

# Human Genetics of WHO



**EUROCAT meeting  
Prevention of Birth Defects:  
A view from WHO, Geneva  
Budapest, 6-7 March 2007**

*Dr Victor Boulyjenkov  
Human Genetics (HGN)  
World Health Organization*

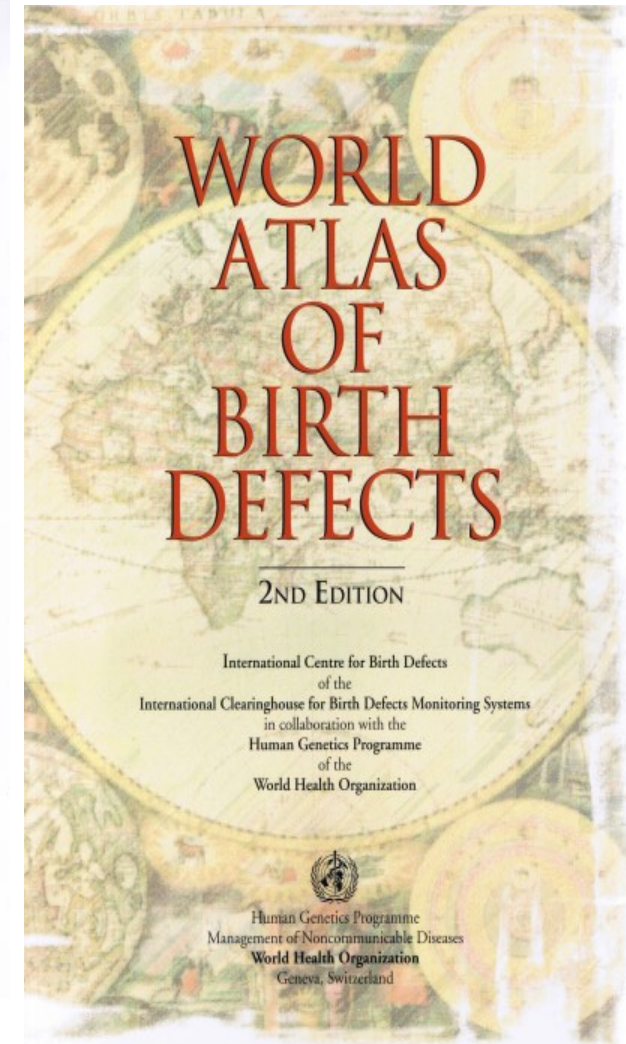
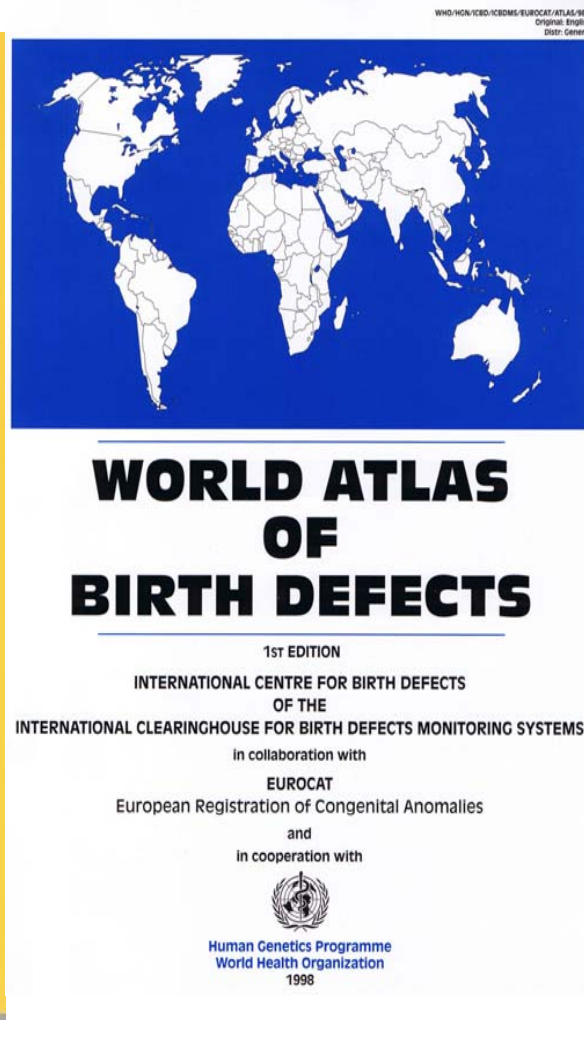
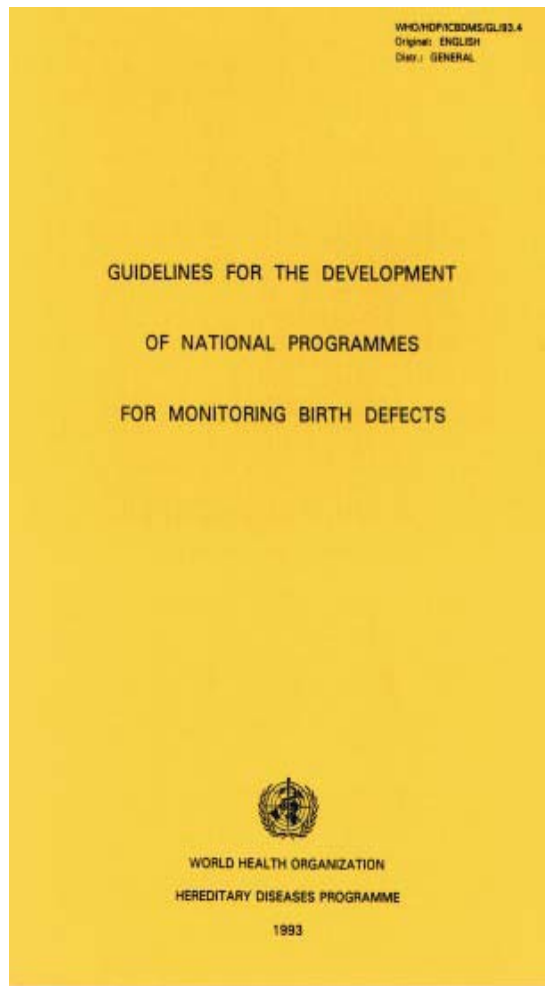


