preconception care and screening, and newborn screening.

In preconception care, the HNA process spotlighted current strengths in rubella immunisation and flour fortification, and an effective teratogen information service. However, it showed that policies and interventions for other preventable conditions such as fetal alcohol syndrome are lacking.

Actions resulting from the HNA process included a pilot initiative on preconception care in a primary care unit of a hospital in Porto Alegre. A questionnaire was also sent to all Brazilian clinical geneticists to assess current knowledge and practice, with a view to developing an effective community based service.

The needs assessment on newborn screening identified an effective bloodspot screening programme but a need to develop better services for newborn hearing screening and physical examination.

The team were impressed by the systematic approach adopted in the HNA Toolkit and its usefulness in identifying and prioritising needs and actions.

Lavinia Schuler-Faccini, Hospital de Clinicas de Porto Alegre, Brazil

A second Brazilian experience described using the HNA Toolkit for health needs assessment in relation to neural tube defects (NTD), sickle cell disease, and fetal alcohol syndrome. These conditions were chosen because of the availability of effective interventions, and because social inequalities play a role in both prevalence and in outcomes.

For NTD, conclusions from the HNA were that these conditions are under-reported due to lack of effective training in newborn physical examination, and that the current level of folic acid flour fortification is likely to be sub-optimal.

Sickle cell disease was found to be more prevalent than suggested by epidemiological modelled data, suggesting that skin colour is not a good indicator of risk and the condition should be considered as a general, national issue rather than an ethnic problem.

The HNA exercise for fetal alcohol syndrome highlighted a serious lack of information on this condition and a lack of any effective health programmes, despite evidence of widespread risky drinking behaviour in women of reproductive age.

Lavinia also described the potential for modifying the HNA Toolkit for other applications. As an example, it is being developed for use in relation to the teratogenic effects of thalidomide, currently used as a treatment for leprosy in Brazil.

The HNA Toolkit can be used to estimate risk, to plan strategies to prevent exposure, and to monitor the effectiveness of these strategies.

Boris Groisman, National Ministry of Health, Argentina

This team selected neural tube defects (NTD) and orofacial clefts (OFC) as the two topics to pilot because together these disorders represent a significant burden of mortality and disability, yet effective preventive and/or therapeutic interventions exist. The team were also able to exploit existing links with key stakeholders who were invited to participate in the HNA process.

With the HNA Toolkit Boris and his team identified gaps in services and actions to address these, including:

- Inclusion of congenital disorders in the national Plan Nacer which covers pregnant women and children up to the age of six
- Improvements in genetics services and professional education; improved treatment for OFCs
- Preparation of information sheets for affected families
- Lobbying for legalisation of pregnancy termination in cases of severe abnormality.

The team summed up specific strengths of the HNA Toolkit as

- Its comprehensive scope
- The different types of questions enabling both qualitative and quantitative information to be used
- The prompts to note any gaps and inadequacies in the available data
- The encouragement to build national and international links and collaborations.

A broad range of government ministries, hospitals, specialist clinicians and patient support groups were engaged and motivated by their participation in the HNA process.

Maria Paz Bidondo, Garrahan Hospital, Buenos Aires, Argentina

Another way of using the HNA Toolkit is to compare health needs in relation to congenital disorders in one region with those in another region. The provinces of Chaco and Nequén in Argentina differ markedly in key socioeconomic and health service indicators.

The topics chosen for comparison were health services, congenital heart disease, chromosomal disorders, and prenatal care and screening.

The team in Buenos Aires also used the prioritisation software that can be used with the HNA Toolkit.

The HNA Working Group, coordinated by a team from Garrahan Hospital, included representatives from hospitals in the two provinces, as well as parents associations, community leaders and educators.

Using the HNA Toolkit Maria Paz and the team were able to:

- Highlight major differences between the two provinces, particularly the need to develop genetics services and prenatal screening in Chaco province
- Make new connections with patient support organisations, and with the National Programme for Congenital Heart Disease, enabling changes and new interventions to be suggested at both regional and national levels.

As a direct result of the evidence collected using the HNA Toolkit, a cytogeneticist has now been employed to help improve services in Chaco.

