Module 5

THE IMPACT OF WHO RESOLUTIONS ON GENETIC DISORDERS:

EXAMPLE OF HEMOGLOBINOPATHIES

Author: Patricia Aguilar Martinez, France

OUTLINE

- WHO resolutions
- Hemoglobin disorders at the global level
- WHO & Hemoglobin Disorders

- Implementation of WHO Resolutions on Hb Disorders

WHO Resolutions

WHAT ARE WHO RESOLUTIONS?

Resolution

A **resolution** is a written motion (formal proposal) adopted by a deliberative body.

WHO resolutions

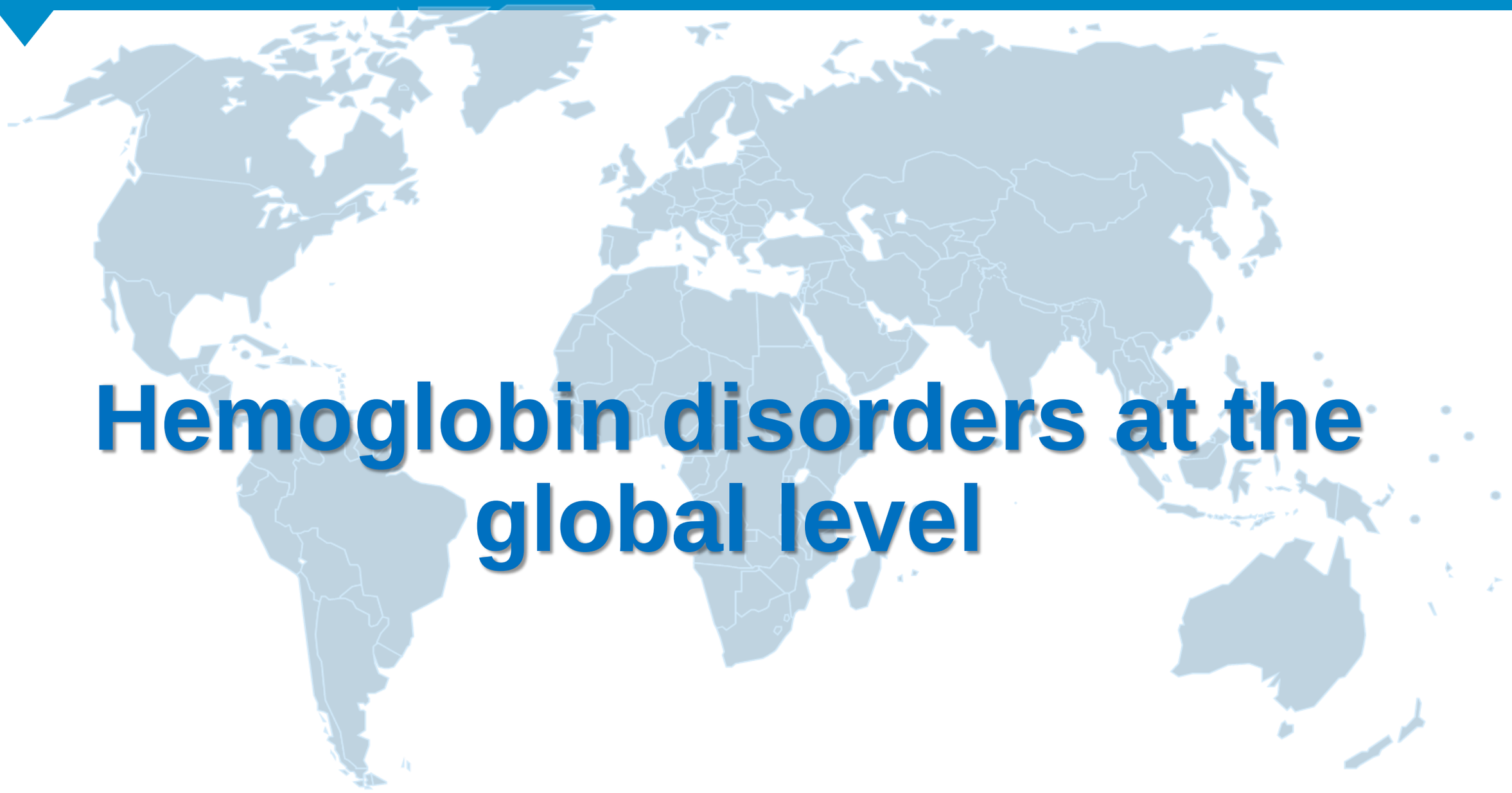
They are discussed and approved by the **World Health Assembly** (**WHA**) during its annual meeting

Or by the **Executive Board** (**EB**)

WHAT IS THE IMPACT OF WHO RESOLUTIONS?

