LSSG-CT-2006-037275

CAPABILITY

Capacity Building for the Transfer of Genetic Knowledge into Practice and Prevention: An International Collaborative Network

Instrument **Specific Support Action**

Thematic Priority **Life sciences, genomics and biotechnology for health**

Title of report ***Final activity report***

Period covered: from 01.07.2008 to 31.12.2009 Date of preparation: 11.02.2010

Start date of project: 01.01.2007 Duration: 36 months

Project coordinator name: Univ.-Prof. Dr. Irmgard Nippert
Project coordinator organisation name: Westfälische Wilhelms-Universität Münster

Table of contents

Abbreviations

COVs	community outreach visitors
CUGC	Clinical Utility Gene Cards
EC	European Commission
ELSI	Ethical, Legal, and Social Issues
EMEA	European Agency for the Evaluation of Medicinal Products
ESHG	European Society of Human Genetics
EU	European Union
EuroGentest	Genetic Testing in Europe – Network for test development, harmonization, validation and standardization of services
GenTEE	Genetic Services in Emerging Economies
GIG	Genetic Interest Group
GraSCOP	Greater Sekhukhune-CAPABILITY Outreach Project
HNA	Health needs assessment
IHCP	Institute for Health and Consumer Protection
IPTS	Institute for Prospective Technological Studies
JRC	Joint Research Centre
KAP	Knowledge, Attitudes and Practice
LU	Lund University
MDG4	Millennium Development Goal 4
MGEP	Medical Genetic Education Programme
MHH	Medizinische Hochschule Hannover
MoH	Ministry of Health
MOHP	"Children with Special Needs" Department/Ministry of Health and Population
NHLS	National Health Laboratory Services Division
NoE	Network of Excellence
PAHO	Pan American Health Organization
SAMIC	Hospital de Pediatría SAMIC
SSA	Specific Support Action
UK	United Kingdom
UKGTN	United Kingdom Genetic Testing Network
WHO	World Health Organisation
WITS	University of the Witwatersrand
WWU	Westfälische Wilhelms-Universität

1. The CAPABILITY Project, Introduction and Overview

1.1 Background

Human genome research is leading to the rapidly increasing translation of genetic/genomic data into clinical application. Genetic tests[1] have been developed for the vast majority of recognised genetic conditions, including DNA-based tests for more than 1.100 diseases, with more than 1.000 currently available for clinical testing.

The number of DNA-based tests performed annually in Europe is now over 700.000 and is increasing. The economic dimensions of this activity is estimated around 900.000 € – 28.000.000 € / million citizens/year[2] and is expected to grow rapidly by the next decade. Although the majority of these tests are used for the diagnosis of rare disorders, ***a growing number of tests have broader population based applications*** including predictive testing for inherited risk for common disorders, carrier identification and pharmacogenetic testing for variations in drug response.

The use of genetic tests for diagnostic, confirmatory and predictive purposes is expanding in all European countries at an estimated growth rate between 100% and 300% per year.

These genetic tests and other anticipated applications of genome technologies for screening and prevention will impact on the future provision of primary care and prevention services.

The European Union (EU) faces the dual challenge of developing adequate standards for genetic testing while bridging the national, socio-economic and socio-cultural and linguistic differences of its Member States. Accordingly in its report "Towards quality assurance and harmonisation of genetic testing services in the EU" the Institute for Prospective Technological Studies (IPTS) states that genetic

[1] Genetic test = the analysis of human DNA, RNA, chromosomes, proteins, and certain metabolites in order to detect disease – related genotypes, mutations, phenotypes or karyotypes for clinical purposes, including: risk predictions and identifying carriers.
[2] Jean-Jacques Cassiman: "EuroGentest -Genetic Testing in Europe. A network for test development, harmonization, validation and standardization of services". Presented at the 19th Annual Meeting of the German Society of Human Genetics April 8-10, 2008

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. The European Commission's Expert Group on Genetic Testing, the IPTS and the European Society of Human Genetics (ESHG) have all expressed ***the need for harmonising quality standards within genetic testing and genetic service provision***. To meet this challenge the FP6 funded Network of Excellence (NoE), "*Genetic Testing in Europe – Network for test development, harmonization, validation and standardization of services*" (*EuroGentest*) was launched in 2005 (www.eurogentest.org). *EuroGentest* will end in June 2010. The World Health Organisation's (WHO) Genomic Resource Centre, when reviewing the current situation in the EU, recommended that ***it could be beneficial for other jurisdictions, such as countries from Latin America or developing countries such as South Africa, to participate in these EU harmonisation activities***, as they may benefit more from the EU approach than from one developed for a single country (e.g. the United States) and because these countries face similar challenges regarding adequate genetic service provision and quality assurance. **This comes at a time when the WHO's Executive Board has recommended the priorisation of genetic services in developing countries for the management and prevention of genetic/congenital disorders to assist them to attain their MDG4[3]**.