# Global Epidemiology of Genetic Disorders

Disorders	Global Average Data	Global number of Affected Population
<b>Congenital Disorders</b> (Down 's Syndrome; Cleft lip/palate)	5% of all newborns	<b>7 000 000</b> infants born each year
<b>Monogenic Diseases:</b>	1-1.5 % of newborns	1 400 000 to 2 100 000 infants born each year
Haemoglobinopathies (thalassaemias, sickle cell anaemia)	5% of population carry an abnormal Hb gene	300 000 infants born each year
Haemophilia	0.01% of newborn (males)	12 000 newborns (males) each year
<b>Common (multi-factorial) disorders</b> affecting middle- and later-life (cancer, CVD, asthma, diabetes, mental diseases)	About 40-60% of population may have a genetic predisposition to common NCDs	



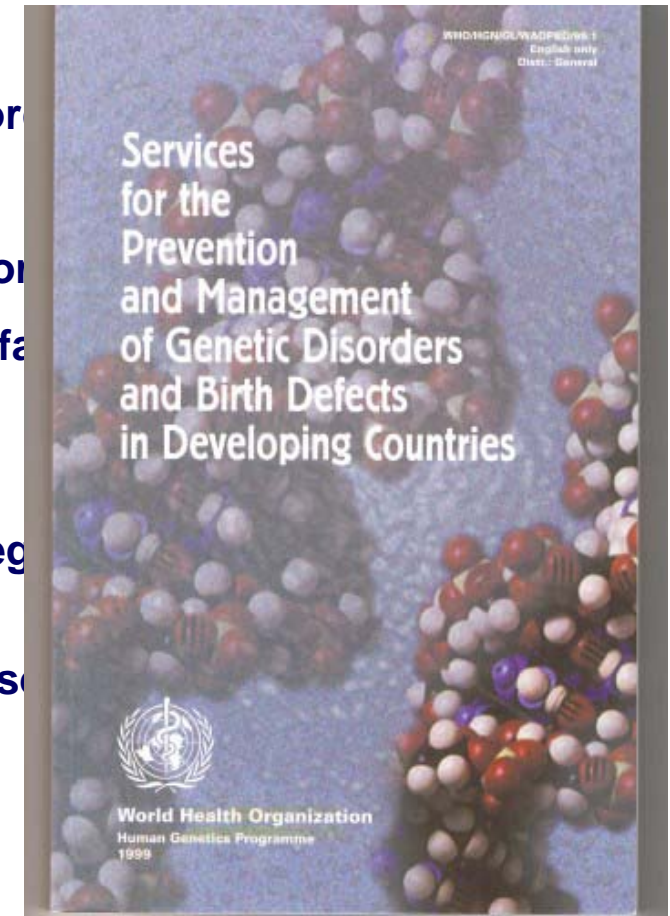
# Joint publications



# WHO/WAOPBD meeting on Prevention and Management of Genetic Disorders and Birth Defects in Developing Countries

## Main conclusions:

- Need to recognise the **burden** imposed by genetic disorders
- Need for **political** will and commitment
- Improve **epidemiological** knowledge about genetic disorders
- Define **goals** of genetic services in terms of individual/family and public health
- Improve pre- and **perinatal services**
- Organize genetic services in a comprehensive and integrated manner at the **primary health care** level
- Select programs and targets according to **prevalence**, social and economic outcomes
- Respect **ethical principles** and cultural diversity
- **Train** health professionals in genetics
- **Educate** the public in genetics
- Encourage the formation of **parent/patient organizations**



# WHO meeting on Primary Health Care Approaches for Prevention and Control of Congenital and Genetic Disorders

## Participating country

## WHO regional office

## WHO headquarters

National multidisciplinary group

Regional intersectoral collaboration

Global steering group, intersectoral collaboration

Critical examination of the evidence base for the recommended interventions in the country context

Critical examination of the evidence base for the interventions in a regional context

Critical examination for the inter

Country report and plan for intervention

Regional assessment

Global assessment

Baseline epidemiological study ("mother and baby study")