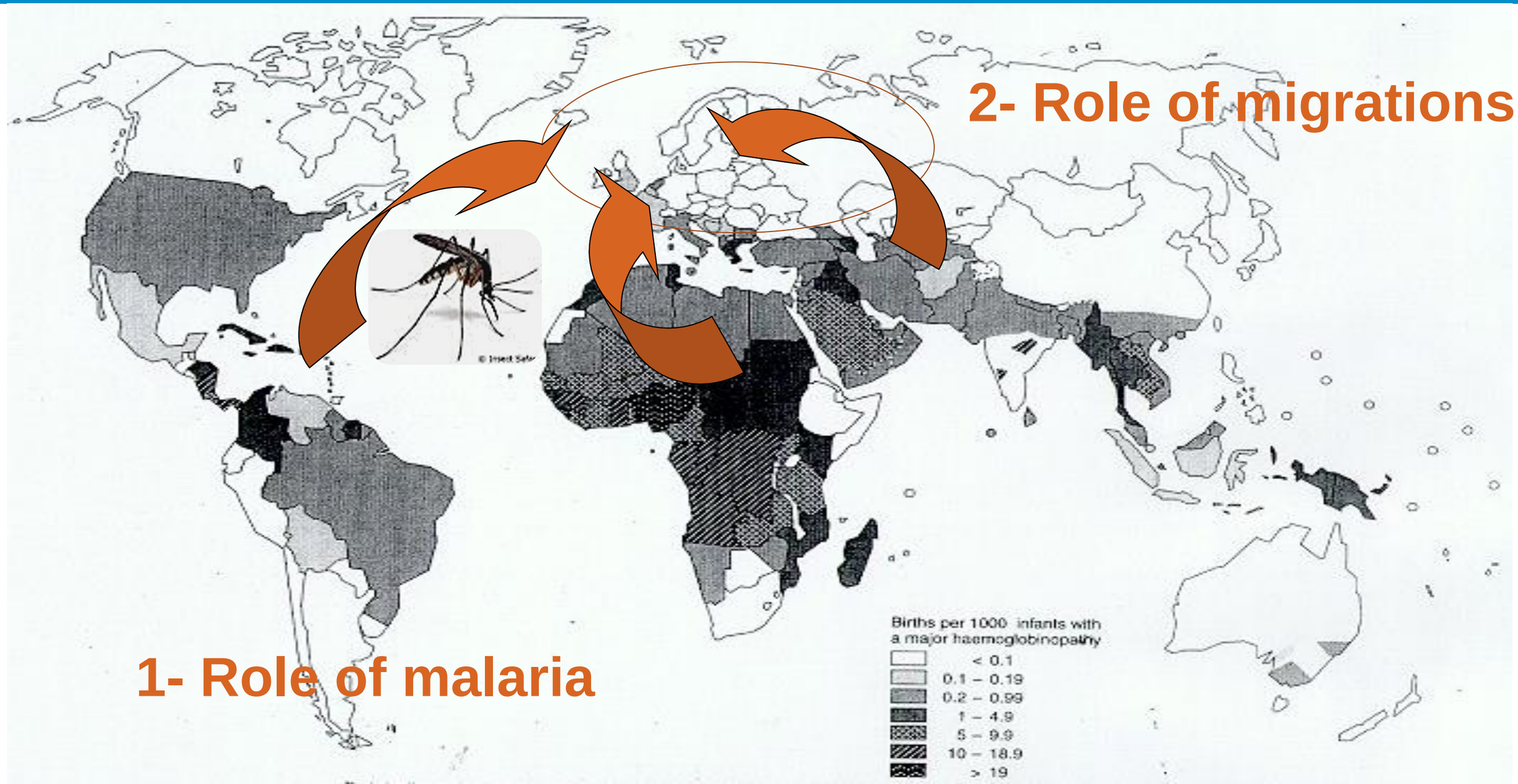
WHO resolutions

- Have a global diffusion to all **member states**
- Imply that the **WHO Director General** (DG) is committed in their application



Hemoglobin disorders at the global level

GLOBAL DISTRIBUTION OF HEMOGLOBIN DISORDERS (BIRTHS OF AFFECTED INFANTS PER 1000 BIRTHS, WHO, 1996)



PREVENTION PROGRAMMES FOR THALASSEMIA

Example of Cyprus

Preventive Medicine

PREVENTION OF THALASSAEMIA IN CYPRUS

M. A. ANGASTINIOTIS M. G. HADJIMINAS

Cyprus Thalassaemia Centre, General Hospital, Nicosia, Cyprus

Summary A programme for the prevention of β-homozygous thalassaemia has been operating in Cyprus from 1973. From 1976 there has been an increasing gap between the number of homozygotes born and the number expected, calculated as 1 in 135 of the total births. In 1978 23 homozygotes are known to have been born compared with 71 expected, and in 1979, 18 compared with 77 expected. All cases may not have yet come to notice. The programme consists of public education, population screening, genetic counselling, and antenatal diagnosis.

INTRODUCTION

THE difficulties of population screening for the prevention of inherited disease have recently been discussed,[1] and doubts have been expressed about whether such programmes can succeed, especially in areas where they are most needed. The budget of the Ministry of Health. Patients do not contribute to this cost.

Development of Prevention Programme

Thoughts on population screening and prevention started with the visit to the island in 1971 of Prof. G. Stamatoyiannopoulos, a World Health Organisation adviser on thalassaemia to the Cypriot Government.[6]

By 1973 much preparatory public education had been done and population screening was started. 1321 were tested, mostly single people (high school girls, army recruits, and relatives of homozygotes) with a view to preventing marriages between heterozygotes and, perhaps later, establishing a "pre-marital certificate".

In 1974 the programme continued on similar lines but was interrupted by political events and the separation of the two main communities (Greek Cypriots and Turkish Cypriots) which excluded the Turkish Cypriot population from our programme.

From 1974 to 1977 the programme had many organisational and financial difficulties. The people tested were those who came forward voluntarily.

In 1977 antenatal diagnosis for β-thalassaemia had become a reality[7,8] and we began concentrating our efforts on people of reproductive age. In 1978 a team separate from other hospital laboratories was established to concentrate on the prevention and study of thalassaemias.

PRESENT PROGRAMME

PREVENTION PROGRAMMES FOR THALASSEMIA

Example of Sardinia

Am J Hum Genet 33:592–605, 1981

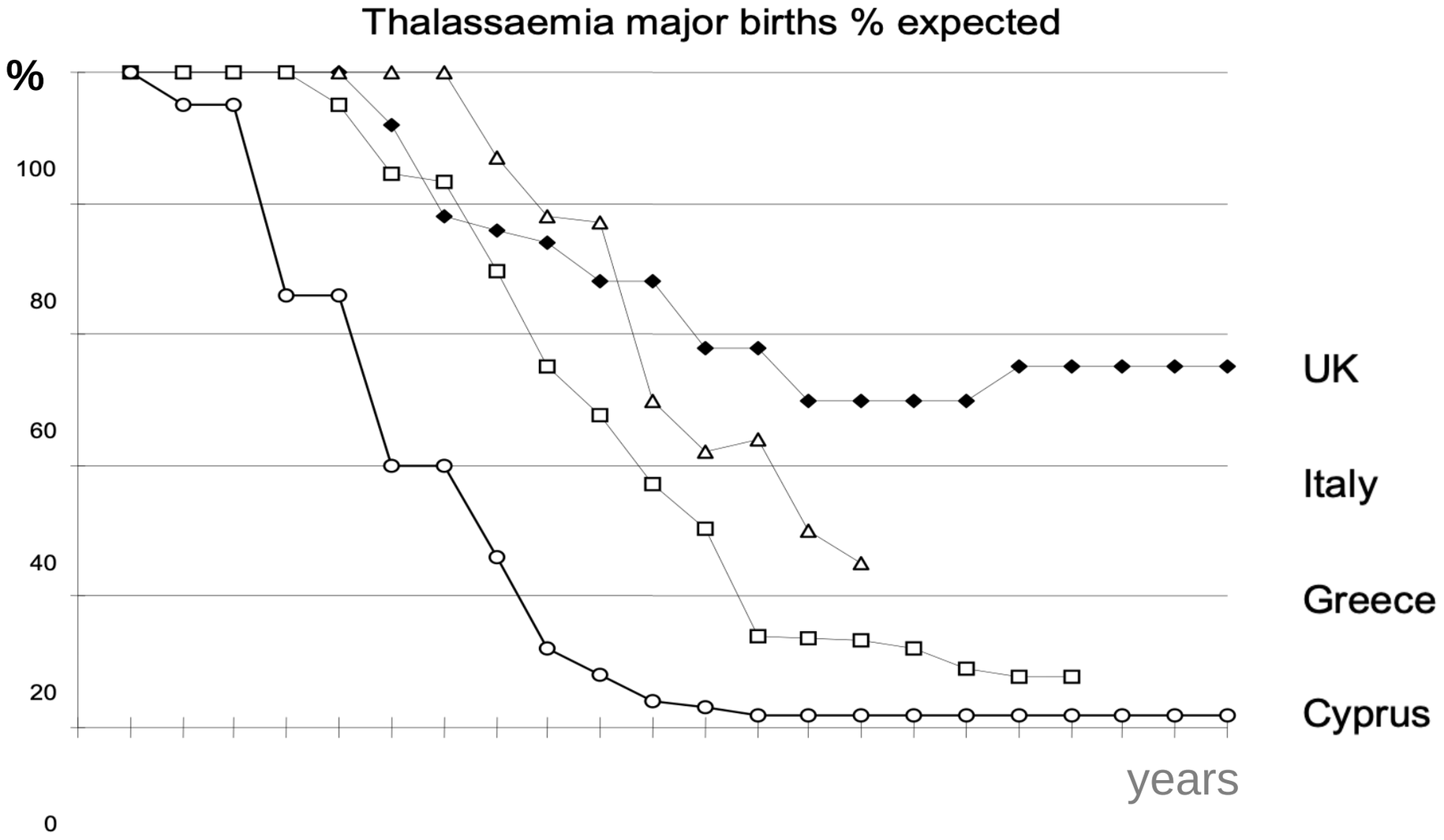
Prevention of Homozygous β-Thalassemia by Carrier Screening and Prenatal Diagnosis in Sardinia

