

**Minutes of the First meeting of the Steering Group for the Framework for Action on Birth Defects held on 11<sup>th</sup> November 2010, PHG foundation, Cambridge, UK**

**Members present:**

Alan Bittles	Director, Centre for Human Genetics, Edith Cowan University, Joondalup, Australia,
Hilary Burton	Director, PHG Foundation, Cambridge,
Arnold Christianson	Professor and Principal Specialist, Division of Human Genetics, National Health Laboratories Service, University of the Witwatersrand, Johannesburg, S Africa
Christopher Howson	Vice-President for Global Programs, The March of Dimes Foundation, White Plains, New York USA
Alastair Kent	Director, Genetic Alliance, London,
Irmgard Nippert	Professor and Chair for Women's Health Research, Institute for Human Genetics, Universitaetsklinikum Muenster, Muenster, Germany
Ysbrand Poortman	Chair, Preparing for Life Initiative, International Genetic Alliance, Baarn, Netherlands,
Peter Turnpenny	Consultant Clinical Geneticist & Hon. Senior Lecturer, Clinical Genetics Department, Royal Devon & Exeter Hospital, Exeter, Devon

**Also in attendance:** Corinna Alberg, Carol Lyon, Luis Nacul (all PHG Foundation, Cambridge)

**Apologies:** Apologies were received from Betty Kirkwood, Bernadette Modell and Mikkel Oestergaard

Item	Notes	Action
1	<p><b>Introduction and overview</b> Hilary Burton welcomed all those present to the first meeting of the steering group for the Framework for Action on Birth Defects and thanked participants for their attendance. All those at the meeting introduced themselves.</p>	
2.	<p><b>Introduction to the programme</b> Hilary Burton explained that the meeting would focus on the functions of the steering group and its planned method of operation. Discussion of the content of the Toolkit would be reserved for the workshop over the subsequent one and a half days.</p> <p>It is envisaged that the project will take place over 3-5 years, the first 2 years being a development phase and the subsequent 3 years a phase of putting the Toolkit into action. The PHG Foundation has been working on the Framework since February 2009 and the completion of the first phase will coincide with the workshop to be held in June 2011. In the longer term it is hoped that the Framework and use of the Toolkit would continue to develop without the lead being taken by the PHG Foundation.</p>	

	<p>Hilary Burton described the main objectives of the PHG Foundation - to bring a public health focus to genomics. She noted that the Framework for Action on Birth Defects builds on the work of the March of Dimes, that of Bernadette Modell and Arnold Christianson and the Capability Programme. Importantly, since the World Health Assembly meeting in May 2010, there is a mandate from the WHO to raise the position of birth defects on the international health agenda.</p> <p>The purpose of the Framework for Action on Birth Defects is to build a network of champions for action on birth defects and the Toolkit is a means of identifying needs and prioritizing actions in relation to birth defects in individual countries or regions within countries. Hilary Burton noted that although the term 'birth defects' was problematic it is the best recognized term for conditions that are present from birth, but if a better term was agreed upon the terminology could be changed.</p> <p>Chris Howson noted that the WHO will need something concrete to enable the WHA resolution to be transformed into action and that the Toolkit may fulfill that need. Arnold Christianson added that certain countries in the South are becoming aware of the impact of birth defects and since many countries in the South rely on the WHO to set the health agenda this may motivate governments to take action on birth defects.</p> <p>There was discussion on the lack of data and quality of data on birth defects in many countries. Peter Turnpenny highlighted that lack of high quality data should not stop action being taken.</p>	
<p><b>3.</b></p>	<p><b>Process discussion</b></p> <p>The purpose of the steering group and terms of reference were agreed. Hilary Burton said that she was willing to chair the steering group unless another chair was suggested. It was agreed that Hilary should chair the steering group. The draft terms of reference for the steering group were agreed.</p> <p>The membership of the steering group and any gaps was discussed. It was felt that an additional member from the South would be valuable. PHG will approach a suitable person to address this gap.</p> <p>Chris Howson noted that from the March of Dimes perspective the Framework for Action on Birth Defects was very important and that it was also critically important for all members of the steering group to work together.</p> <p>Hilary Burton noted that there are three categories of tasks for the steering group:</p> <ol style="list-style-type: none"> <li>1. Content issues - it was agreed to address this by circulating drafts and at times members of the steering group would be asked to comment on particular sections</li> </ol>	<p><b>PHG to recruit additional member.</b></p>