Technical support

Technical support

Initiation of selected initiatives in primary health care

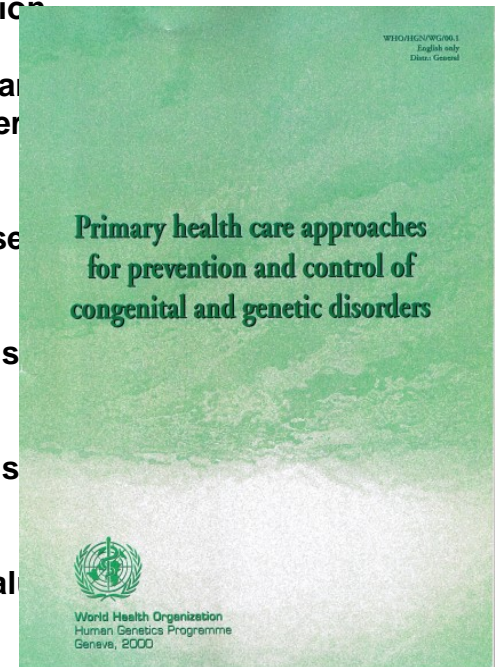
Technical support

Technical support

Evaluation of process and outcomes

Regional evaluation

Global evaluation



# Implementing Genetics into Primary Health Care

## Pilot Studies in 11 Member States