A. Cao,[1] M. Furbetta, R. Galanello, M. A. Melis, A. Angius, A. Ximenes, C. Rosatelli, R. Ruggeri, M. Addis, T. Tuveri, A. M. Falchi, E. Paglietti, and M. T. Scalas

SUMMARY

We report here results of a 3-year pilot voluntary screening program coupled with prenatal diagnosis directed to the prospective prevention of homozygous β-thalassemia (β-thal) in Sardinia. The screening program took two approaches: outreach community testing and hospital testing on request after a period of sensibilization. The outreach testing was very effective as, taking into account the already known number of couples at risk with an affected proband (20), 74% of the couples at risk expected (61) on the basis of the carrier rate were identified. Less effective was the hospital testing in which half of the couples at risk expected were detected

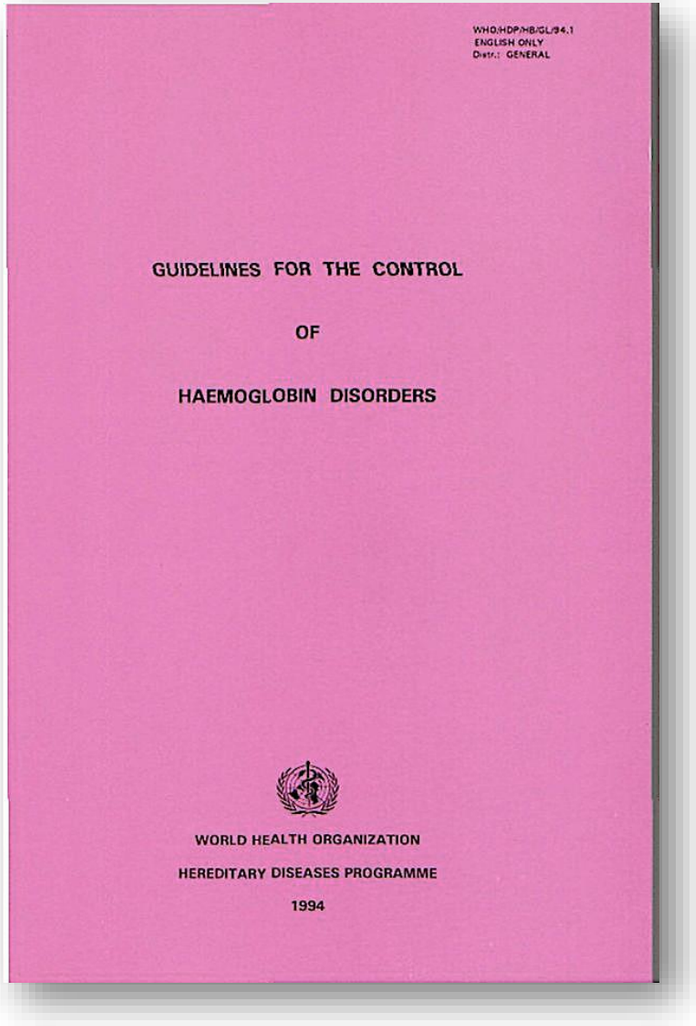
FALL IN THE BIRTH RATE OF CHILDREN WITH THALASSEMIA IN SELECTED COUNTRIES



WHO & Hemoglobin Disorders

CONTROL OF HEMOGLOBIN DISORDERS

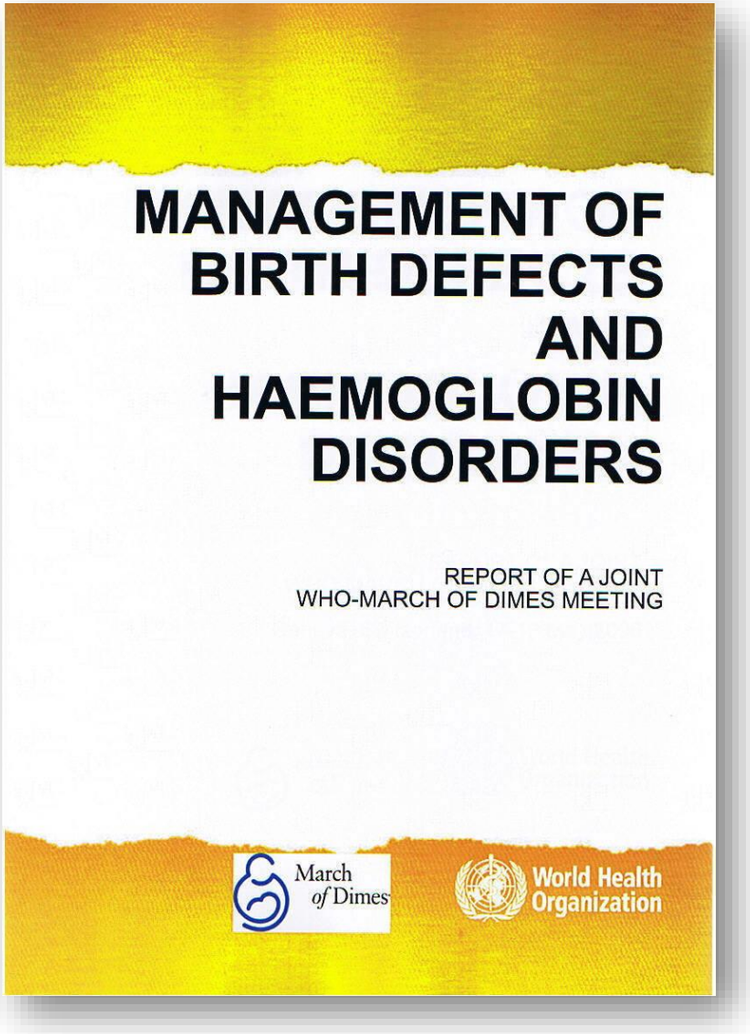