	<p>of the materials. This may be distributed through the Toolkit website.</p> <ol style="list-style-type: none"> <li>2. Process issues - helping the PHG Foundation in piloting materials.</li> <li>3. Strategic tasks- particularly embedding the Framework and Toolkit in the contexts and institutions in which it can be used.</li> </ol> <p>Alastair Kent noted that a key role for the steering group was making the programme sustainable.</p>	<p>All</p> <p>All</p>
4.	<p><b>Toolkit development</b></p> <p>Luis Nacul described the health needs assessment and prioritization process of the Toolkit. He explained that sections were being piloted in a number of countries, mostly in South America but that there had also been interest from a variety of other countries. It was felt that countries which are starting to recognize the impact of birth defects on infant mortality and childhood morbidity will be keenest to invest in the Framework and Toolkit. In addition, a key community of interest will be affected families, and pressure from them will change the health systems and make the project sustainable. Birth defects have to become part of public <i>health</i> policy. Chris Howson noted the effectiveness of such a grassroots project called Little Big Souls in Nigeria which was community based.</p> <p>There was discussion on the terminology and it was felt that terms such as RNA, DNA and Corporate were confusing and should be changed and terminology should be simple and clear.</p> <p>Many members of the steering group noted the importance of disseminating the work being undertaken through conferences and publications. A conference in Lodz, Poland will take place in 2011 part sponsored by the March of Dimes and the Toolkit should be presented at the event. Chris Howson will suggest to the organizers that information on the Toolkit should be part of the programme. Members of the steering group agreed to share information on relevant meetings and conferences with the PHG Foundation and suggest that events on birth defects should include information on the Toolkit.</p> <p>Chris Howson noted that having a demonstration project using the Toolkit is important, such as use in a South American context, and the results from that pilot should be widely disseminated.</p> <p>Steering group members were asked to let the PHG Foundation know if there is further information that they require when networking about the Framework and Toolkit.</p>	<p>PHG</p> <p>Chris Howson</p> <p>All</p> <p>PHG</p> <p>All</p>
5	<p><b>Dissemination and implementation</b></p> <p>In addition to the work on the Toolkit, the Foundation aims to develop a wider community of interest in 2011 who all would be</p>	

	<p>stakeholders in taking the birth defects agenda forward. Members of the steering group were asked to let Corinna Alberg know if there are organizations to approach to be part of the community of interest.</p>	<b>All</b>
<b>6</b>	<p><b>Funding and Fundraising</b>  Carol Lyon explained the PHG Foundation's financial investment in the Framework to date and clarified that external funding would be needed to disseminate the Toolkit more widely and for field trials beyond the current piloting of the materials.</p> <p>Irma Nippert noted that EU funding was potentially available. Hilary Burton felt that the PHG Foundation does not have sufficient experience or capacity to act as the principal coordinator for EU funding but that the PHG Foundation would be very willing to be a partner organization for EU funded projects led by others.</p>	
<b>7</b>	<p><b>Next steps</b>  Regarding the next steps, a workshop is planned for 27-29<sup>th</sup> June 2011 to present the Toolkit to key individuals working in the field of birth defects. This will be combined with the next steering group meeting. In the meantime a progress report will be sent to steering group members.</p>	
<b>8</b>	<p><b>Date of next meeting</b>  27<sup>th</sup> June 2011 London</p>	