spanning all WHO regions\*

### **AFR**

Nigeria, South Africa

### **AMR**

Brazil, Chile

### **EMR**

Bahrain, Egypt

### **EUR**

Cyprus, Russian Federation

### **SEAR**

India, Thailand

### **WPR**

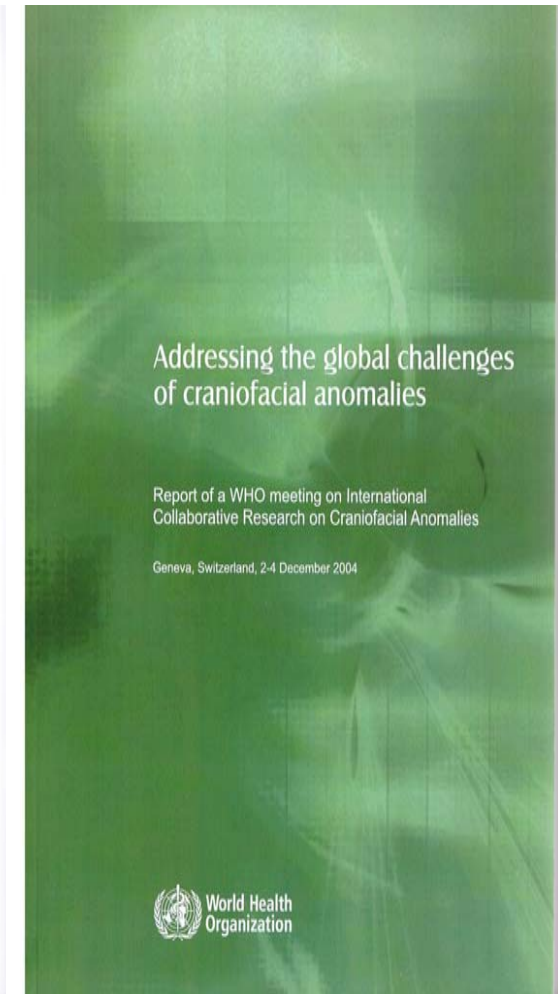
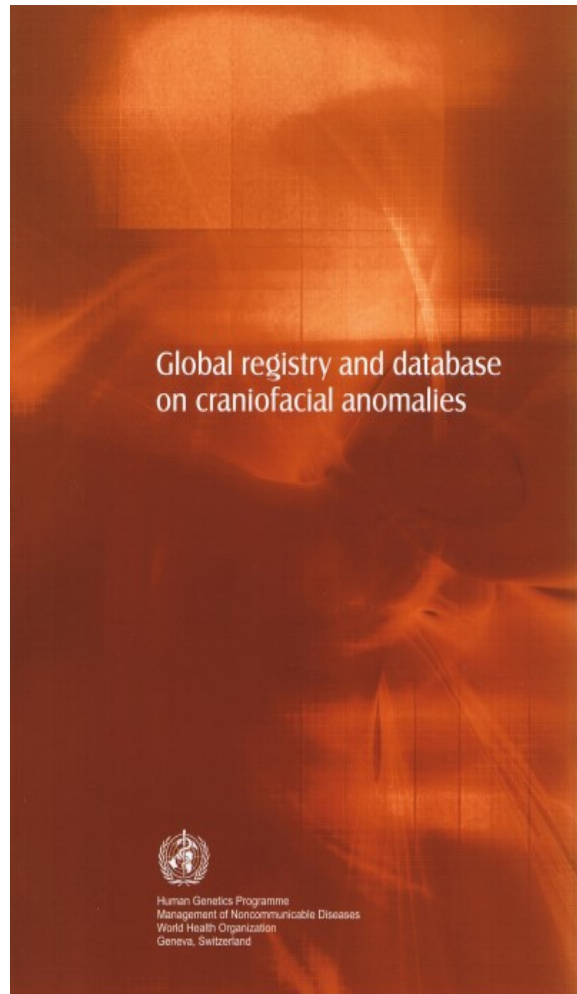
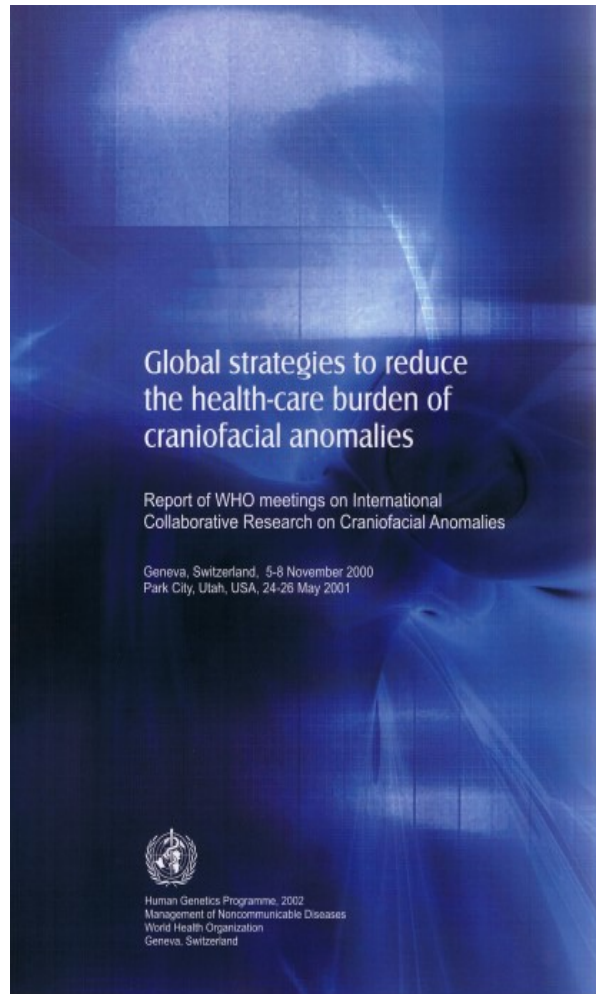
China