Middle income developing countries, such as those participating in this CAPABILITY project, are facing an epidemiological transition. As national health care systems improve an increasing proportion of morbidity and mortality is due to congenital and genetic conditions. Health care systems in these nations are starting to respond to the rising needs for genetic services in primary care and public health. Like in Europe, but to a greater extent, there is a scarcity of personnel trained in genetics. Services are predominately tertiary specialist medical genetic services, usually situated in academic centres because service evolution is highly research dependent. Consequently these services have been developed with little contact with primary care or community-based services. As in Europe the extraordinary diversity of the natural history and clinical implications of genetic conditions has favoured the

[3] WHO Executive Board, May 14th, 2009

development of disease specific services (e.g. for cystic fibrosis, haemoglobinopathies, metabolic disorders, neurological conditions, etc.), that often have little contact with each other and fail to recognise their commonalities. ***As in Europe, albeit with lower available resources, these countries need to develop the necessary infrastructure, tools, resources, guidelines and procedures leading to the establishment of harmonised quality medical genetic services.***

Because the use of genetic tests for diagnostic, confirmatory and predictive purposes is expanding worldwide. The following challenges are common to both EU and middle income countries:

i) The need to develop an evidence-based evaluation process for translating genetic tests or other applications of genomic knowledge into health care.

ii) The need for capacity building to enable health care systems to integrate adequately genetic/genomic applications with proven clinical utility into practice and prevention.

iii) The need to establish an evidence-based implementation process for genetic/genomic knowledge into routine practice, including the review of testing practices, to ensure quality standards of medical genetic services and to allow policy makers and governments informed decision making.

CAPABILITY is a first step to address these challenges.

1.2 Objectives and Methods

CAPABILITY addresses:

- the lack of internationally shared standards for medical genetic service provision including genetic testing.
- the lack of evidence-based models for assessing the need for medical genetic services.

CAPABILITY is a 3-year model project, funded from 2007-2009 by the European Commission (EC) FP6. It is a Specific Support Action (SSA) for the NoE *EuroGentest* (http://www.eurogentest.org).

The project was developed jointly by the NoE *EuroGentest* unit 3 "Clinical Genetics, Community Genetics and Public Health" represented by U. Kristoffersson, Lund University, Sweden (LU*)*, J. Schmidtke, Medizinische Hochschule Hannover, Germany (MHH) and I. Nippert, Westfälische Wilhelms-Universität Münster, Germany (WWU) who acted as co-ordinator of CAPABILITY and unit 6 "Education" leader A. Kent, Genetic Interest Group, London, U.K. (GIG) and by leading experts from: Argentina C. Barreiro, Hospital de Pediatría SAMIC, Buenos Aires (SAMIC), Egypt R. Raouf, "Children with Special Needs" Department/Ministry of Health and Population, Cairo (MOHP) and South Africa A. Christianson, University of the Witwatersrand, Johannesburg (WITS), the latter being currently engaged in major development projects to integrate genetic services in primary care and prevention in their countries.

There is a commonality of interests shared by the CAPABILITY participants to promote an internationally shared set of basic quality standards for genetic testing and the provision of adequate genetic services based upon systematic health needs assessment (HNA).

To achieve its objectives CAPABILITY has established an international multidisciplinary working group (members see Annex) representing expertise in:
clinical genetics, genetic epidemiology, health service research, health technology assessment, evidence-based medicine, molecular genetics, public health genetics

and ethics. The working group also includes representatives from parent and patient organisations and primary care providers.

The following tasks were performed by the CAPABILITY working group:

- examination of existing national and international genetic test evaluation approaches that could be adopted to evaluate the utility of genetic tests/genetic knowledge for health care in developing countries;
- identification of priorities for HNA in medical genetic services and priorities for capacity building;
- validation of its capacity building approach by means of a joint demonstration project sensitive to specific health care contexts and service patterns in developing countries, including the magnitude of assessed needs, available resources and capacities, service gaps, as well as ethical, socio-cultural and legal implications;
- dissemination of the report "Model Approaches for Capacity Building for the Translation of Genetic Knowledge into Practice and Prevention" to targeted audiences worldwide.

