Partnership for Perinatal Health Networks

First Annual Meeting

March of Dimes Foundation

White Plains, NY
17-18 December 2007
Capacity Building for the Transfer of Genetic Knowledge into Practice and Prevention

The CAPABILITY Project

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The Challenge…

• Genetic tests and genetic knowledge applications are increasing in number and complexity

• Increasing knowledge about gene-disease associations will lead to new opportunities to apply genetic knowledge in practice and prevention
The Challenge...

- A growing number of genetic tests / genetic knowledge applications have broader population based applications.

- Genetic testing / genetic knowledge applications will move into primary care and prevention services.
The Challenge...

- Genetic testing is currently highly fragmental
- Diverse and heterogeneous quality schemes
- Lack of adequate counselling
The Challenge…

• 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)
Before genetic tests are introduced into general practice the benefits of their use must be evaluated.
Worldwide, health care systems are facing the same challenges:

• The need to develop an evidence-based evaluation process for genetic tests or other applications of genetic knowledge in transition from research into practice.

• The need for capacity building to enable health care systems to make effective use of genetic/genomic applications with proven clinical utility.
Evolution to CAPABILITY

• Reports and recommendations
  
  ➢ Institute for Prospective Technological Studies (Seville 2003)
Genetic testing in Europe, while based upon high quality scientific know-how, suffers from disorganisation and fragmentation of services, caused by a lack of structuring and complementarity at the European level and the absence of a common European objective to provide quality services to all its consumers:

“A European, or even higher international level, approach is required.”

(Towards quality assurance and harmonisation of genetic testing services in the EU, Report EUR 20977 EN, 2003 Executive Summary, Page 4.)
Evolution to CAPABILITY

• Reports and recommendations

  - Organisation for Economic Co-operation and Development (Paris 2005)
Organisation for Economic Co-operation and Development (Paris, France - 2005)

Recommendation:

The “internationalisation” of genetic testing for medical and research purposes and the establishment of genetic testing networks has become inevitable and necessary.

(Paris, 2005)
EU Funded Projects

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

www.eurogentest.org
Harmonizing genetic testing across Europe

What is EuroGentest?

EuroGentest is an EU-funded Network of Excellence (NoE) with 8 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 all these areas throughout the EU and beyond.

What can EuroGentest offer you?

- info for public
- info for laboratories
- info for industry
- info for clinical genetists
- info for students

Find a lab or test

Find Information on laboratories across Europe, their tests and their Quality Assurance (QA) data.

- Go to QA database
- Go to orphanet

How to register?

Find out more about the world of Genetic Testing by registering. You will get access to additional information if you

Roadshow: OECD Guidelines

New OECD Guidelines for Quality Assurance in Molecular Genetic Testing will set a framework for genetic testing services. EuroGentest members have been heavily involved in the preparation of these guidelines as well as other guidelines to harmonize the practice of genetic testing. EuroGentest is organizing a series of roadshow presentations at national level to explain to stakeholders the implications of the new guidelines and suggested pathways to implementation. The first is scheduled for Poland in September.

Upcoming Events

04 Sep 2007 Hamburg (DE), EURLNG Workshop on DPT schemes 2007 and EOA and diagnosis of peroxisomal disorders (WP1.5)

The 5 individual DPT meetings will be followed by a workshop for all EURLNG and open to SIEVA participants. The topic will be diagnostic aspects of peroxisomal disorders.

05 Sep 2007 Hamburg (DE), EURLNG Board meeting, Best practice in BGT testing (WP1.5)

Coordination and implementation of strategies and measures for best practice and improvement of quality of BGT testing and linking these to other EUGT activities.

Guidelines on counselling

As patients' understanding of the results and consequences of the test is an integral part of genetic testing, EuroGentest also aims at improving the
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, proper utilization, quality control and assurance and adequate management of genetic services
• Establish procedures and guidelines for the validation of methods and technologies

• Provide genetic healthcare workers, the end-users and healthcare authorities with a portfolio of quality-assured information sources and informatic tools that are subject to validation and quality procedures

In addition, EuroGentest aims at becoming a model for similar initiatives in developing countries and will provide appropriate support for their development.
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).