WHO guidelines, 1994



MEETING OF EXPERTS, GENEVA, MAY 2006

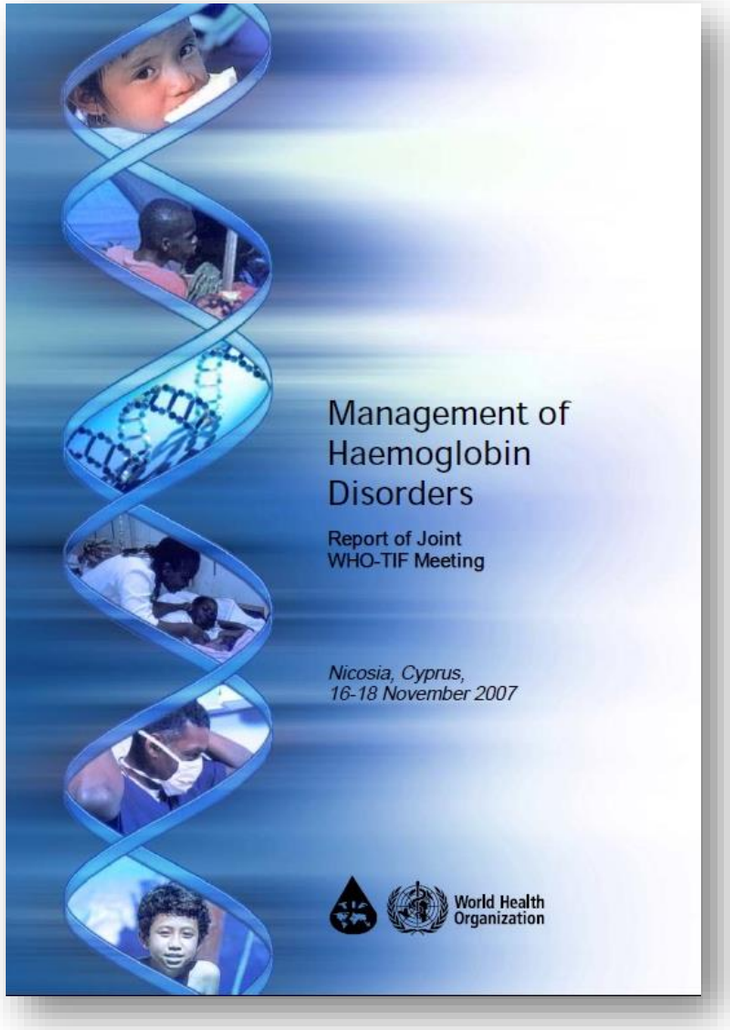
WHO-March of Dimes meeting

Management of the disease



WHO - TIF MEETING, CYPRUS, 2007

Management of Hb disorders



WHO resolutions on Hemoglobinopathies

LIST OF WHA / EB RESOLUTIONS (IN GENETICS)

WORLD HEALTH ASSEMBLY

WHA57.13	22 May 2004	Genomics and world health
WHA59.20	27 May 2006	Sickle cell anaemia
WHA63.17	21 May 2010	Birth defects

EXECUTIVE BOARD

EB116/3	21 Apr 2005	Control of genetic diseases
EB117.R3	25 Jan 2006	Sickle cell anaemia
EB118.R1	29 May 2006	Thalassaemia and other haemoglobinopathies

LIST OF WHA / EB RESOLUTIONS: SICKLE CELL ANEMIA

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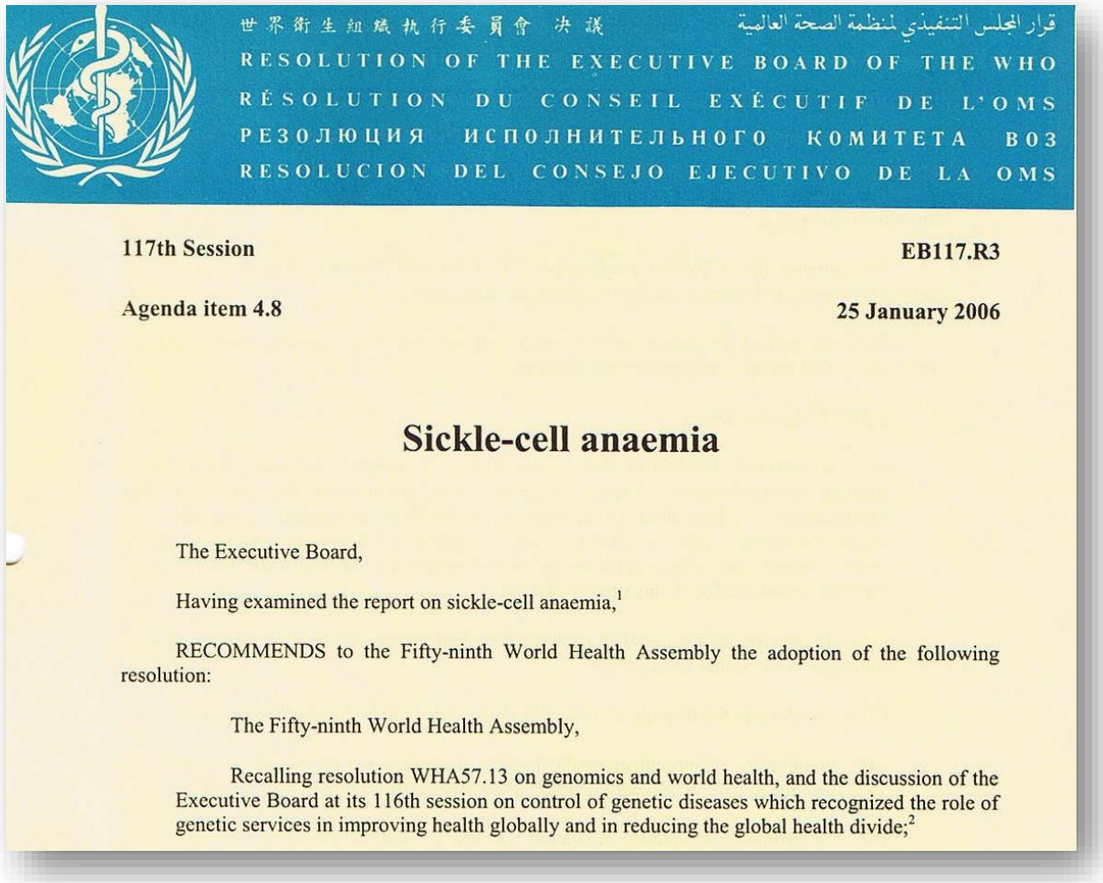
WORLD HEALTH ORGANIZATION: RESOLUTION ON SICKLE CELL ANEMIA



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

117th Session EB117.R3

Agenda item 4.8 25 January 2006

Sickle-cell anaemia

The Executive Board,

Having examined the report on sickle-cell anaemia,[1]