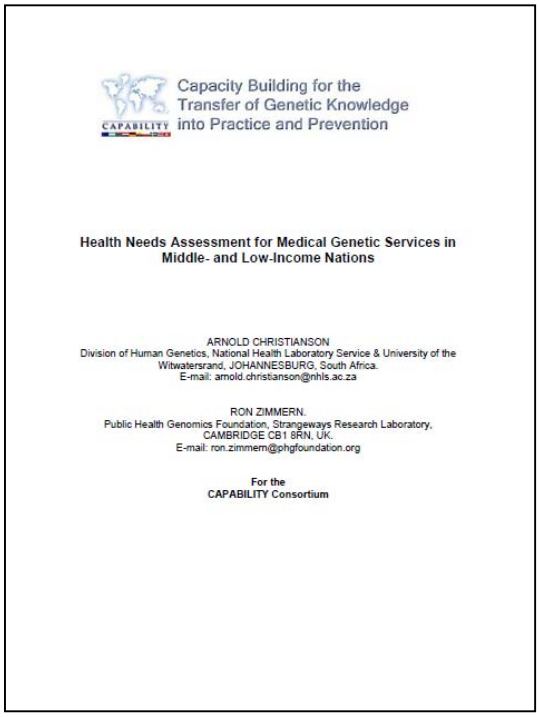
To start the project a systematic HNA for medical genetic services was conducted in Argentina, Egypt and South Africa. Because limited cogent literature on HNA and examples of its application to medical genetic services are available in the literature, CAPABILITY had to develop its own approach to HNA for medical genetic services with particular reference to middle- and low-income nations.

The CAPABILITY project set out to collect information taking into account: socio-demographic and epidemiological data, health care system data, situational analysis of the availability of services, gaps in service provision, ELSI implications taking into account national/regional cultural norms and practices and national laws and guidelines to identify health care needs in these countries and to plan the demonstration project.

The data obtained from Argentina, Egypt and South Africa were presented at an international experts workshop for discussion and critical assessment (needs assessment reports available on CAPABILITY website: http://www.capabilitynet.eu).

Subsequently small scale projects for capacity building were designed to address needs identified by the national/local surveys. Capacity projects were conducted in the Chaco Region Las Breñas, Argentina, Om-Khenan area in Giza governorate, Egypt and the Greater Sekhukhune district, Limpopo Province, South Africa.

In the light of the experiences of the conducted surveys and the input from international experts a general methodology for conducting HNA as a rational evidence-based foundation for capacity building for medical genetic services was developed by CAPABILITY (the report "HNA for Medical Genetic Services in Middle- and Low-Income Nations" is available on CAPABILITY website *http://www.capabilitynet.eu/Documents/Documents.html*).


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

Health Needs Assessment for Medical Genetic Services in Middle- and Low-Income Nations

ARNOLD CHRISTIANSON
Division of Human Genetics, National Health Laboratory Service & University of the Witwatersrand, JOHANNESBURG, South Africa.
E-mail: arnold.christianson@nhls.ac.za

RON ZIMMERN.
Public Health Genomics Foundation, Strangeways Research Laboratory, CAMBRIDGE CB1 8RN, UK.
E-mail: ron.zimmern@phgfoundation.org

For the
CAPABILITY Consortium

The project "Public Private Partnership" was conducted in Germany (J. Schmidtke, MHH). The project addresses the problem how to attract private funds in order to satisfy the public need for genetic services in countries with very limited resources (see below).

1.3 Demonstration project overview

1.3.1 Argentina - Implementing genetic services in the Chaco region of Argentina

(report available on CAPABILITY website: http://www.capabilitynet.eu/Documents/presentation/CAPABILTY%20Argentina%20CHACO%20Outreach%20Project.pdf)

The aim of the project was to design and implement a model for local delivery of medical genetic services in primary health care by means of an educational programme, taking into account the characteristics of the province. Thereafter, using funds generated from within Argentina, the aim was to maintain and extent the structures achieved in Chaco and encourage the development of complementary projects in other areas of the country using the knowledge, experience and educational programmes gained from the CAPABILITY project.

The province of Chaco was chosen for this project, in agreement with the Provincial and National Ministries of Health. Chaco is one of the 10 provinces in Argentina lacking genetic services, located in the north-west of the country. Chaco has 1.042.881 inhabitants of whom almost 50.000 belong to Toba, Wichi, and Mocoví communities.

The project was co-sponsored by SAMIC by building and staffing a cytogenetic laboratory in Chaco.

To undertake the project a local working group was established in each Sanitary Area of Chaco to liaise with and assist the CAPABILITY team. By means of a questionnaire the health professional resources, patient profile, available means of communication (print, computers, internet availability, and others) and the preference for the type of teaching material wanted in each Sanitary Area was assessed.

Educational material was then developed to meet the needs in the Sanitary Areas. Two syllabi were prepared, one for people working in primary health care (nurses, physical therapists, clinical psychologists, health care agents, and midwives) and the second for medical practitioners. Based on these syllabi, teaching material in the forms of a book, CD, and PowerPoint presentations was developed. A web page was

designed to make this material easily available and as a resource for continuing medical genetic education and on-line consultation for health professionals in Chaco with the Human Genetics Department in the Hospital de Pediatria Juan P. Garrahan, SAMIC, Buenos Aires.

