

OXFORD



**2nd CAPABILITY Workshop
Oxford, UK
May 21 -23, 2008**



CAPABILITY is a 3-year (2007-2009) model project developed jointly by Eurogentest's units 3 & 6 and by leading experts from: *Argentina, Egypt and South Africa*, the latter being currently engaged in major development projects to integrate genetic services in primary care and prevention in their countries.

Funded by EC Contract no.: FP6-037275



EuroGentest

Genetic testing in Europe -
A network for test development,
harmonization, validation and
standardization of services

FP6 2005-2009

www.EuroGentest.org

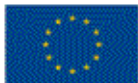


Major aims of the NoE

- Harmonization and quality improvement of genetic services
- Defining the European dimensions which are compatible with and complementary to national approaches

FOUR major domains of activities

- Quality of the laboratories
- Quality of the clinical aspects of the services
- Translation of technologies into diagnostic practice
- Educational aspects



Issues to start from

- More than **1,000 laboratories/centers** in different settings
- More than **1,000 rare diseases** can be tested

- Lack of **centralized and uniform information** about services
- Limited networking

- Lack of harmonized and standardized PT/EQA
- Lack of reference materials
- Limited number of ISO 15189 **accredited labs**

- Limited analytical and clinical validation of tests
- Diverse regulatory environments

- Insufficient **counselling**/lack of harmonized approach

Source: IPTS/JRC-EC: Ibarreta et al. Towards quality assurance and harmonization of genetic testing services in the EU.

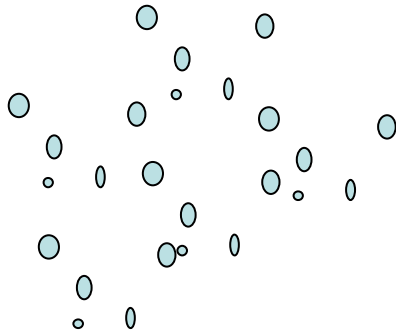
Report EUR20977, 2003



Euro**Gentest**

Present

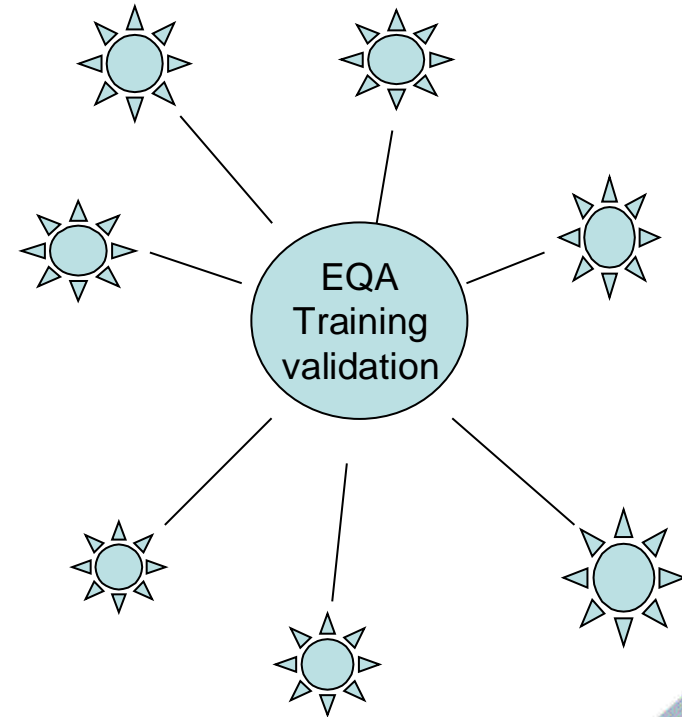
individual facilities



Future

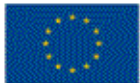
Virtual Institute:

Structured Network of expert
and peripheral centers



Highly variable quality
Individual rules and procedures

Common goals for quality, criteria, training
Centralized information for all
EQA provision
Validation of new technologies





Genetic testing in Europe

Network for test development, harmonization, validation and standardization



Quality Management

Information Databases

Public Health

Ethical & Legal

New Technologies

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Find out more about the world of Genetic Testing!