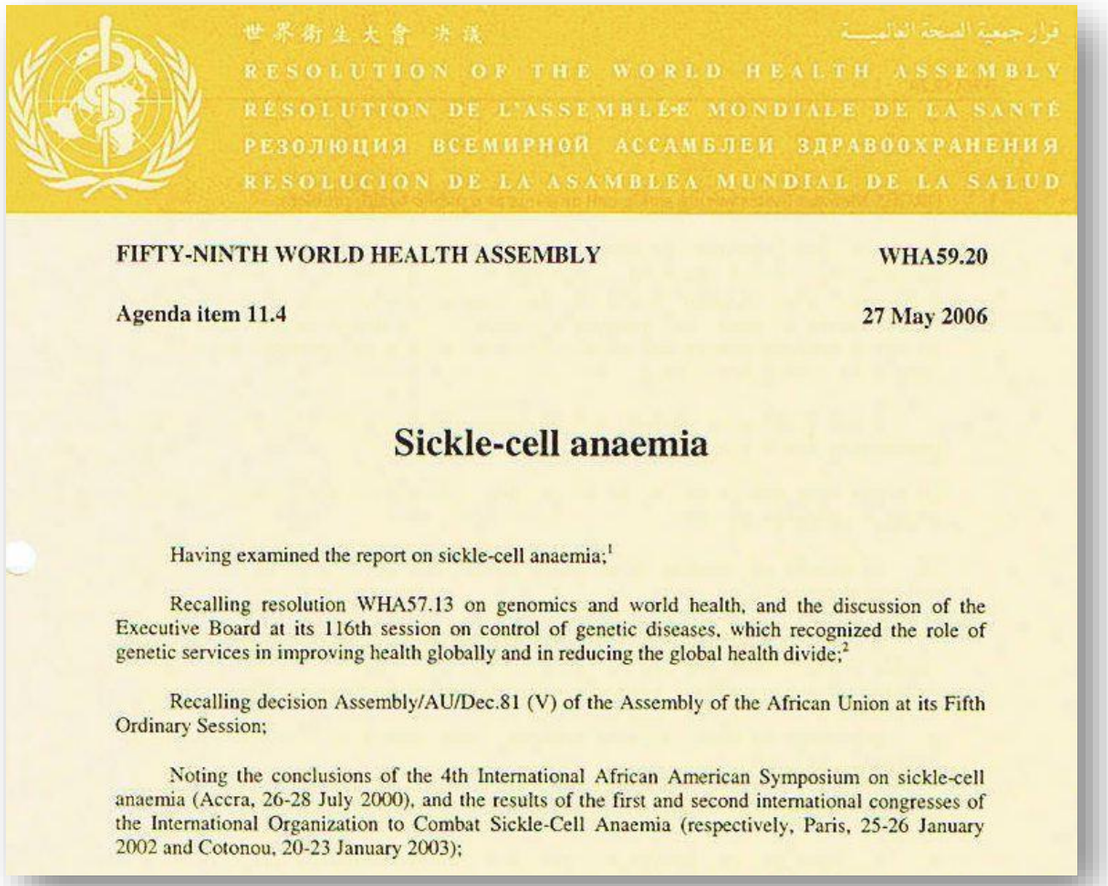
RECOMMENDS to the Fifty-ninth World Health Assembly the adoption of the following resolution:

The Fifty-ninth World Health Assembly,

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;[2]

- **EB117.R3 Resolution, 2006**

WORLD HEALTH ORGANIZATION: RESOLUTION ON SICKLE CELL ANEMIA



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

RESOLUCIÓN 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;[1]

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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);

- **WHA59.20 Resolution, 2006**

LIST OF WHA / EB RESOLUTIONS: THALASSEMIA

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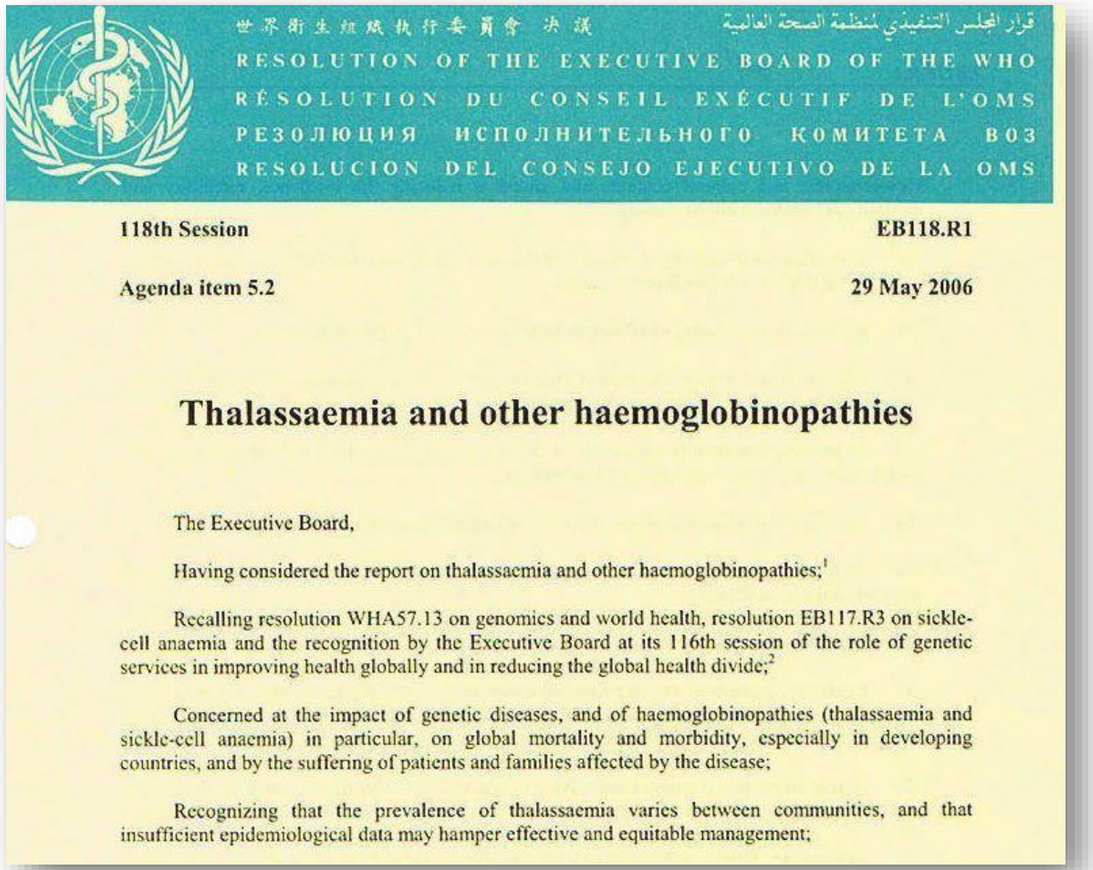
WORLD HEALTH ORGANIZATION: RESOLUTION ON THALASSEMIA



世界衛生組織執行委員會 決議 قرار المجلس التنفيذي لمنظمة الصحة العالمية

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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;[1]

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;[2]

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;

- EB118.R1 Resolution, 2006

LIST OF WHA / EB RESOLUTIONS (IN GENETICS)

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WORLD HEALTH ORGANIZATION

EB118.R1 Resolution, 2006

World Health Organization

SIXTY-THIRD WORLD HEALTH ASSEMBLY — A63/10
Provisional agenda item 11.7 — 1 April 2010

Birth defects

Report by the Secretariat

1. The report aims to inform the discussion on birth defects, including definition, epidemiology, burden of disease and interventions for prevention and care, as well as indications of how these interventions might be integrated into existing health services. An earlier version of this report was considered by the Executive Board at its 126th session,[1] following which the Board adopted

Also mentions hemoglobinopathies

detection rate of congenital disorders in the first trimester through biochemical screening is improved when it is undertaken in tandem with ultrasound screening involving nuchal translucency and other ultrasonographical assessments. Ultrasonography in the second trimester is useful to detect major structural defects.