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

CAPABILITY: a 3 year model project (2007-2009)
Funded by FP6 - 037275
CAPABILITY is a Specific Support Action (SSA) for EuroGentest units 3 (Public Health) and 6 (Education)

Improving quality of genetic counselling, clinical validation and clinical utility

Generating shared skills and knowledge about genetic testing

Public Health

Education
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)
CAPABILITY is a 3-year model project developed jointly by EuroGentest’s units 3 & 6.

UNIT 6: EDUCATION

Unit Leader: Alastair Kent, Director of GIG and President of the European Alliance of Genetic Support 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)
Arnold L. Christianson
The Clinical Diagnostic and Genetic Counselling Division

(Participant 5)
Proposed Greater Sekhukhune Outreach Programme: an evidence based approach for developing medical genetic services in South Africa

• Arnold Christianson
• Division of Human Genetics
• National Health Laboratory Service &
• University of the Witwatersrand, Johannesburg, South Africa.

Source: CAPABILITY Website - http://www.capabilitynet.eu/
Limpopo

Demography

- No of births ~140 2900
- Fertility rate 3.7/woman (2.8)
- U5MR 57/1000 live births
- Life expectancy 56 years (50.7)
- Piped water 73.3%
  Sanitation 85.1%

Source: CAPABILITY Website - http://www.capabilitynet.eu/
Limpopo

Health care

• Antenatal Care coverage 93.4%
• Assisted birth 87.7%
• 88% clinics with Early Primary Immunization
• 97% clinics with Family Planing
• Doctors 780+75 specialists (26.8% posts vacant)
• Nurses 5827 (15% posts vacant)

Source: CAPABILITY Website - http://www.capabilitynet.eu/
Randa Kamal Abdel-Raouf, Children with Special Needs Department. CAPABILITY Demonstration Project 2 (Participant 6)
Genetic Services in Egypt

MOH&P

Primary Care
- 20% of 4000 PHC:
  - Early detection
  - Referral to genetic counseling clinics

Secondary Care
- 10 genetic counseling clinics:
  - Diagnosis
  - Counseling
  - Treatment
  - Referral for Rehabilitation

Tertiary Care
- 5 genetic depts/units in Universities
- Genetic Department in the National Research Center (NRC)
- Health Insurance Organization (HIO)
- Private Sector

NGO’s
- ≈10 organizations working in the field of genetics
- ≈ 120 organizations working in the field of MCHC

Source: CAPABILITY Website - http://www.capabilitynet.eu/
National Committee for Community Genetics / 2002

Objectives of the program:
• Provide Community genetic services
• Determine the prevalence of congenital and genetic disorders
• Raise community awareness

Genetic counseling clinics:
• 1 in Giza
• 3 in Cairo
• 4 in Alexandria
• 1 in Port-Saiid
• 1 in Sharkia
• 1 in Al-Minia
• 1 in Assiut

Supervision
• Genetic Dept. National Research Center/Giza
• Genetic Unit, Ain-Shams University/Cairo
• Genetic Dept. Alexandria University
Egyptian Module for Genetic Counseling Program

Genetic Depts.:
Universities and NRC
Training + Consultation + Supervision

Health insurance org.
Investigations + Treatment + Rehabilitation

Genetic counseling clinics
Diagnosis + Counseling + Follow-up

PHC facilities
Detection + Referral

Source: CAPABILITY Website - http://www.capabilitynet.eu/
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

Source: CAPABILITY Website - http://www.capabilitynet.eu/
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)
Cristina Barreiro, Medical Genetics Service.  
(Participant 7)  
CAPABILITY Demonstration Project 3
Genetic Centres in Argentina

(red dots)
Newborn Screening in Buenos Aires (90% coverage)