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Project Activities

Welcome to the EuroGentest website. We aim to provide you up to date general information on genetic testing in Europe, together with specific insight on how we are working to harmonise standards across the EU. Whatever your particular interest, click on the appropriate square to find out more.

Qm Quality Management	Id Information Databases	Ph Public Health
Et Ethics & Legal	Nt New Technologies	Ed Education

Genetic testing touches us all

As science unravels the genetic basis of disease, the opportunities offered by increased genetic testing become more and more apparent. EuroGentest is an EU-funded project that aims to ensure these opportunities are secured for the benefit of all.

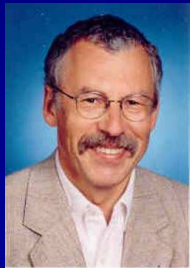
EuroGentest is organised as a Network of Excellence with 6 Units looking at all aspects of genetic testing – Quality Management, Information Databases, Public Health, New Technologies and Education. Through a series of initiatives EuroGentest encourages the harmonisation of standards and practice in



UNIT 3: CLINICAL GENETICS, COMMUNITY GENETICS AND PUBLIC HEALTH



Unit Leader: Ulf Kristoffersson, Head of the Department of Clinical Genetics at the University Hospital in Lund, Sweden
(Participant 2)



Co-leader: Joerg Schmidtke, Director of the Institute of Human Genetics at Hannover Medical School, Germany
(Participant 3)

Clinical validity and utility of genetic testing

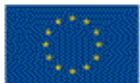
- Defining **clinical utility** criteria of genetic testing
- Investigating access to and utilisation of genetic services in EU
- Initiating an international collaborative network in order to promote internationally shared quality standards for clinical utility assessment
- Investigating criteria and mechanisms to maximize the output of genetic services, given limited resources
- Validate and translate the **utility guidelines** developed by the GSHG

Genetic Counselling

- Aim to improve the quality of genetic counselling

Specific Support Action to integrate partners from non-EU countries

“CAPABILITY Project” (2007-2009)



UNIT 6: EDUCATION



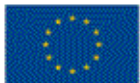
Unit Leader: Alastair Kent, Director of GIG and President of the European Alliance of Genetic Support Groups, London, UK
(Participant 4)