The training took place at 3 levels

a) *Initial training:* Training in genetics of health care professionals working at health centers in different sanitary areas in Chaco
b) *Continuous education:* Communication to achieve periodic updates for the health care professionals to improve the quality of care.
c) *Amplifying effect:* Design of a module for training new teachers (according to the trainer's profile: a group will act as Education Management/and the others will be Trainer Effectors).

Contents according to the profile will be transferred and connections with the specialized referral center will be maintained to update them.

During the initial training health care workers and professionals in the Sanitary Area were taught and trained by means of a series of seminars, workshops and teaching consultations (hands-on experience to observe physician-patient relationships and use of skills).

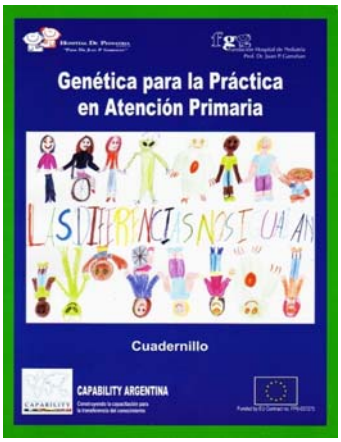
A total of 413 health care workers were trained. While this teaching and training was being undertaken, engagement of the community with the project and with children with congenital and genetic disorders was achieved through the media (newspapers, TV, radio, internet), open talks for the community and workshops for parents with children with congenital and genetic disorders.

After a pilot project in sanitary zone V, it was decided to incorporate in the remaining sanitary zones community activities in the definition, implementation, development, and continuation of the project in time. From then on, training was not only addressed to health care professionals, but also to health care managers who are leaders in their communities. They served not only as a source of consultation on the community needs, but also as a control mechanism, where adequate solutions related to prevention can be looked for. They facilitate the access of patients to the health care network with key locations for genetic diagnosis and counseling.

The success of the Chaco Outreach Project has become apparent in several ways:

- Referrals of patients with congenital and genetic disorders for medical genetic consultation increased significantly: 3.5 times in those Sanitary Areas comparing before and after the training period, and notably, all patients referred did have a congenital disorder. (The total number of consultations from Chaco was 268 after the training period in 2008-2009 and 77 before the training period in 2006-2007).
- The Ministry of Health in Chaco has decided to extend the project to all 7 Sanitary Areas, and is opening a cytogenetic laboratory because of the anticipated increase in demand.
- Based on the results of the Chaco Outreach Project, the National Ministry of Health has decided to implement and support medical genetic care programmes in 4 provinces in North-East Argentina, including Chaco.

- Because of the demand, the educational material in book and CD format has been distributed widely throughout the country by the Garrahan Hospital Foundation.



1.3.2 Egypt – Knowledge, Attitudes and Practice (KAP) of Medical Genetics in a Rural Egyptian Community

(report available on CAPABILITY website: http://www.capabilitynet.eu/Documents/presentation/Community%20Education%20in%20Health%20Aspects%20of%20Genetics_A%20Capaci-205.pdf)

This project addressed barriers identified by the Egyptian survey in integrating new medical genetic health services into the existing public health services such as:

- Difficulties in integrating new services into a public health service with many existing vertical programmes.
- Lack of knowledge of health professionals on where and how to access already existing services for the care and prevention of congenital and genetic disorders.
- Long distances from residence to medical genetic facilities, exacerbated by the unavailability of these facilities in some governorates (provinces).
- The financial cost of the services available and lack of insurance coverage or adequate reimbursement.
- Cultural and religious beliefs
- Concerns about privacy and confidentiality.

The project was conducted in the Om-Khenan area in the Hawandia district in the Giza governate.

The following tasks were worked upon: The knowledge, attitudes and practices with respect to congenital and genetic disorders of the villages' 21.525 inhabitants and the medical and para-medical professions responsible for their care were surveyed and analysed.



To achieve the task the first step was the acquisition and evaluation of demographic and other health related data of Om Khenan. Its inhabitants are mostly of low socio-economic status (58%) with low education, 68% having attended primary school education and 30% secondary school. Most males (85%) work as farmers or manually labourers and 68% of women are housewives. Primary health care ensures 98.8% vaccination coverage and 99.7% coverage for neonatal thyroid screening. There are just over 500 births annually with 19% occurring at home. This represents 23.6 births per 1000 population a figure exacerbated by the low use of contraception (32.4%). It can thus be estimated that up to 40 infants are born each year with a congenital or genetic disorder in Om Khenan.

Religion is a strong force in the community, 85% of people being Muslims and 15% Christians. This underpins their beliefs in God and fate, the sanctity of family and their rejection of abortion which is considered a sin. It also motivates their efforts to ensure the health, education and future wellbeing of their children.

With this background a questionnaire for the community knowledge, attitudes and practices with respect to congenital and genetic disorders was developed and piloted.