DETECTION, TREATMENT AND CARE

11. Screening of newborn infants for congenital disorders facilitates early detection, treatment and care. Neonatal screening programmes (physical examination of all neonates and screening for congenital hypothyroidism, phenylketonuria, sickle-cell disease and glucose-6-phosphate dehydrogenase deficiency) and training of primary health-care providers support the diagnosis and appropriate referral for treatment of infants with congenital disorders. Physical examination of all newborn infants by trained primary health-care practitioners is feasible in most health systems and allows the identification of many birth defects, including cardiovascular defects that are associated with a high risk of early mortality and referral.

12. Treatment of birth defects depends on the level of health care available. It comprises medical therapy, surgery, rehabilitation and palliative care when appropriate.

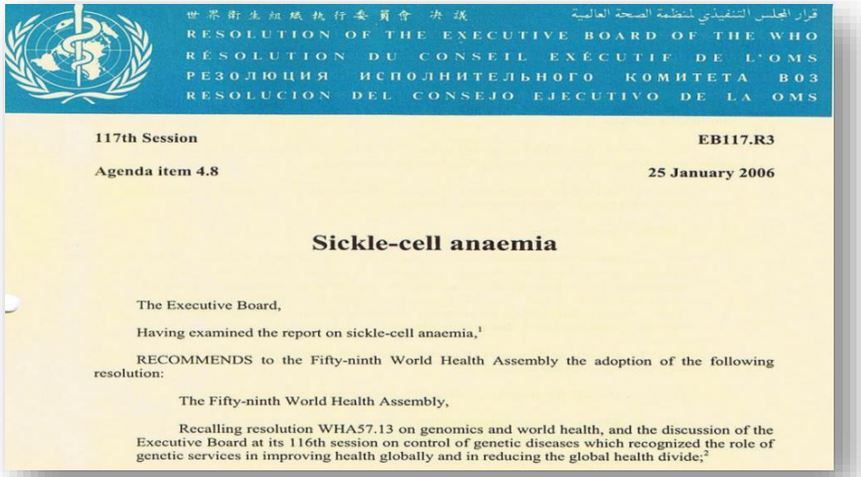
13. Effective life-saving medical treatment is available for several birth defects, including some common functional single-gene defects. Examples include treatment of neonatal jaundice in glucose-6-phosphate dehydrogenase deficiency and in Rhesus incompatibility, and therapy for congenital hypothyroidism, sickle-cell disorders, thalassaemia, haemophilia, cystic fibrosis, and other inborn errors of metabolism. Other treatment options include in utero therapy and postnatal surgical corrections; these are now under research and evaluation in a few selected centres for a number of conditions (e.g. congenital diaphragmatic hernia, congenital heart lesions, myelomeningocele, twin-to-twin transfusion syndrome).

WHO RESOLUTIONS ON HEMOGLOBINOPATHIES

- Sickle cell anemia (SCD)
- Thalassemia (Thal) and other hemoglobinopathies
- Birth defects (including SCD & Thal)

What do they say?

WHO RESOLUTIONS ON HEMOGLOBINOPATHIES



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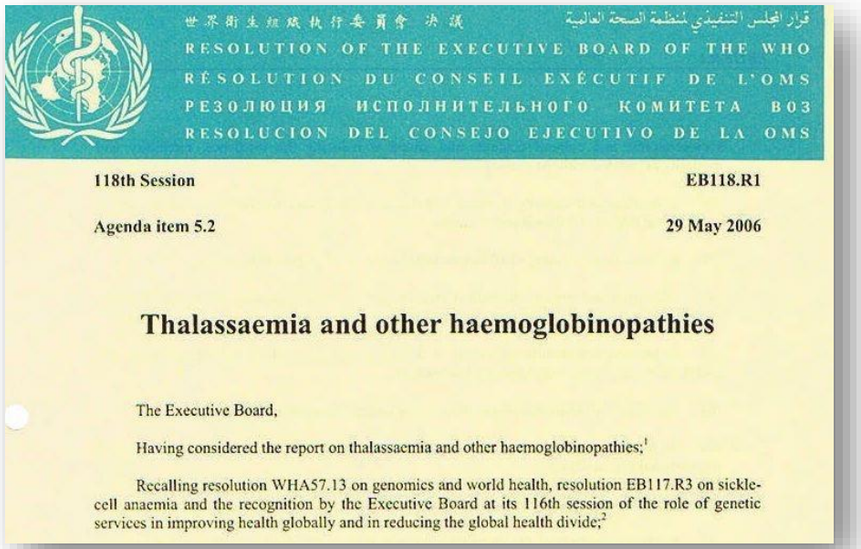

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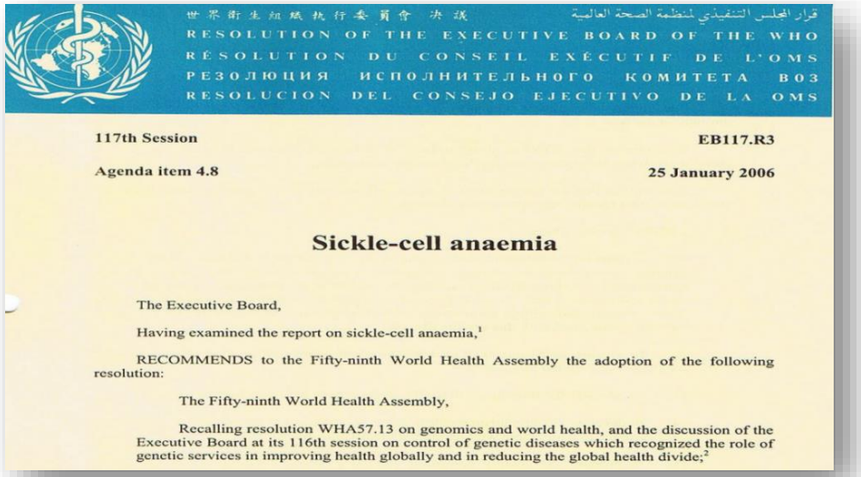
Urge Member States:

- To implement and reinforce national programmes on HB disorders
- To evaluate the impact of national programmes
- To intensify the training of all health professionals
- To promote community education
- To promote international cooperation
- To develop and strengthen medical genetic services
- To support basic and applied research