Education and patient issues

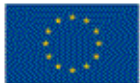
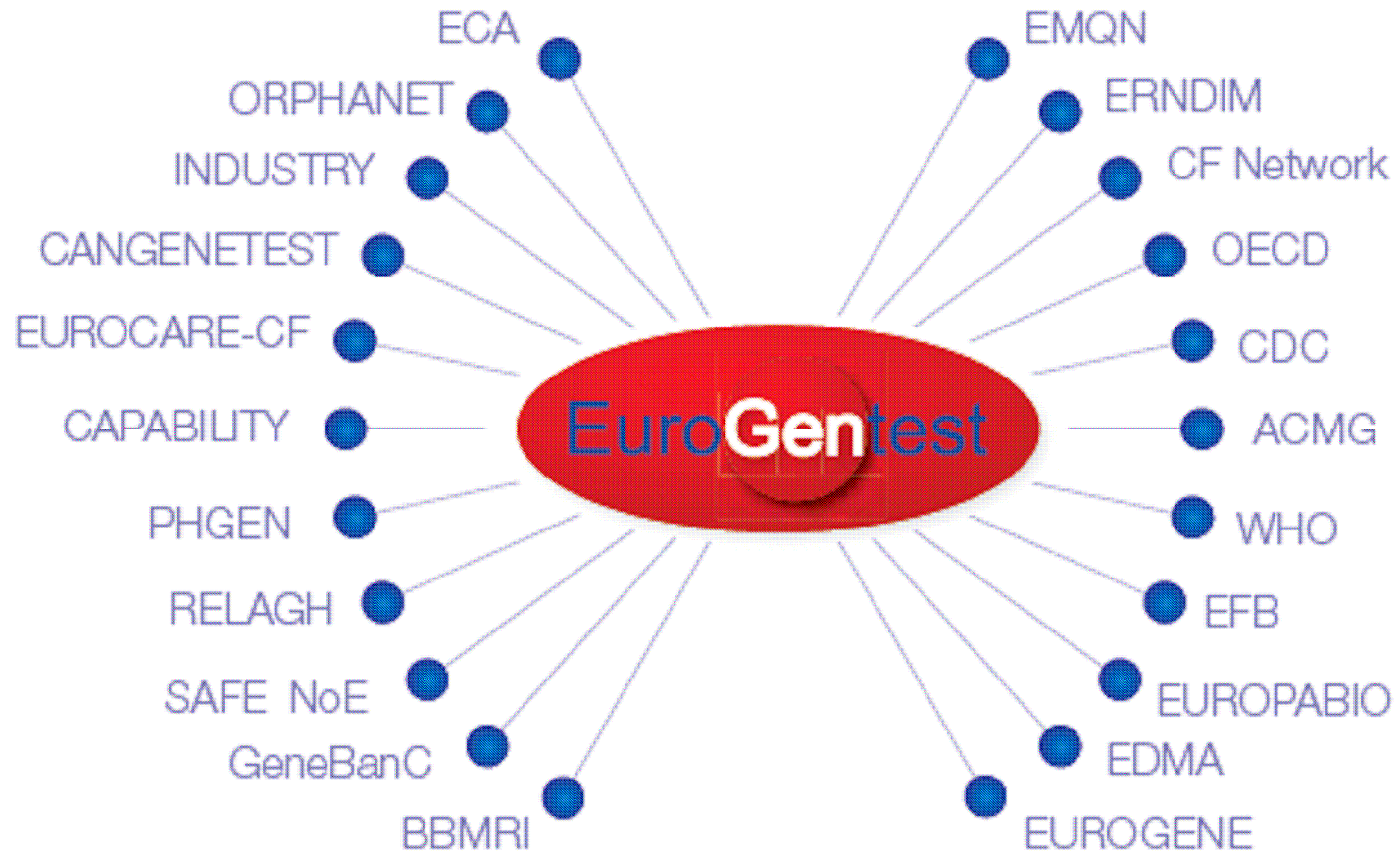
- Updated list of National **Societies** of Genetics and others relevant ones
- List of European and National **genetic courses**
- List of other **educational material** and list of relevant institutions involved in genetics education
- Network of **national contact persons** in the European countries
- 1st Workshop on “Patient and Professional Perspective of Genetic Information/Education in Europe”
- **Core competences** for geneticists and allied health professionals



Developing informative guides



Creating a network of networks



CAPABILITY's overall objectives are to contribute to:

- **the efforts to establish and sustain a worldwide harmonisation process for quality standards for the integration of genetic test/genomic knowledge applications into practice and prevention and**
- **to serve as a model project for successful, sustainable collaboration between EU research centres and centres from non-EU countries, especially emerging economies (middle income countries).**

CAPABILITY

International Participants...





NATIONAL HEALTH
LABORATORY SERVICE

The Department of Human Genetics

School of Pathology, Faculty of Health Sciences,
University of the Witwatersrand



Arnold L. Christianson

The Clinical Diagnostic and Genetic Counselling
Division

Medical Genetic Services

CAPABILITY Demonstration Project 1
(Participant 5)



EuroGentest

Greater Sekhukhune Outreach Programme: an evidence based approach for developing medical genetic services in South Africa

Project Leader: Arnold L. Christianson




جمهورية مصر العربية
وزارة الصحة و السكان

اتصل بنا
 خريطة الموقع
 زيارته

أمانة الوزارة | عن الوزارة | إحصائيات

ابحت | أدوية | التلقيح الصحي | هيئات ومراكز الوزارة

الأخبار :

- 10 آلاف تكوّن من فرنسا لمصر
- إحتفال وإشياء 59 مجتمع طبي بالمعاقلات
- خمس وحدات للمناظير وإفتتاحات بالمدينة

• الحركة الإستثنائية لترشيح للتخصصات الطبية المختلفة مايو 2006

• جمعية الصحة العالمية الثامنة و الخمسين تتيح اللوائح الصحية و الدولية

• دراسة عناصر تدبير الخدمة

• نتيجة الممولين بالزمانة




Ministry of Health and Population



Randa Kamal Abdel-Raouf, Children with Special Needs Department.
CAPABILITY Demonstration Project 2
(Participant 6)

Community Education in Health Aspects of Genetics

Project Leader: Randa Kamal Abdel-Raouf

Capacity Building Project (Community Education in Health Aspects of Genetics)

Goal:

- ❖ To develop and implement a module for community education on the prevention and care of genetic disorders.