The questionnaire interrogated community needs with regards congenital and genetic disorders, including available services for care and prevention, and issues related to the need for, how to, and to which group to deliver public education on the topic. It

further explored women's knowledge to congenital and genetic disorders; their needs related to these disorders and how to improve the public health services; their needs for and attitudes to prevention of congenital and genetic disorders; and their practices during pregnancy to prevent these disorders. Specially trained community outreach visitors (COVs) administered the questionnaire to 531 randomly selected families in the village. The informants were mostly wives (80%).

The survey confirmed that public knowledge in the village about congenital and genetic disorders was poor, but 87% of informants considered they needed information on these disorders with 69% preferring to receive it just before marriage and 30% while at school. The latter is 10% more than the percentage of people that attended secondary school. There was a significant relationship between informant knowledge and age, marital status and level of education. Informants (86%) considered the public required access to medical genetic services with 47% considering that this should include mandatory pre-marital consultation and genetic counselling. Interesting only 8% considered consanguinity should not be allowed, and not surprisingly 69% of informants were against prevention of congenital and genetic disorders by abortion.

Using this and other knowledge gained, an educational programme for nurses and COVs to deliver simplified medical genetic health awareness to the community was developed. To assist them in this task a simple manual was also developed. Work on the project tasks included community education through home visits, group discussions seminars and public education in health care facilities.

The knowledge, attitudes and practice of physicians (general practitioners/ family practitioners, paediatricians and, obstetrician/gynaecologists) in Om Khenan and

surrounding health facilities were evaluated by a survey, a focus discussion group and a questionnaire using open ended questions. The results indicated the majority acknowledge the need for further training in medical genetics including literature, an illustrated book detailing diagnosis, treatment and prevention on common congenital and genetic disorders. Some admitted not having any information on available services and requested information on these and official referral cards to these facilities be made available to them. Regarding public education the majority considered expectant mothers to be the best target and recognised COVs as the most suitable route for community education. The role of the media was noted, with some also proposing the youth population as being a suitable target for education including adding relevant information into the school curriculum. Seminars for physicians working in the community were held to inform them of approaches to common congenital and genetic disorders; current medical genetic services in Egypt; and methods of prevention and early intervention for congenital and genetic disorders.

The Knowledge, Attitudes and Practice Survey on Medical Genetic in a Rural Egyptian Community documented important information for medical genetic services in Egypt and for middle- and low-income nations with similar circumstances. Undoubtedly Egypt has reached a stage of health transition and development that recommend the Egyptian Ministry of Health and Population efforts to develop medical genetic services. They are confronted by the barriers to their task including those documented above and others revealed by the CAPABILITY approach including:

- The lack of health professional knowledge on available services in the country was confirmed. However, this is amplified by the lack of health professional knowledge on congenital and genetic disorders revealed by the project.

- The Knowledge, Attitudes and Practice Survey confirmed that religious and cultural belief will impinge on the implementation of preventive services and interventions- medical genetic screening, prenatal diagnosis and termination of pregnancy, were these to be offered, and attempts to reduce consanguineous marriage.

- The low percentage use of contraceptives and concomitant high birth rate are increasing the birth prevalence of congenital and genetic disorders- chromosomal trisomies and autosomal recessive disorders, in the country. Improving access to and use of contraceptive services would be an effect public health intervention to reduce the burden of these disorders.

- Reducing the frequency of consanguinity is unlikely for a long time. Therefore pre-conception screening and counselling for common recessive disorders (thalassaemia), possibly before marriage which seems acceptable to a large proportion of the Om Khenan population, is a consideration. The early postnatal diagnosis of haemoglobin disorders and inborn errors of metabolism will become a consideration to add to the presently available neonatal screening for congenital hypothyroidism.

- Poor public knowledge and appreciation of congenital and genetic disorders is a barrier to the implementation of medical genetic services in Egypt, but one that is amenable to remediation. In rural situations like Om Khenan public education by COVs and nurses will be important due to the high percentage of people with low levels of education and poverty which would diminish the likelihood that they would benefit from education in school and through the media. However the situation might be different in urban areas were all forms of public education would probably be beneficial. Education should also target males, as over 60% of married women considered their husbands as their preferred adviser. For the same reason all genetic counselling, where possible, should include the husband.

1.3.3 South Africa - Greater Sekhukhune-CAPABILITY Outreach Project (GraSCOP)

(report available on CAPABILITY website: http://www.capabilitynet.eu/Documents/presentation/GraSCOP%20report.pdf)

This project was based on the CAPABILITY HNA survey for this region and on a successful clinical genetic outreach programme to hospitals in Limpopo Province (formerly Northern Province) undertaken in the 1990s. In 2006 the Limpopo Provincial Department of Health and Social Development initiated a task team to plan and implement medical genetic services in the province.