WHO RESOLUTIONS ON HEMOGLOBINOPATHIES



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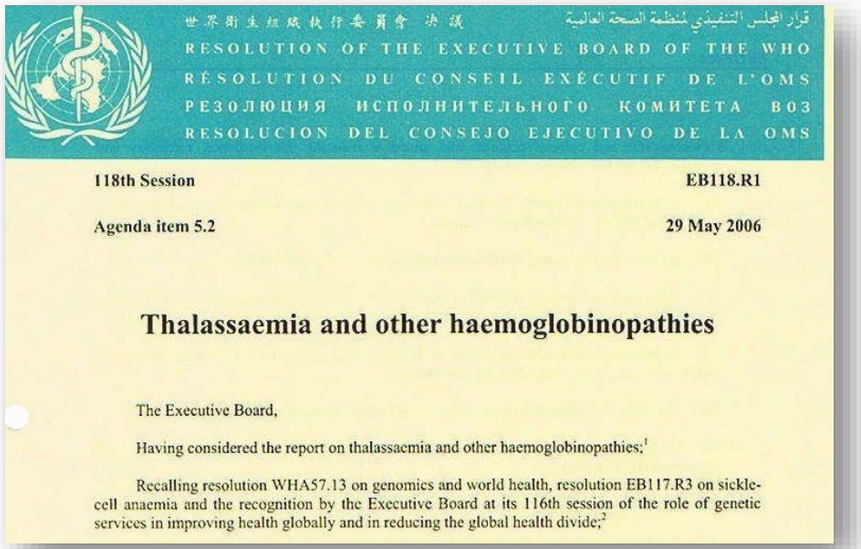

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Request the Director-General:

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

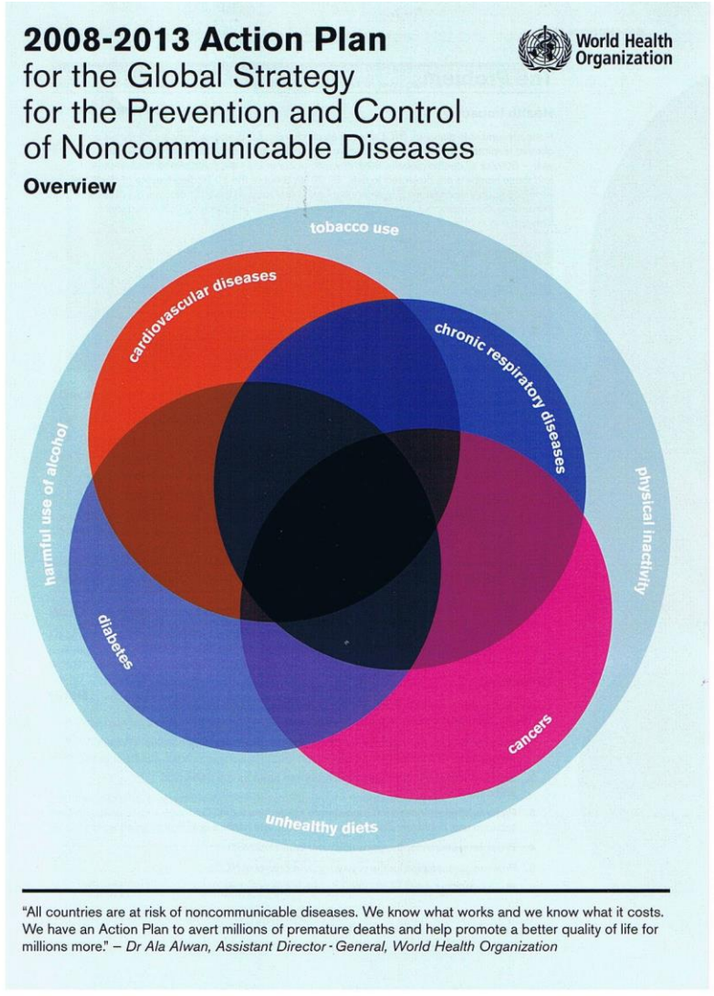
2008-2013 Action Plan
for the Global Strategy
for the Prevention and Control
of Noncommunicable Diseases
Overview

World Health Organization

Implementation of WHO Resolutions on Hb Disorders

"All countries are at risk of noncommunicable diseases. We know what works and we know what it costs. We have an Action Plan to avert millions of premature deaths and help promote a better quality of life for millions more." – *Dr Ala Alwan, Assistant Director-General, World Health Organization*

WHO ACTION PLAN (2008-2013) FOR THE PREVENTION AND CONTROL OF NCDS



- Integrate NCD prevention into the development agenda, and into policies across all government departments
- Establish and strengthen national policies and plans for the prevention and control of NCDs
- Promote interventions to reduce the main risk factors for NCDs: tobacco use, unhealthy diets, physical inactivity and harmful use of alcohol
- Promote research for the prevention and control of NCDs
- Promote partnerships for the prevention and control of NCDs
- Monitor NCDs trends and assess progress made at country level

WHO - HUMAN GENETICS: PARTNERSHIPS

Partners

- WHO Regions
- WHO Collaborating Centres
- NGOs in official relations with WHO-HGN (ex: TIF)
- Other International Organizations…

IMPLEMENTATION OF WHO RESOLUTIONS

Role of Partners

The 6 WHO Regions:

- Eastern Mediterranean
- Africa
- South East Asia
- Western Pacific
- Europe
- Americas