The global policy dimension

In addition to sessions devoted to practical evaluation of the HNA Toolkit, the conference also considered current perspectives on the global burden of congenital disorders and on efforts to address the problem in different world regions.

Epidemiology: reviewing the evidence base

Arnold Christianson (University of the Witwatersrand, Johannesburg) and Matthew Darlison (University College London) discussed current epidemiological evidence on congenital disorders. Sources of information included estimates by the WHO's Child Health Epidemiology Reference Group (2005) and the Global Burden of Diseases (GBD) Congenital Disorder Expert Group (2010). There are differences between these estimates depending on whether the underlying cause of death – which may often be a congenital condition – is taken into account.

In 2010 the GBD Congenital Disorder Expert Group attributed nearly 30% of all deaths in the under-5 age group to congenital disorders. If we benchmark the current under-5 mortality ratio in different world regions to Millennium Development Goal 4 (MDG4) targets we find that, in many regions, achievement of MDG4 would require the complete elimination of all cases of under-5 deaths other than congenital and genetic disorders. Unless action is taken on these disorders, MDG4 is simply not feasible.

An important aspect of the evidence base is information on the survival of infants born with congenital disorders, and an understanding of the factors that predict survival rates.

Judith Rankin (University of Newcastle, UK) presented findings from a study by the Northern Congenital Abnormality Survey (NorCAS), which collects data on all congenital anomalies occurring in the north of England.

Between 1985 and 2003, overall 10-year survival rates for liveborn individuals with congenital abnormalities improved from 80% to 93%. Of course, survival rates for different types and numbers of anomalies vary greatly. For example, one-year survival for babies born with cleft palates was 99.3%, but no babies born with anencephaly survived their first year.

Data from the study apply to a high-income country with developed health services, including access to prenatal diagnosis and termination of pregnancy, but provide a useful indicator of future trends and possibilities for countries developing services for prevention, care and treatment of congenital disorders.

International perspectives on the care and prevention of congenital disorders

Experts presented a snapshot of current efforts to develop policies and services for the care and prevention of congenital disorders in different countries and regions.

Gustavo Giachetto (Ministry of Public Health, Uruguay) drew attention to the establishment of a National Registry of Birth Defects and a centralised information system for epidemiological surveillance in Uruguay.

Victor Penchaszadeh painted a picture of highly variable service provision throughout the 37 countries of Latin America. Provision tends to be focused in urban areas and genetics services tend not to be integrated. However, positive moves forward can be seen in inititatives such as the Latin American Collaborative Study of Congenital Malformations, which registers and describes all congenital malformations in liveborn infants born in a network of participating maternity hospitals across South America.

Of the 136 million annual births worldwide, 68 million are in Asia. Carmencita Padilla (University of the Philippines, Manila) described the development of newborn screening services in the region, where coverage ranges from 99% in the richer countries to less than 1% in the poorest.

In an informal survey of colleagues from countries with widely varying levels of socioeconomic development, Carmencita found that overall health service provision for congenital disorders is also highly variable across the Asian region.

Laos, for example, has no data or systems for registration of congenital disorders and no dedicated governmental support, while Singapore, where congenital disorders are the main cause of infant mortality, has a thalassaemia prevention programme, focused screening programmes for specific conditions, and a national birth defects registry.

Patient involvement

There is ample evidence that involving patients in the planning and development of healthcare leads to improvements in the quality, responsiveness and accessibility of services.

Celine Lewis (Genetic Alliance UK) described some current examples of patient involvement in improving services for congenital disorders in low and middle income countries, where patients have often been regarded as passive recipients of healthcare rather than active stakeholders.

The South African Inherited Disorders Association, a patient/parent group, has helped to drive the agenda of primary genetic healthcare, particularly in deprived rural areas, by training over 300 nurses to recognise potential causes of congenital disorders, to advise and assist pregnant women and, where necessary, arrange referral.

In Brazil, the Brazilian Fragile X Association promotes professional and public awareness of the condition and supports families seeking diagnosis and care.