GraSCOP was initiated from that task team to pilot a primary and secondary health care medical genetic service in the Greater Sekhukhune district with the objectives of:

- Testing and developing the principles and practices of primary health care based medical genetic services as outlined in the South African National Department of Health's 'National Guidelines for the Management and Prevention of Birth Defects and Disabilities'.
- Further assessing and developing the Medical Genetic Education Programme (MGEP), a distance learning education programme currently used by the National Department of Health for post graduate nurse training.
- Re-evaluating the epidemiology of congenital disorders in this setting
- Testing the clinical utility of DNA based medical genetic tests and technologies
- Using the knowledge and experience acquired from the project to assist the implementation and development of medical genetic services throughout Limpopo and other provinces in South Africa.

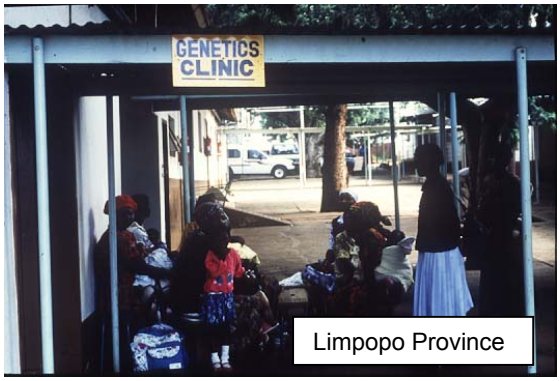
To initiate GraSCOP 38 nurses and 6 primary health care doctors from St Rita's Hospital and it referring primary care hospital underwent training with MGEP. A feature of this teaching was that the inclusion of primary care doctors in the courses was encouraged. Results obtained in the examination rendered were similar to those from previous courses held elsewhere in the country, for those that completed the course. What was different was that 8 candidates did not attend all the contact days and complete the course, and therefore could not write the examination.

In addition during the GraSCOP programme a trial was undertaken of the MGEP contact day teaching by tele-teaching to hospital based tele-conferencing facilities in Limpopo. This was the first attempt at undertaking the MGEP contact day teaching in this manner and was very successful with 86% of the candidates passing the examination. Further piloting of tele-teaching of the MGEP programme contact days will now be undertaken with hopefully similar results. Doing the contact day teaching for MGEP in this manner ensures the course can be taught to more nurses and doctors at significantly less cost, and travel and inconvenience to both students and lecturers. The second objective of the GraSCOP programme was thus achieved.

The training of the doctors and nursing staff with MGEP was to enable them initially to recognise infants and children with congenital disorders in their hospitals possibly clinically diagnose the more common congenital disorders and initiate relevant investigations and treatment. It was then intended that they refer their patients with their parents, and attend with them, to outreach clinics held at St Rita's Hospital by medical geneticists and medical genetic counsellors of the Division of Human Genetics, National Health Laboratory Services Division (NHLS) and WITS.

The purpose of nursing staff and doctors attending the outreach clinics with their patients was so that they could receive further 'on the job' teaching and training from the outreach clinic staff. Despite efforts to try and achieve this, it never occurred during the programme. Patients seen at the outreach clinics were only those diagnosed and being treated at St Rita's Hospital.

Reasons for this failure to network the primary care hospitals to St Rita's, the secondary care facility for the district, for the outreach clinics were sought. Two cogent and interconnected reasons were determined. When the protocol for GraSCOP was developed in 2007 the 2006 figures for vacancies in medical practitioner posts (26.8%) and nursing posts (15%) were available. The 2008 figures record a significant increase in vacancies, to 35.4% for medical practitioners and 43.7% for nursing staff. In 2008, 42.1% of all health professional posts in the public health sector in Limpopo were vacant. This, with the burden of HIV/AIDS and TB in the Province, are placing huge stress on the health services, including available health professionals, in the Province[4]. The care and prevention of congenital and genetic disorders must rate a lower priority in these circumstances, and hospitals quite obviously could not release doctors and nurses from their post to attend the outreach clinics. Additionally, undertaking epidemiological studies in these circumstances was not possible. This is also an explanation for why so many students did not complete the MGEP course.