\* to assess the health burden of congenital and genetic disorders and improve their control in primary health care



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# Congenital malformations and CFA



# International Database on Craniofacial Anomalies (IDCFA)



The screenshot shows a web browser window displaying the WHO website for the International Database on Craniofacial Anomalies (IDCFA). The address bar shows the URL: <http://www.who.int/genomics/anomalies/idcfa/en/>. The page features the WHO logo and the text "World Health Organization" in a blue header. Below the header, there is a navigation menu on the left and a main content area on the right. The navigation menu includes links for Home, About WHO, Countries, Health topics, Publications, Research tools, WHO sites, Genomics home, Health professionals, Policy makers, Patients and public, Ethical, legal and social implications, Research, and Craniofacial anomalies. The main content area displays the title "International Database on Craniofacial Anomalies (IDCFA)" and provides information about the database, including a link to a printable version and data sets for typical orofacial clefts.

Address <http://www.who.int/genomics/anomalies/idcfa/en/>

Google Search Web 484 blocked AutoFill Options

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**Countries** WHO > WHO sites > Genomics home > Craniofacial anomalies

**Health topics** [printable version](#)

**Publications**

**Research tools** **International Database on Craniofacial Anomalies (IDCFA)**

**WHO sites**

**Genomics home** **About the IDCFA**

**Health professionals** - [Click here](#)

**Policy makers**

**Patients and public** **Data: Typical orofacial clefts cumulative data by register**

**Ethical, legal and social implications** - [Click here](#)

**Research**

**Craniofacial anomalies** **Data: Typical orofacial clefts by set of registries**

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# **IDCFA**

## **International Database of Craniofacial Anomalies**

**A world-wide initiative promoted by  
WHO – Human Genetics Programme and  
NIDCR – National Institute of Dental and Craniofacial Research**

**Coordinated by the ICBD – International Centre on Birth Defects**

- **Principal Investigators: Pierpaolo Mastroiacovo and Elisabeth Robert Gnansia**
- **Advisor: Eduardo E Castilla**

**Participating Registries – 63**



# WORLD HEALTH ORGANIZATION

**EXECUTIVE BOARD**  
**116th Session**

**EB116/3**  
**26 May 2005**

**Agenda item 4.1**

## **Control of genetic diseases**

### **Report by the Secretariat**

1. Increased knowledge of genomics over the past two decades has made it apparent that the traditional category of *genetic diseases* represents only those conditions in which the genetic contribution is particularly marked, whereas in fact diseases can be arrayed along a spectrum representing the varied contribution of genes and the environment. The beneficial *applications* of genomic knowledge are still evolving, but it is expected that in the future genomics will have “a significant contribution to make to the area of public health”.