Phases of the Project:

I. Preparatory Phase:

1) Selection of:

- a) setting
- b) target population

❖ Criteria for selection of the setting:

- Rural area
- Closed Community
- Inaccessible tertiary care
- High prevalence of genetic disorders

❖ Criterion for selection of target population:

- Persons who have an influence on the community:

a) Community Leaders:

- Mayor of the village
- Emam, Priest
- Members of popular assembly

b) Traditional birth attendants (Daya)

c) Influential persons in the family:

- Mothers, mothers in law, grandmothers

2) Situation analysis: Study of the sociodemographic characteristics of the selected community

- * Education
- * occupation
- * resources
- * Family size and family planning
- * Consanguinity
- * M/F ratio
- * Culture (beliefs, behaviors, attitude)

*Hospital de Pediatría S.A.M.I.C.
"Prof. Dr. Juan P. Garrahan"*

*Buenos Aires
Argentina*



Cristina Barreiro, Medical Genetics Service.
CAPABILITY Demonstration Project 3
(Participant 7)

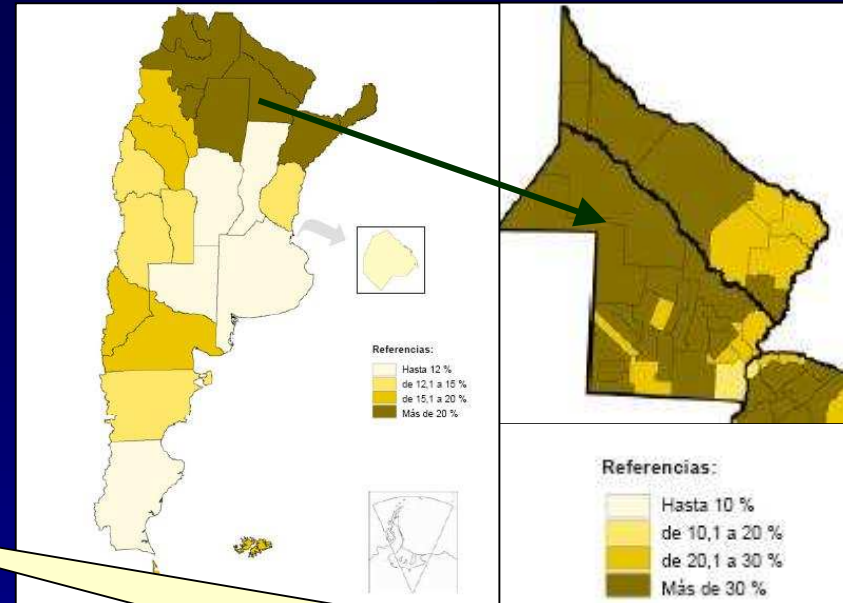
Chaco “Las Breñas”

Project Leader: Cristina Barreiro

The province of Chaco was chosen for a demonstration project

Local Characteristics:

- **Office for Distance Communication** connecting local health care centers and the Garrahan Hospital in Buenos Aires.
- **Public Health Project:** designed to improve quality of medical assistance.
- **Current Project: On-line Clinical Records (HCUA)**
- **Well-functioning Referral System** (ambulances, cars)



Our aim is to develop a program of genetic diagnosis and counseling in the province of Chaco through capacity building

When social conditions are hard, the burden of genetic disease is increased



Co-ordinator: Irmgard Nippert,
Women's Health Research,
Department of Human Genetics.
(Participant 1)

Co-ordination Office



Karina Bertmaring
Secretariat



Luis Robles-Treviño
IT & Communications

CAPABILITY

International Advisory Board...





