



**1st Central and Eastern European Summit on
Preconception Health and Prevention of Birth Defects**

Budapest, Hungary, August 27-30, 2008

Capacity Building for the Transfer of Genetic Knowledge into Practice and Prevention

The CAPABILITY Project

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The Capability Project

the challenge

eurogentest

capability

3 models

summary



International Partners



The Challenge...

Genetic tests &

genetic knowledge applications:

a. increase in number and complexity

b. increase in knowledge about
gene-disease associations

leading to new opportunities

in practice & prevention

c. broader population based applications

d. move into primary care & prevention services



The Challenge...

- **Genetic testing**
 - currently highly fragmental
 - diverse and heterogeneous quality schemes
 - lack of adequate counselling
- **Widening gap between the technology led supply of testing and services to meet patient needs**
- **Rising demands for genetic services**



The Challenge...

- Lack of health professionals educated in genetics
- Lack of an informed public (genetic illiteracy)
- Evaluation of benefits before introduction



International Partners



Worldwide, health care systems are facing the same challenges:

- The need
 - to develop an evidence-based **evaluation process** in transition from research into practice.
 - for **capacity building** to enable health care systems to make effective use of genetic/genomic applications with proven clinical utility.



EU Funded Projects



A European Network of Excellence aiming at harmonizing genetic testing services (2005-2009)

www.eurogentest.org



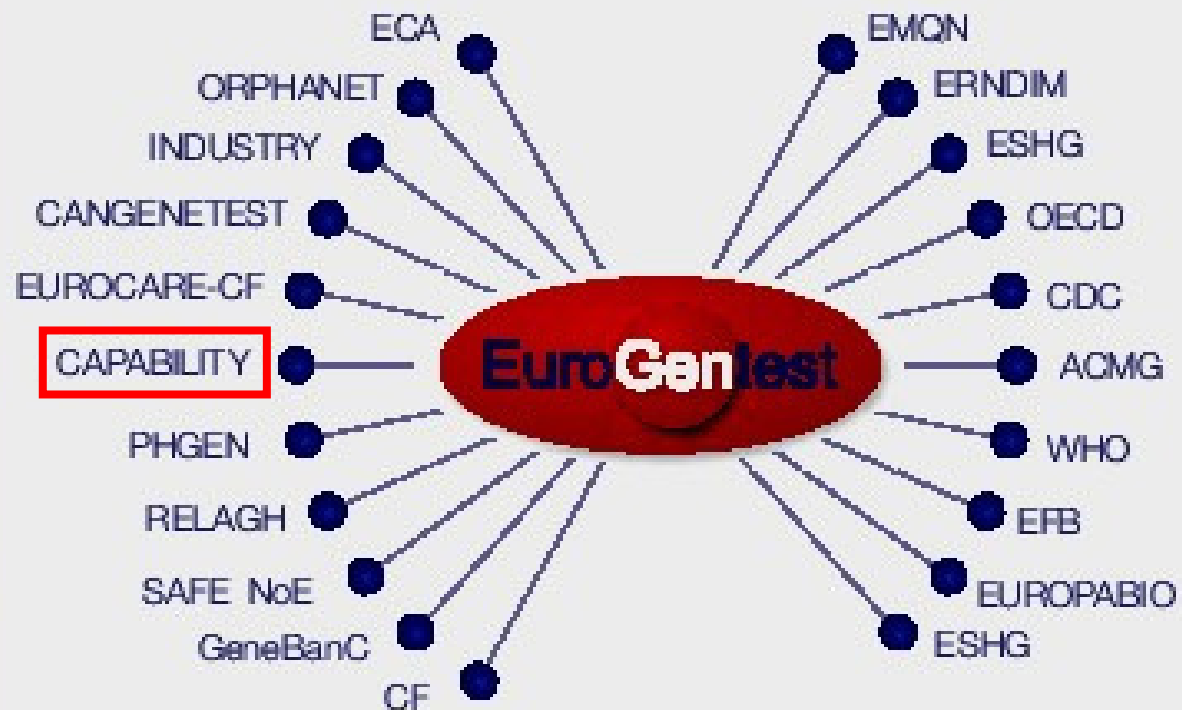
International Partners



EuroGentest

creating a network of networks

EuroGentest has become in effect a “network of networks” through collaborations with various national and international bodies involved in genetic testing. These range from the OECD through the European Society of Human Genetics to patient interest groups. Through such interactions, we aim to accelerate the harmonization of genetic testing services throughout the EU and associated states.





- Establish a network of quality across Europe
- Promote research, adequate management of genetic services
- Establish procedures and guidelines for the validation of methods and technologies



WHO recommendations



World Health Organization

Genomic resource centre

Case Study: European Union

It may be beneficial in the longer term for other jurisdictions, such as the Latin American countries, the Middle East, South Africa, to participate in similar harmonization activities, as they may benefit more from this model than from one developed for a single country (e.g. the United States).

Source: <http://www.who.int/genomics/policy/eu/en/index.html>, accessed: 03/06/2006



Capacity Building for the Transfer of Genetic Knowledge into Practice and Prevention: The CAPABILITY Project



International
Partners



**CAPABILITY: a 3 year model project
(2007-2009)**

Funded by FP6 - 037275



CAPABILITY Methods

- **Bring together key stakeholders**
academics, clinicians, policy makers,
(patient organizations)
- **Seek agreement on standards**



CAPABILITY Objectives

- Identify priorities by systematic needs assessment surveys
- Evaluate the approach via demonstration projects to be conducted at 4 target sites



CAPABILITY is a 3-year model project developed jointly by Eurogentest's units 3 & 6.