- Congenital hypothyroidism
- Phenylketonuria
- Cystic Fibrosis
- Congenital Adrenal Hyperplasia
- Galactosemia.
- Biotinidase Deficiency
- Chagas' disease
- Syphilis
- Congenital Hearing Loss

Source: CAPABILITY Website - http://www.capabilitynet.eu/
Some Problems

• 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)
- Creation of a National Commission of Genetics and Health
- Implementation of a needs assessment of genetic services based on a national survey conducted in 2006, to be followed soon by:
  - creation of new clinical positions and laboratory equipment and personnel
  - implementation of a regionalized network of genetic services

Source: CAPABILITY Website - http://www.capabilitynet.eu/
Pilot project: Chaco “Las Breñas”
Local Characteristics:

- **Chaco** has an infant mortality rate of 19.7% (INDEC 2005)
- Unsatisfied basic needs: > 30%
- Endogamic aboriginal population
- Geographically isolated regions: The province of Chaco was chosen for a demonstration project

- **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.
CAPABILITY Advisory Board...
Ron Zimmern, Director PHG Foundation, Cambridge, UK.
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.
• 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).
Evaluator

March of Dimes, Global Programs
White Plains, USA

Represented by Christopher Howson – Vice President

http://www.marchofdimes.com/professionals/
CAPABILITY Components
CAPABILITY Overview

MoD Global Programs

Researchers/Scientists Laboratories
Consumers’ patient organisations Industry
Policy makers Health care providers

Evaluation

International Demonstration Projects

Systematic reviews
Identify knowledge gaps & data needed
Draft model approach

CAPABILITY Working Group

Model for test evaluation & capacity building

Scientific community
Consumers
Health policy makers
Health care providers
NGOs
Private sector

Refer for appraisal
CAPABILITY Working Group

15 experts:
- 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 open for international attendants
CAPABILITY Methods

• Bring together key stakeholders for the systematic transfer of genetic testing / genetic knowledge applications from research into clinical practice by means of frameworks for an evaluation process agreed by academics, clinicians and policy makers

• Seek agreement on standards
CAPABILITY Objectives

• Identify priorities for capacity building for providing appropriate services by systematic needs assessment surveys

• Evaluate the approach via demonstration projects to be conducted at 4 target sites
A key objective of CAPABILITY is to engage the interest of a wide range of stakeholders.
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
• Disseminate the model approach to targeted audiences
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
CAPABILITY will help

- To enable health care systems to integrate genetic knowledge appropriately, based upon local needs and priorities

- To strengthen the kinds of basic capacities that will allow participating low- and middle-income countries to more easily incorporate the benefits of genetic/genomics research as they unfold

- To reduce inequalities in genetics health care between low- and middle-income and high-income countries
"Genetic Testing in Developing Countries: An international survey by the Institute for Prospective Technological Studies (IPTS) in collaboration with EuroGentest Unit 3 and its international CAPABILITY project (WP3.3)"

Dolores Ibarreta announced the new project at the EuroGentest General Assembly. Nov 21-23, 2007, Leuven, Belgium

The project will start in 2008
CAPABILITY

CAPABILITY is a 3-year model project developed jointly by Units 3 (Clinical Genetics, Community Genetics and Public Health) and 6 (Education) of the Network of Excellence Genetic Testing in Europe Network for test development, harmonization, validation and standardization of services (EuroGentest) 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.

CAPABILITY will:

- develop an analytic framework for evidence-based genetic test evaluation including the domains: efficacy (evidence of utility in controlled settings) and effectiveness (evidence of utility in real settings), identify priorities for capacity building by a systematic needs assessment survey and
- validate the project's approach by means of a demonstration project.

CAPABILITY's overall objectives are to contribute to the efforts to establish and sustain a worldwide harmonization 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 developing countries.

The following challenges are common to both EU and middle income countries:

1. The need to develop an evidence-based evaluation process for genetic tests or other...
Thank you for your attention!