The PHG Foundation is an independent international charity working to achieve the responsible and evidence-based application of biomedical science for health

We undertake and fund research, policy analysis, education and service development projects

About us
[Click here to find out more](#)

Resources
for researchers and health professionals

 Recent highlights

- Newly updated policy database
- Report: an assessment of the Human Tissue Act
- Report: Array CGH in the diagnosis of learning disability
- Biomarkers in familial colorectal cancer

 RSS feeds

- Recently added items
- News articles
- Upcoming events

Latest news [Go to news page](#)

-  Human Tissue and Embryos (Draft) Bill: Joint Parliamentary Committee publishes call for evidence
-  Genome sequencing cost drops below US \$1 million
-  UK Biobank Ethics and Governance Council reports on its work
-  New breast cancer susceptibility genes identified
-  Genetic disorders information resources
-  NICE report on only in research status for health interventions

Our new book

Genetics, Health Care and Public Policy: An Introduction to Public Health Genetics

This book is an introduction to the new discipline of public health genetics. It brings together the insights of genetic and molecular science as a means of protecting and improving the health of the population.



[View details on Amazon](#)



Ron Zimmern, Director PHG Foundation, Cambridge, UK.

Organización Panamericana de la Salud | REPRESENTACION ARGENTINA

Martes 5 de Junio de 2007

Solidaridad

Acerca de OPS/OMS
 Acerca de OPS
 Acerca de OMS
 Preguntas Frecuentes
 Días Mundiales

Links institucionales
 Sede de la ONU
 Sede de la OPS
 Oficinas en los países
 Centros Panamericanos
 Centros Colaboradores
 ONU en Argentina
 Organismos Oficiales

Fuentes de información
 BVS Argentina
 BIREME
 RENICS
 REMAR
 RECIARIA
 CEPS
 PALTEX
 Softwares

Noticias

31 de mayo de 2007 - Día Mundial sin Tabaco
OPS ENTREGA PREMIOS A MINISTERIOS DE SALUD DE CORDOBA, SANTA FE Y TUCUMAN
 La Organización Panamericana de la Salud celebrará este año el Día Mundial Sin Tabaco transmitiendo una videoconferencia el día 31 de mayo, a las 11:30hs., en la Oficina de la Comisión Económica para América Latina y el Caribe en Buenos Aires

Una llamada a la acción para que no haya humo en los lugares cerrados de uso público
 Los espacios 100% libres de humo de tabaco ya son lo normal. Este año el Día Mundial Sin Tabaco se enfoca en los ambientes 100% libres de humo de tabaco, como la única medida efectiva de proteger al público – incluidas mujeres y niños, y las personas en su lugar de trabajo – de la exposición al humo de tabaco ajeno.

WORLD HEALTH STATISTICS 2007
 La OMS ha publicado el World Health Statistics 2007, el sistema más completo de estadísticas de la salud de sus 193 estados miembros. Esta edición también destaca tendencias de las 10 variables estadísticas de salud observadas. Es la referencia anual de un sistema de 50 indicadores de la salud en países alrededor del mundo.

OPS anuncia el XI Congreso CREAD/MERCOSUR 2007 Educación a Distancia,


Destacados
 NOTICIAS, DOCUMENTOS Y ENLACES DESTACADOS
 Documento sobre Análisis de situación - Estrategias de Cooperación Técnica OPS/Argentina 2006-2007 [analisis de situacion.pdf](#)
 FORO Latinoamericano y del Caribe en VIH/SIDA e ITS

Publicaciones
 INFORMACION SOBRE INFLUENZA

Publicaciones
 Preguntas frecuentes sobre la pandemia <http://www.ops.org.ar/pandemia.asp>
 Desastres



Victor B. Penschaszadeh, Program on Genetics in Public Health and Human Rights, Pan American Health Organization (PAHO).



MARY SEACOLE
1805 - 1881

VOTED THE GREATEST BLACK BRITON

ABOUT MARY SEACOLE | STATUE APPEAL | CENTRE FOR NURSING PRACTICE | CONTACT | RESOURCES

MARY SEACOLE WAS A NURSE AND CARER WHO WENT TO WAR IN THE 1850'S. Her *adventures* took her far from home, the island of Jamaica in the Caribbean. She met members of the *British Royal Family*, and was *cheered by thousands* of people in London. She wrote a book about her life, telling how she helped soldiers during the Crimean War.