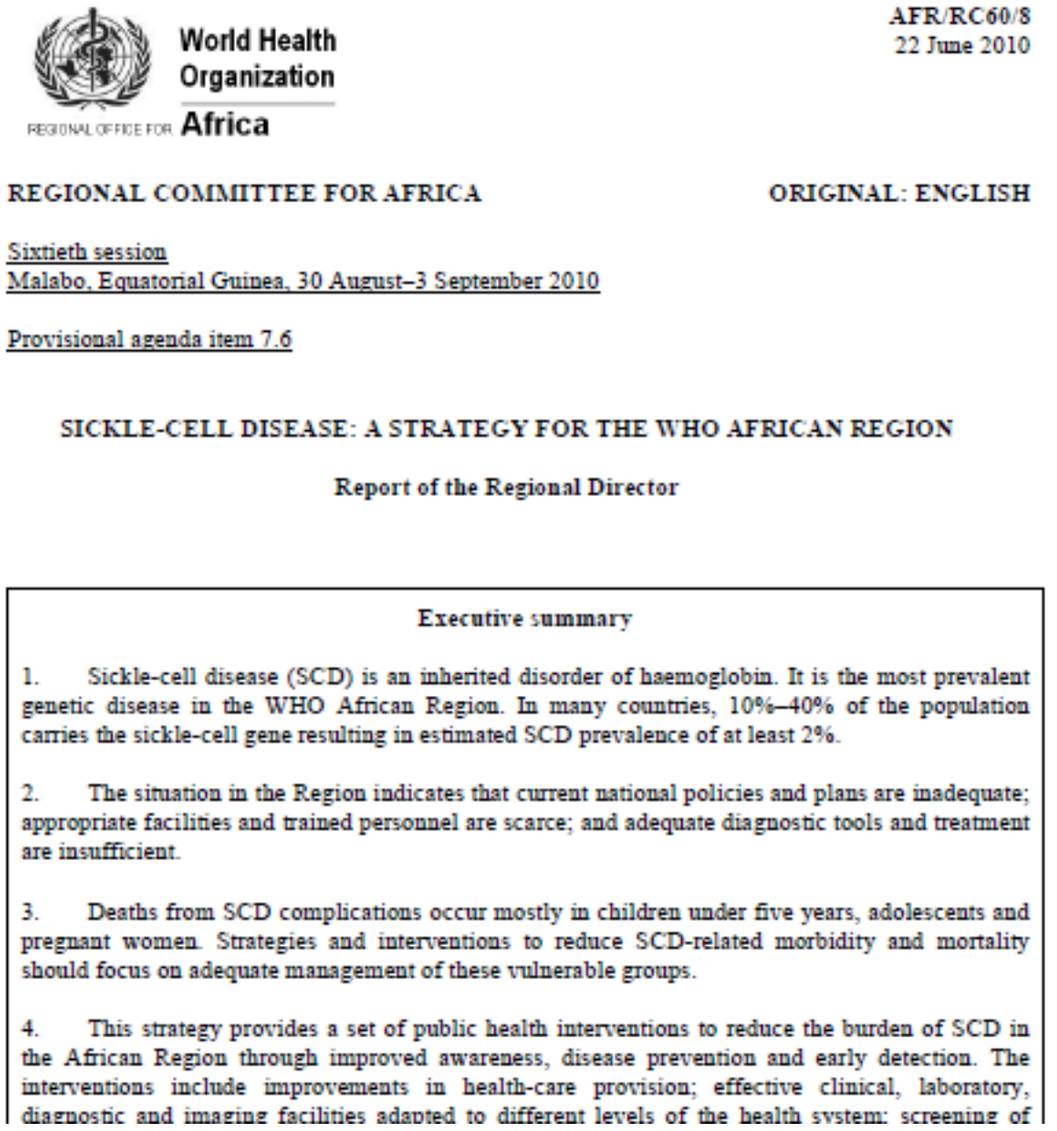
IMPLEMENTATION OF WHO RESOLUTIONS

Role of Partners

The 6 WHO Regions:

- Eastern Mediterranean
- **Africa**
- South East Asia
- Western Pacific
- Europe
- Americas

Adoption by the WHO regional Committee of SCD strategy in Africa



World Health Organization
REGIONAL OFFICE FOR Africa

AFR/RC60/8
22 June 2010

REGIONAL COMMITTEE FOR AFRICA — ORIGINAL: ENGLISH

Sixtieth session
Malabo, Equatorial Guinea, 30 August–3 September 2010

Provisional agenda item 7.6

SICKLE-CELL DISEASE: A STRATEGY FOR THE WHO AFRICAN REGION

Report of the Regional Director

Executive summary

1. Sickle-cell disease (SCD) is an inherited disorder of haemoglobin. It is the most prevalent genetic disease in the WHO African Region. In many countries, 10%–40% of the population carries the sickle-cell gene resulting in estimated SCD prevalence of at least 2%.

2. The situation in the Region indicates that current national policies and plans are inadequate; appropriate facilities and trained personnel are scarce; and adequate diagnostic tools and treatment are insufficient.

3. Deaths from SCD complications occur mostly in children under five years, adolescents and pregnant women. Strategies and interventions to reduce SCD-related morbidity and mortality should focus on adequate management of these vulnerable groups.

4. This strategy provides a set of public health interventions to reduce the burden of SCD in the African Region through improved awareness, disease prevention and early detection. The interventions include improvements in health-care provision; effective clinical, laboratory, diagnostic and imaging facilities adapted to different levels of the health system; screening of

CONCLUSION

- WHO resolutions on hemoglobin disorders: a health priority at the global level
- Implementation at the country level warrants collaborations in all the fields

WHO PUBLICATIONS ON HEMOGLOBIN DISORDERS

- **WHO, 1989.** *Report of the fifth WHO working group on the feasibility study on hereditary disease community control programmes (Hereditary anaemias) .* **WHO, Geneva, Switzerland. (WHO/HDP/WG/HA/89.2)**
- **WHO, 1991.** *Guidelines for the Management of Sickle Cell Disease.* **WHO, Geneva, Switzerland (WHO/HDP/SCD/91.2)**
- **WHO, 1993.** *Report of a joint WHO/TIF meeting on the prevention and control of haemoglobinopathies*. **WHO, Geneva, Switzerland (WHO/HDP/TIF/WG/93.1)**
- **WHO, 1994.** *Educational materials on prenatal diagnosis for Sickle-cell disorder.* **WHO, Geneva, Switzerland (WHO/HDP/EM/PN.SCD/94.2).**
- **WHO, 1994.** *Guidelines for the Control of Haemoglobin Disorders.* **WHO, Geneva, Switzerland (WHO/HDP/HB/GL/94.1).**
- **WHO, 1995.** *Prevention and Control of Haemoglobinopathies*. **WHO Bulletin, v73(3):375-386.**
- **WHO, 1997.** *Inherited Haemoglobin Disorders: an increasing global health problem*. **WHO Bulletin, v.75 (3):15-39.**
- **WHO, 1999.** *Services for the Prevention and Management of Genetic Disorders and Birth Defects in Developing Countries.* **WHO, Geneva, Switzerland (WHO/HGN/WAOPBD/99.1)**
- **WHO, 2000.** *Primary Health Care Approaches for Prevention and Control of Congenital and Genetic Disorders.* **WHO, Geneva, Switzerland (WHO/HGN/WG/00.1)**
- **WHO, 2002.** *Minutes of a WHO meeting on haemoglobin disorders*. **WHO, Geneva, Switzerland (WHO/HGN/HB/02.4)**
- **WHO, 2002.** *Report of the Advisory Committee on Health Research.* ***Genomics and World Health*****. WHO, Geneva, Switzerland (ISBN 92 4 154554 2).**
- **WHO, 2003.** *Genetic Approaches to Haemoglobin Disorders and Primary health Care.* **WHO, Geneva, Switzerland (WHO/HGN/TIF/CONS/03.1)**
- **WHO, 2006.** *Report by Secretariat to Executive Board: Sickle-cell anaemia*. **EB117, Doc. EB117/34. WHO, Geneva, Switzerland**
- **WHO, 2006.** *Report by Secretariat to World Health Assembly: Sickle-cell anaemia.* ***WHA59, Doc.A59/9*****. WHO, Geneva, Switzerland**
- **WHO, 2006.** *Report by Secretariat to Executive Board: Thalassaemia and Other Haemoglobinopathies*. **EB118, Doc. EB118/5. WHO, Geneva, Switzerland**
- **WHO, 2006.** *Executive Board Resolution on Sickle Cell Anaemia.* **EB117.R3. WHO, Geneva, Switzerland**
- **WHO, 2006.** *World Health Assembly Resolution on Sickle Cell Anaemia.* **WHA59.20. WHO, Geneva, Switzerland**
- **WHO, 2006.** *Executive Board Resolution on Thalassaemia and Other Haemoglobinopathies.* **EB118.R1. WHO, Geneva, Switzerland**
- **WHO, 2006.** *Report of a joint WHO/MOD meeting on Management of Birth Defects and Haemoglobin Disorders.* **WHO, Geneva, Switzerland.**
- **WHO, 2007. Report of Joint WHO/TIF Meeting.** ***Management of Haemoglobin Disorders*****, Nicosia, Cyprus, 16-18 November 2007**
- WHO, 2010 *Report by Secretariat to World Health Assembly: Birth defects.* ***WHA63.17,*** **WHO, Geneva, Switzerland ….**

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