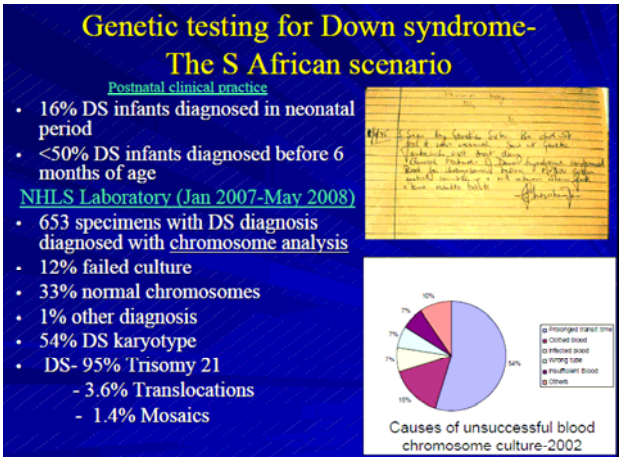
Limpopo Province

One positive development resulted from this situation. A cell phone was given to the paediatrician and the neonatal ward at St Rita's Hospital. With a cell phone photographs of infants and children with congenital disorders with dysmorphic feature were taken and MMSed to the Division of Human Genetics. With the photographs and other clinical details obtained by fax or through cell phone conversation, a clinical geneticist in the Division of Human Genetics offered a tentative diagnosis and suggested relevant investigations and treatment to the attending staff at St Rita's Hospital. Although in its early stages this appears to by an acceptable way to offer clinical support to clinicians in rural areas like Greater Sekhukhune.

[4] Health Systems Trust. South Africa Health Reviews 2006 & 2008. [www.hst.org]

Finally, during GraSCOP, the clinical utility of QF-PCR for the postnatal diagnosis of Down syndrome, using the *EuroGentest* criteria, was evaluated in the circumstances pertaining to South Africa.

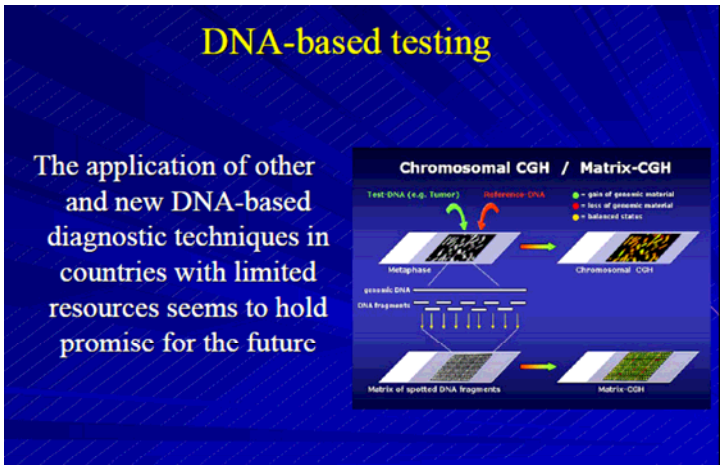
NATIONAL HEALTH LABORATORY SERVICE



A retrospective audit of chromosomal analyses done in the Division of Human Genetics cytogenetic laboratory from January 2007 to May 2008 documented that 653 specimens were received with a diagnosis of Down syndrome. Of these 12% were unsuccessful due to failed lymphocytes culture growth due to problems occurring before the specimens arrived at the laboratory, mainly prolonged transit time (54%). Normal chromosomes were found in 33% of analyses and 1% had a different diagnosis. A diagnosis of Down syndrome was confirmed in only 54%. Problems of making a clinical diagnosis of Down syndrome in African infants in South Africa has been previously recorded, with only 16% of infants diagnosed in the early neonatal period and less than 50% before 6 months of age.

Initial it was planned to use only specimens from Greater Sekhukhune for the clinical utility evaluation of QF-PCR for the diagnosis of Down syndrome. Then academic paediatricians at the University of Limpopo requested that the postnatal tests for the diagnosis of Down syndrome from the whole province be included in this research. Due to the Division of Human Genetics cytogenetic laboratory loosing 50% of its staff in mid 2008 (still not replaced), after consultation with senior academic paediatricians at the Universities of Pretoria and the Witwatersrand in Gauteng Province, the project was extended, as a necessity, to include all postnatal specimens with a clinical diagnosis of Down syndrome received in the laboratory. Lectures were given to paediatricians in the referral area of the Division of Human Genetics on QF-PCR and its use, including counselling information for patients with positive results. A fact

sheet on this was sent to doctors referring patients to the Division of Human Genetics, and accompanies test results for patients confirmed as having Down syndrome. Information from a medical geneticist or genetic counsellor is also available on a designated phone line in the Division of Human Genetics.



Between July 2008 and February 2009, 223 specimens with a clinical diagnosis of Down syndrome were analysed by QF-PCR. A diagnosis of Down syndrome was confirmed in 64% and not confirmed in 36%, results similar to the audit in the cytogenetic laboratory, considering the problem of failed lymphocyte culture was eliminated. QF-PCR cannot differentiate translocation Down syndrome and misses 30% of mosaic Down syndrome in those patients with low mosaicism in the blood. When evaluated against the criteria for clinical utility proposed by *EuroGentest*, it was considered that the use of QF-PCR for the postnatal diagnosis of Down syndrome in the circumstances currently pertaining in South Africa had standing. Although there are disadvantages to using QF-PCR for this purpose these were far outweighed by the advantages including cost-saving and because of staff shortages not being able to offer a confirmatory diagnostic test for Down syndrome.