LATEST NEWS

FORTHCOMING EVENTS

MARY SEACOLE 1805-1881
Jamaican Nurse who went to war in the Crimea

PLACES WHERE MARY SEACOLE IS REMEMBERED

RACE IN VICTORIAN BRITAIN

MARY'S JOURNEY: [interactive map]

MEDICINE IN THE VICTORIAN AGE

HISTORY OF THE CRIMEAN WAR

1805
TIMELINE
1881

GALLERY



Elizabeth Nneka Anionwu, RN HV Tutor PhD CBE, Professor of Nursing, Head of Mary Seacole Centre for Nursing Practice, Thames Valley University, London, UK.

Advisory Board



Hilary Harris, MD, is the senior partner in a South Manchester general practice, UK.

CAPABILITY

International Observers...



A light blue world map is centered in the background of the slide. The text is overlaid on the map.

World Alliance *of* Organizations

for the prevention and treatment *of*
genetic *and* congenital conditions





- **Stephen T.S. Lam**
Clinical Genetic Service Centre, Department of Health, Hong Kong Special Administrative Region.
Secretary of Asia Pacific Society of Human Genetics (APSHG).

Asia Pacific Society of Human Genetics (APSHG)



Represented by Carmencita Padilla





**Nanbert Zhong, Director, Chairman
Peking University Center of Medical Genetics
Beijing, China**

**New York State Office of Mental Retardation and
Developmental Disabilities.**

**Institute for Basic Research in Developmental Disabilities.
Staten Island, New York, USA.**



Evaluator

March of Dimes, Global Programs White Plains, USA



Represented by Christopher Howson – Vice President

<http://www.marchofdimes.com/professionals/>



CAPABILITY Components

**Multidisciplinary
International
Experts**

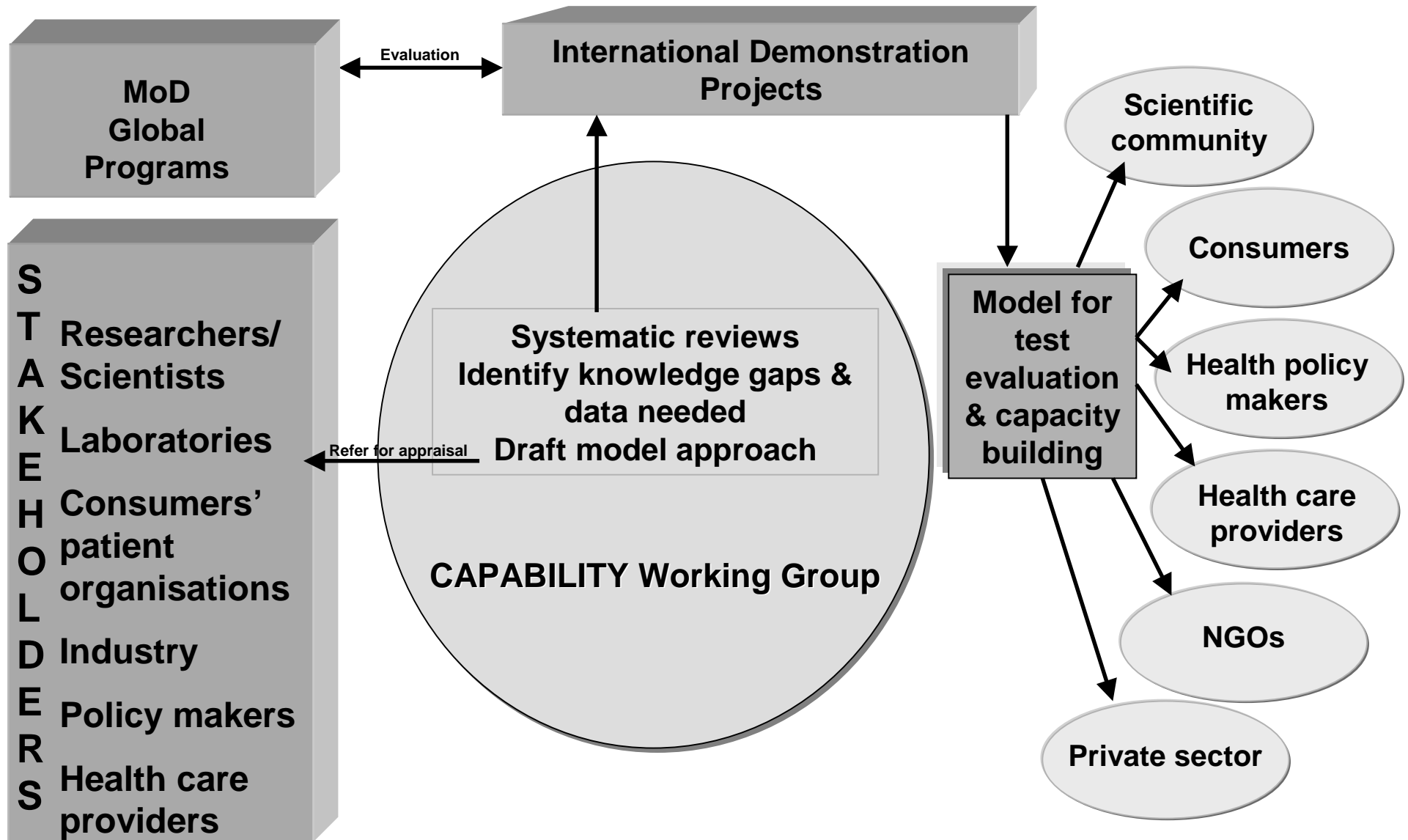
Representing:

- clinical genetics
- genetic epidemiology
- health service research
- health technology assessment
 - evidence-based medicine
 - molecular genetics
- parent and patient organisations
- primary care providers
- public health genetics
- ethics

Regular workshops & international meetings



CAPABILITY Overview
















**1st CAPABILITY Workshop
Trolleholm, Sweden
May 7 – 9th, 2007**






Home Participants Expert Advisors Demonstration Projects Meetings Documents

Home	Participants	Expert Advisors	Demonstration Projects	Meetings	Documents
	The CAPABILITY Project		1st. CAPABILITY Workshop / Trolleholm, Sweden	I. Nippert	May 7-9th, 2007
	CAPABILITYNET		1st. CAPABILITY Workshop / Trolleholm, Sweden	L. Robles-Treviño	May 7-9th, 2007
	Proposed Greater Sekhukhune Outreach Programme: an evidence based approach for developing medical genetic services in South Africa		1st. CAPABILITY Workshop / Trolleholm, Sweden	A. Christianson	May 7-9th, 2007
	Current strategies for implementing and strengthening public health genetics in Egypt		1st. CAPABILITY Workshop / Trolleholm, Sweden	R. Raouf	May 7-9th, 2007
	Genetic service provision in Argentina		1st. CAPABILITY Workshop / Trolleholm, Sweden	C. Barreiro	May 7-9th, 2007
	Translating genetic tests from research into practice in Germany: Cascade screening for hereditary nonpolyposis colorectal cancer – a model approach		1st. CAPABILITY Workshop / Trolleholm, Sweden	J. Schmidtke	May 7-9th, 2007
	Public Health Genomics Programmes and Policies in the UK		1st. CAPABILITY Workshop / Trolleholm, Sweden	R. Zimmern	May 7-9th, 2007
	Genetic testing in Europe: a network for test development, harmonization, validation and standardization of services		1st. CAPABILITY Workshop / Trolleholm, Sweden	M. Macek	May 7-9th, 2007
	Genetic Disorders and Congenital Defects in Latin America and the Caribbean		1st. CAPABILITY Workshop / Trolleholm, Sweden	Victor B. Penchaszadeh	May 7-9th, 2007
	Medical Genetics in Hong Kong & Networking in Asia		1st. CAPABILITY Workshop / Trolleholm, Sweden	Stephen TS Lam	May 7-9th, 2007
	March of Dimes Global Programs		1st. CAPABILITY Workshop / Trolleholm, Sweden	C. Howson	May 7-9th, 2007