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# World Health Organization WHA59.20 Resolution, 2006



世界衛生大會 決議

قرار جمعية الصحة العالمية

RESOLUTION OF THE WORLD HEALTH ASSEMBLY  
RÉSOLUTION DE L'ASSEMBLÉE MONDIALE DE LA SANTÉ  
РЕЗОЛЮЦИЯ ВСЕМИРНОЙ АССАМБЛЕИ ЗДРАВООХРАНЕНИЯ  
RESOLUCION DE LA ASAMBLEA MUNDIAL DE LA SALUD

FIFTY-NINTH WORLD HEALTH ASSEMBLY

WHA59.20

Agenda item 11.4

27 May 2006

## Sickle-cell anaemia

Having examined the report on sickle-cell anaemia;<sup>1</sup>

Recalling resolution WHA57.13 on genomics and world health, and the discussion of the Executive Board at its 116th session on control of genetic diseases, which recognized the role of genetic services in improving health globally and in reducing the global health divide;<sup>2</sup>

Recalling decision Assembly/AU/Dec.81 (V) of the Assembly of the African Union at its Fifth Ordinary Session;

Noting the conclusions of the 4th International African American Symposium on sickle-cell anaemia (Accra, 26-28 July 2000), and the results of the first and second international congresses of the International Organization to Combat Sickle-Cell Anaemia (respectively, Paris, 25-26 January 2002 and Cotonou, 20-23 January 2003);



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# World Health Organization EB118.R1 Resolution, 2006



世界衛生組織執行委員會決議

قرار المجلس التنفيذي لمنظمة الصحة العالمية

RESOLUTION OF THE EXECUTIVE BOARD OF THE WHO  
RÉSOLUTION DU CONSEIL EXÉCUTIF DE L'OMS  
РЕЗОЛЮЦИЯ ИСПОЛНИТЕЛЬНОГО КОМИТЕТА ВОЗ  
RESOLUCION DEL CONSEJO EJECUTIVO DE LA OMS

118th Session

EB118.R1

Agenda item 5.2

29 May 2006

## Thalassaemia and other haemoglobinopathies

The Executive Board,

Having considered the report on thalassaemia and other haemoglobinopathies;<sup>1</sup>

Recalling resolution WHA57.13 on genomics and world health, resolution EB117.R3 on sickle-cell anaemia and the recognition by the Executive Board at its 116th session of the role of genetic services in improving health globally and in reducing the global health divide;<sup>2</sup>

Concerned at the impact of genetic diseases, and of haemoglobinopathies (thalassaemia and sickle-cell anaemia) in particular, on global mortality and morbidity, especially in developing countries, and by the suffering of patients and families affected by the disease;

Recognizing that the prevalence of thalassaemia varies between communities, and that insufficient epidemiological data may hamper effective and equitable management;