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)



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UNIT 6: EDUCATION



Unit Leader: Alastair Kent, Director of GIG and President of the European Genetic Alliances' Network & Genetic Interest Groups, London, UK
(Participant 4)



CAPABILITY Co-ordinator:



Irmgard Nippert,
Women's Health Research,
Department of Genetics,
Muenster, Germany
Participant 1

(EuroGentest Participant 35, Unit 3 WP 3.3)



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

(Participant 5)



Objectives

CAPABILITY Demonstration Project – Limpopo, South Africa

- **test and develop the principles and practices of primary health care based medical genetic services** as outlined in the National Department of Health's national policy guidelines and in the CAPABILITY model approach.
- re-evaluate the **epidemiology** of congenital disorders in this setting



- test the **clinical utility, including cost efficiency**, of DNA based medical genetic tests and technologies (QF-PCR, MLPA, Microarray CGH when available) in these circumstances
- use the knowledge and experience gained from the project to assist the **implementation and development of medical genetic services** throughout Limpopo and other provinces in South Africa.

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الأخبار:

- ◆ 10 آلاف كتكوت من فرنسا لمصر
- ◆ إحلال وإنشاء 59 مجمع طبي بالمعافظات
- ◆ خمس وحدات للمناظير وافتتاحات بالمنيرة

الحركة الإستثنائية للترشيح
للخصصات الطبية المختلفة مايو
2006

- ◆ جمعية الصحة العالمية الثامنة و الخمسين تنبج اللوائح الصحية و الدولية
- ◆ دراسة عناصر تقديم الخدمة بالزمالة
- ◆ نتيجة المتبولين




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

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, grand-mothers

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.
(Participant 7)

CAPABILITY Demonstration Project 3



Some Problems in Argentina

- Economic difficulties which affect the country are a hindrance to progress in the field.
- Until recently, government officials were not considering genetic services as a priority.
- Therefore, the development of clinical genetics as well as the access of the population to specialized care has been deficient.

Confronting the Problems

- Need for **training** of more professionals in the different genetic sub-disciplines and the creation of **hospital positions** in the neglected areas of the country.
- Development of a regional network of **genetic services**, with interconnected levels of complexity and roots at the primary health care level

Recent Actions of the Ministry of Health

Creation of

- a National Program of Community Physicians for Primary Health Care (genetic training is pending)
- a National Commission Genetics and Health
- implementation of a
 - needs assessment of genetic services
 - regionalized network of genetic services

CAPABILITY

Argentina

Pilot project: Chaco “Las Breñas”

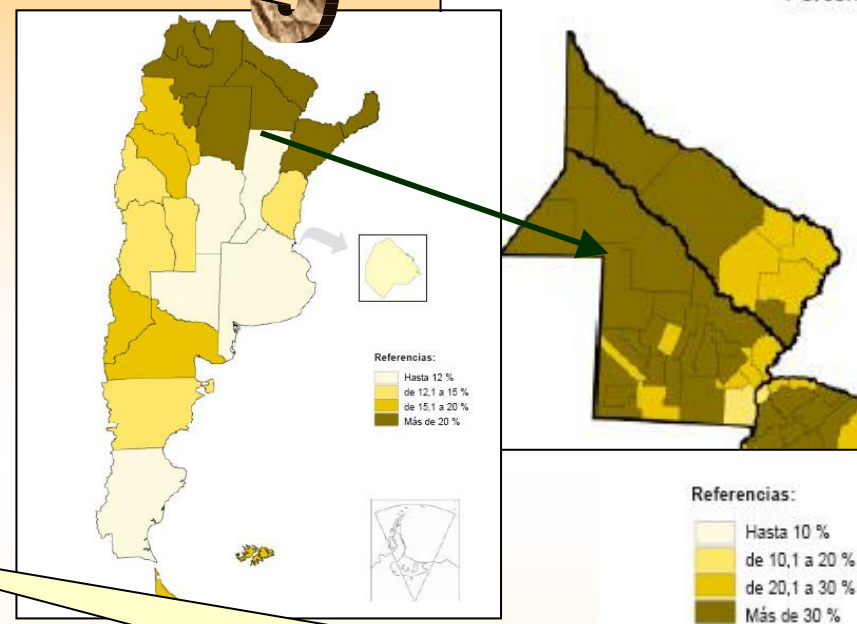


CAPABILITY Argentina

- 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

**A key objective of CAPABILITY is
to engage the interest of a wide
range of stakeholders**



International
Partners



CAPABILITY Networking Goals

- **To promote**
 - an internationally shared set of basic quality standards for genetic testing / genetic knowledge applications and
 - the provision of appropriate genetic services in primary care and public health
- **To lay the foundation for a sustained collaboration, that will continue and be open for international partners**
- **To start collaboration by joint projects**



CAPABILITY Objectives

- Disseminate the model approach to targeted audiences



International Partners



CAPABILITY will help

- To promote
 - proper implementation of new genetic knowledge, quality assurance and
 - adequate provision of genetic services at a global level
- To identify present and future needs for capacity building mechanisms



International Partners



CAPABILITY will help

- To enable health care systems to integrate genetic knowledge appropriately, based upon local needs and priorities
- To reduce inequalities in genetics health care between low- and middle- income and high-income countries



CAPABILITY Website



**Thank you
for your attention!**