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Subcommittee 1 Tasks

Chair: Joerg Schmidtke

- **systematic review of existing approaches of genetic test evaluation with an emphasis on clinical utility of tests with a potential to be implemented in general practice care and prevention**
- **included in the review will be genetic test evaluation approaches developed in the USA such as ACCE and EGAPP and the approaches of the United Kingdom Genetic Testing Network and the reviews done so far by Unit 3 of EuroGentest**

Subcommittee 1 Tasks

- **Draft of an analytic framework (=questions that need to be addressed) for evaluating genetic test application in different settings**

Participant 3 will prepare the draft for the CAPABILITY Working Group

Subcommittee 2 Tasks

Chair: Arnold L. Christianson

- **will develop criteria for a systematic needs assessment survey**
- **will identify priorities for capacity building by needs assessment survey**
- **will recommend capacity building mechanisms**

Participant 5 will prepare the report for the CAPABILITY Working Group



CAPABILITY presentation

Event during the "3rd International Conference on Birth Defects and Disabilities in the Developing World"
Conference: "Capacity Building for the Transfer of Genetic Knowledge into Practice and Prevention: The CAPABILITY Project"
Date: Monday June 18th, 2007 - 14:00hrs.
[More Info](#)

CAPABILITY Subcommittee 2 Preparatory Meeting

Date: June 20th, 2007
Time: 18:45 hrs.
Venue: Yellow Room, Copacabana Palace, Avenida Atlântica 1702, Rio de Janeiro, Brasil



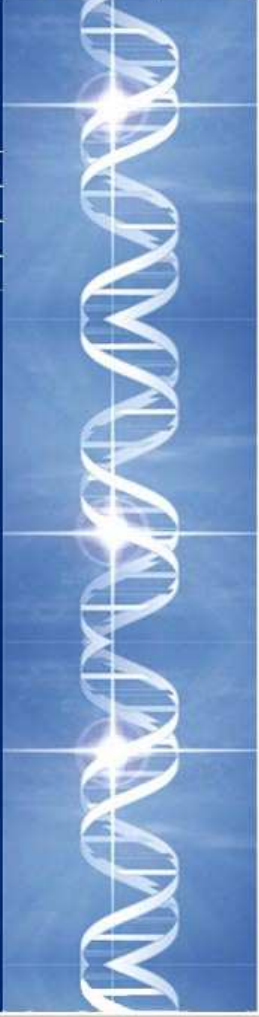
CAPABILITY Subcommittee 1 1st Meeting

Venue: Chesa Laret, Sils Maria, Switzerland
Date: September 21st, 2007

[Presentation](#)



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**WORKSHOP "CLINICAL VALIDITY AND UTILITY OF
GENETIC SUSCEPTIBILITY TESTING IN
COMMON DISORDERS"
OCTOBER 8-10, 2007, SEVILLE, SPAIN**

**CAPABILITY Subcommittee 2
1st Meeting**
Venue: Seville, Spain
Date: October 8th, 2007

Presentation



PRELIMINARY ANNOUNCEMENT

**Fourth International Conference on Birth Defects
and Disabilities in the Developing World**

New Delhi, India 4-7 October 2009

CONFERENCE THEMES

The conference will be organized around five central themes. A focus of each will be on the presentation of successful models of care and prevention around the world.

- 1. Surveillance, monitoring and needs assessment;**
- 2. Care of neonates and children with birth defects and disabilities;**
- 3. Prevention of birth defects and disabilities;**
- 4. Local, regional and global networks to strengthen the quality of health service delivery and promote community awareness; and**
- 5. Clinical genetics in its broad sense, including dysmorphology, metabolic disorders, thalassemia, muscular dystrophy and mental retardation, genetic predisposition to common chronic illnesses, etc.**

The conference will be structured to encourage dialogue and the sharing of experiences among health care providers, researchers, parent/patient group representatives, policy makers and other stakeholders from developing and industrialized countries.

CALL FOR ABSTRACTS

A Call for Abstracts will be issued in July 2008 and early submission is encouraged. A proportion of travel and per diem expenses, to be determined, will be awarded to those whose abstracts are accepted for oral presentation and who require funding



Thank you for your attention!