In Europe, the EU-funded EuroGenTest project included development of appropriate information on genetic testing for patients in a range of European countries with different languages, cultures and legal constraints.

Education

Jhpiego is a non-profit organisation funded by USAID, international donors and private foundations to help some of the poorest countries of the world improve the health of women and children by focusing on reproductive health and family planning, maternal/ newborn health and infectious disease control.

Patricia Gomez (Johns Hopkins University, US) offered some pertinent lessons for Born Healthy based on Jhpiego's 38 years experience as a community of interest in the international health arena.

It is vital, she urged, to influence the curricula for medical and midwifery education, to develop in-country networks to maintain quality and performance of healthcare programmes, and to evaluate and share results in national and international fora.

While government policy, the establishment of registries and the rolling out of national programmes such as newborn screening and flour fortification are clearly important, all countries represented at this workshop session identified education of both health professionals and the public as paramount in efforts to improve services.

Where next?

On the final day of the workshop, there was a group discussion of the strengths, weaknesses, opportunities and threats for the future uptake and use of the HNA Toolkit.

Strengths

Its scope and systematic, inclusive approach to health needs assessment and to the planning and prioritisation of actions; the inclusion of questions enabling both qualitative and quantitative information to be used; the prompts to note any gaps and inadequacies in the available data.

Perhaps most significantly, its encouragement of collaborations.

Weaknesses

A lack of an explicit process for choosing HNA topics, and some lack of clarity about the intended audience; lack of regional data and time consuming nature of the process.

Opportunities

Efforts should be made to link the Toolkit to existing major health programmes (for example, rubella vaccination or programmes to combat sexually transmitted diseases such as syphilis). This could be particularly beneficial in countries that lack the infrastructure or resources for specific health programmes devoted to congenital disorders.

Threats

The challenge of providing accurate epidemiological information; competition for resources.

There is clearly enormous potential for taking the HNA Toolkit to other countries.

The Philippines were reported as ready to undertake a pilot project, targeting the country's two main islands and starting with the topics of health services for congenital disorders, and neural tube defects. The aim will be to present results to a human genetics conference in 2013, to convince others in the Asia-Pacific region of the HNA Toolkit's usefulness.

In India, the regional office of WHO has expressed interest, and funding might be available from the Ministry of Research. An initial task will be to identify key people who need to be involved in the project, with a view to convening a meeting later in 2011.

As ever, funding remains a major constraint. The PHG Foundation will continue to fund core staff time and travel for development of the Toolkit and is willing to support and facilitate bids for other sources of funding, but does not necessarily have the experience to lead such bids.

Other groups that might be involved in, or consulted about, an FP7 bid include the GenTEE (Genetic Testing in Emerging Economies) group.

Several of the conference participants have also participated in EU-funded projects and suggested they might be able to contribute expertise.

Acknowledgements

Our thanks go to all workshop participants and the invited speakers for their time and expertise.

Guest speakers

Maria Paz Bidondo
Garrahan Hospital, Argentina

Arnold Christianson,
University of the Witwatersrand, South Africa

Matthew Darlison
University College London, UK

Gustavo Giachetto
Ministry of Public Health, Uruguay

Roberto Giugliani
Hospital de Clincas de Porto Alegre, Brazil

Patricia Gomez
Johns Hopkins University, US

Boris Groisman
National Ministry of Health, Argentina

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March of Dimes, US

Mariela Larrandaburu
Ministry of Public Health, Uruguay

Celine Lewis
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About the PHG Foundation

The PHG Foundation is an independent, non-profit organisation based in Cambridge, UK, with the mission making science work for health. We identify the best opportunities for 21st century genomic and biomedical science to improve health and tackle disease in ways that are rapid and effective, equitable and responsible. This entails work to promote the prompt translation of scientific innovation into medical and public health policy and practice.

We provide knowledge, evidence and ideas to stimulate and direct well-informed debate on the potential and pitfalls of key genomic and biomedical developments, and to inform and educate stakeholders – policy makers, health professionals, patients and the public. We also provide expert research, analysis, health services planning and consultancy services for governments, health systems, and other non-profit organisations.


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