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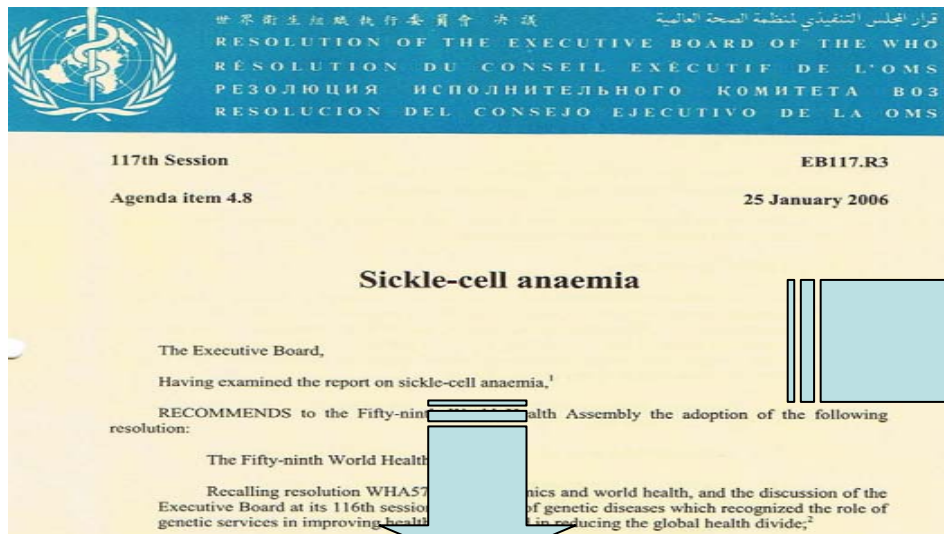
# Resolutions on Haemoglobinopathies

## Thalassaemia and other Haemoglobinopathies

EB118, May 2006 – Resolution EB118.R1

Urges Member States:

- Implement and reinforce **national programmes** on HB disorders
- Evaluate the **impact** of national programmes
- Intensify the **training** of all health professionals
- Promote community **education**
- Promote international **cooperation**
- Develop and strengthen medical **genetic services**
- Support basic and applied **research**



## Sickle cell anaemia

WHA59, May 2006 – Resolution WHA59.20



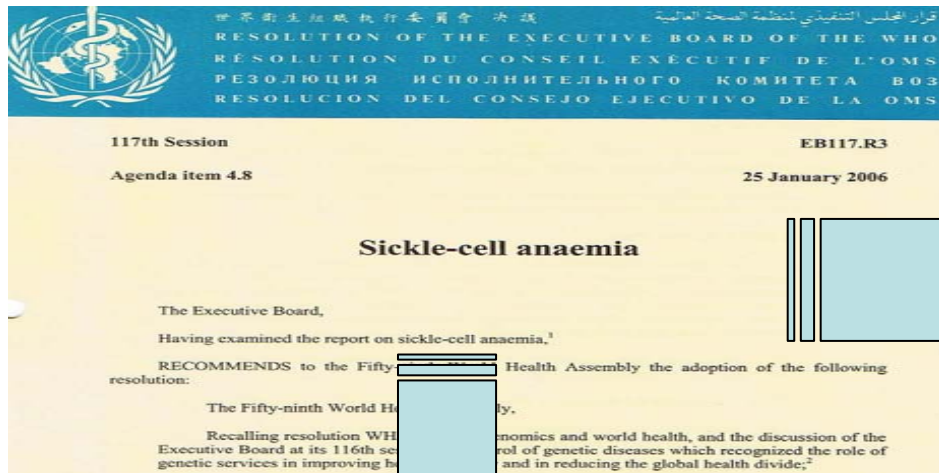
# Resolutions on Haemoglobinopathies

## Thalassaemia and other Haemoglobinopathies

EB118, May 2006 – Resolution EB118.R1

Requests the Director-General

- provide technical **support** and advice to national programmes
- expand the **training** and expertise of personnel
- support the further **transfer** of affordable technologies
- drafting **guidelines** on prevention and management
- fostering the establishment of **regional groups** of experts;
- support needed **research**



## Sickle cell anaemia

WHA59, May 2006 – Resolution WHA59.20



# Meeting of Experts, Geneva, May 2006

## MANAGEMENT OF BIRTH DEFECTS AND HAEMOGLOBIN DISORDERS

REPORT OF A JOINT  
WHO-MARCH OF DIMES MEETING



## Priorities for action

### for the care and prevention of birth defects

- support continued **research for the collection** and refinement of birth defects data to assist the development of medical genetic services
- provide practical advice and support for countries wishing to **develop pre-conception and medical genetic services**
- promote **human resource** capacity development and **technology transfer**.



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## Basic **reproductive approaches** to prevent birth defects that are appropriate for low- and middle-income countries

Promote **family planning**, allowing couples to space pregnancies, plan family size, define the ages at which they wish to begin and complete their families and reduce the proportion of unintended pregnancies and support health education of the public, particularly of women and girls. This will:

- reduce the overall rate of birth defects
- decrease the birth prevalence of Down syndrome by reducing the number of mothers of advanced maternal age
- allow women with affected children the option of not having further children
- introduce women to the concepts of reproductive choice

Before and during a woman's reproductive years, ensure a **healthy, balanced diet** and access to adequate quantities of macronutrients (protein, carbohydrates and fats) and micronutrients, including iodine, provided through universal salt iodization and folic acid through fortification of staple foods or supplementation, where these approaches are required. This will:

- prevent iodine deficiency disorders in women during pregnancy and thereby
- prevent the cognitive impairment resulting from iodine deficiency in their offspring
- decrease neural tube defects and other malformations
- prevent birth defects due to common teratogens such as alcohol and recreational drugs

**Control infections** in all women before and during pregnancy. In particular,

- prevent and treat syphilis
- prevent congenital rubella syndrome through immunization with rubella vaccine

Optimize maternal health through **control of chronic illnesses** associated with increased risk of birth defects. Target, in particular:

- insulin-dependent diabetes mellitus
- epilepsy and its control with anti-epileptic drugs
- women on Warfarin for deep vein thrombosis or cardiac conditions



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# Medical genetic **screening for birth defects** that are appropriate for low- and middle-income countries

## **Preconception**

Use of family history as a screening tool for birth defects and genetic conditions  
Carrier identification using family pedigrees  
Carrier screening for common recessive disorders, the haemoglobin disorders (FBC & indices, electrophoresis, DNA) and cystic fibrosis (DNA)

## **Antenatal**

Rhesus negativity  
Down syndrome (advanced maternal age, maternal serum, ultrasound)  
Neural tube defects (maternal serum & ultrasound)  
Major malformations (fetal anomaly scanning)  
Carrier screening for common recessive disorders, the haemoglobin disorders (DNA) & cystic fibrosis (DNA)

## **Postnatal**

Neonatal screening (using Guthrie cards)  
Congenital hypothyroidism  
Sickle cell disorders  
Neonatal jaundice /G6PD deficiency  
Inborn errors of metabolism



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# Genomic Resource Centre

[www.who.int/genomics](http://www.who.int/genomics)

HGN launched the Genomic Resource Centre (GRC),  
WHO's online resource on genomics and human genetics



The screenshot shows the WHO Genomic Resource Centre website. The address bar displays <http://www.who.int/genomics/en/>. The page features the WHO logo and navigation links for English, Spanish, and Français. A search bar is present. The main content area is titled "Genomic resource centre" and includes a navigation menu on the left with options like Home, Countries, Health topics, Publications, Research tools, WHO sites, Genomics home, Health professionals, Policy makers, Patients and public, Ethical, legal and social implications, Research, and Craniofacial anomalies. The main text describes the GRC as a resource base developed by the WHO Human Genetics Programme (HGN) to provide information and build awareness on human genetics. It mentions that the GRC includes individual sections designed to cater for the needs of the major stakeholder group in genomics, namely the public and the patients, the health professionals and the policy makers. It also notes that information is provided on the ethical, legal and social implications of genomics and the latest updates in genomic research. A sidebar on the right lists "NEW ON THE GRC" with links to "ELSI Genetics Regulation Database", "Community genetic services in Latin America and regional networks on medical genetics", and "Report of a WHO Consultation". Below this, there are sections for "GENOMICS" with links for "Resource submissions", "Feedback forms", "Publications", "Media room", and "News and events".



## For more information on HGN

- Visit to Genomic Resource Centre  
<http://www.who.int/genomics>
- Write to HGN: [genomics@who.int](mailto:genomics@who.int)

*Thank you*