1.3.4 Lessons learned from the national projects

Object lessons for the development of medical genetic services in middle- and low-income nations are available from the national demonstration projects. These include:

- The involvement, including the political will and commitment, of the health authorities of governments in the initiation and development of services for the care and prevention of people with congenital disorders is essential. This was previously highlighted by the WHO[5]. Evidence of the involvement and commitment of government and health authorities was available from the Chaco outreach project which was sustained and supported during the tenure of two different Ministers of Health and their administrations, in office during the project.

- A functional and stable health care system, with adequate staffing, must be in place for the successful initiation and development of medical genetic services. This is highlighted by contrasting the success of the Chaco Outreach Project with the problems encountered in the South African programme GraSCOP. South Africa is currently experiencing a health care crisis consequent on its HIV/AIDS epidemic and exacerbated by a lack of available health care personnel. The latter was incipient when GraSCOP was muted and deteriorated significantly, and unexpectedly, in Limpopo Province, particularly with respect to nursing staff. Consequently achieving some of the objectives of GraSCOP was not possible in contrast to Chaco where the health care service was functioning and well staffed, allowing the project to advance beyond its original expectations.

- Coupled with the need for a functional and stable health care system is the prerequisite of ensuring that previous unmet health needs will not compete for resources and thus limit the development of intended medical genetic services. In the case of South Africa the unmet health needs involved mainly HIV/AIDS and TB, but in other middle- and low-income nations it might involve other infectious diseases.

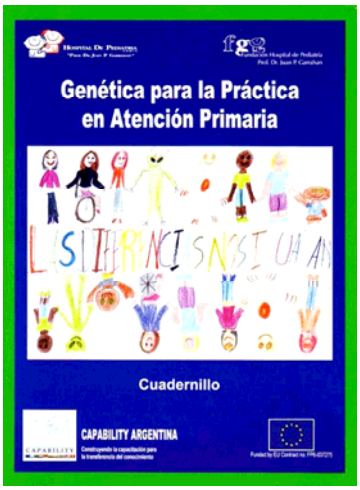
  These points are evidence of the need for middle- and low-income nations to utilise health needs assessment as proposed by CAPABILITY. Undertaken correctly health needs assessment would identify such issues and enable solutions to be sought.

[5] WHO. Services for the prevention and management of genetic disorders and birth defects in developing countries. WHO, Geneva, Switzerland. 1999. [WHO/HGN//GL/WAOPBD/99.1]

- The role of and need for medical genetic education for health care professions and workers was highlighted in all the demonstration projects. This was particularly notable in the Chaco outreach project in which the success of the services developed was reliant on the education of the provinces health care professions and workers. Recent evidence from the development of medical genetic services in Iran recorded that the financial cost of this education is one of the major expenses in initiating and developing medical genetic services[6] This is an important consideration for the future development of medical genetic services in middle- and low-income nations.

- The Egyptian community education project through its knowledge, attitudes and practices survey demonstrated the lack of medical genetic knowledge of health care practitioners and workers working in the primary health care settings common to all three projects. The development of medical genetic education material and its use for teaching and training health care practitioners and workers is a priority in middle- and low-income nations.

- Medical genetic educational material for health care professionals and workers is available word-wide from numerous sources, including the internet. However, it is considered necessary by CAPABILITY for each country to develop it own educational material to meet the specific needs and circumstances of each country, as demonstrated in the Argentinean and Egyptian projects.



[6] Samavat A. Genetic epidemiology in Iran- a basis for service delivery. PhD thesis. University College, London, UK. 2009.

This had previously been done in South Africa. Apart from addressing the particular circumstances of each country, this raises the profile of the authors in the country and through them adds to the awareness of health professionals and workers of congenital disorders, their care and prevention. The process of developing the educational material for national use can also be used to unify national efforts for the care and prevention of congenital disorders by involving people from throughout the country. This was previously done in South Africa.

- The CHACO outreach and the Egyptian community education projects illustrated the importance of community involvement in the development of medical genetic services in middle- and low-income nations. The Egyptian project identified the lack of knowledge of such communities regarding congenital disorders, their care and prevention whilst showing their desire to learn about and understand the issues. The Argentinean project illustrated how the communities involvement, including their education is necessary for the development of services for the care and prevention of congenital disorders.

  The community knowledge, attitudes and practices assessment of the Egyptian project confirmed that such surveys can be successfully undertaken in middle- and low-income nations and their value to the health needs assessment process.

- The Argentinean project in Chaco clearly illustrated that the success of such a project in developing previously unavailable services in one part of a country frequently stimulates the demand for similar services in other areas. Thus the initiation of such a project needs to be carefully planned to maximise the chances of success, raising the issue of health needs assessment.

- All of the projects confirmed that health needs assessment, as delineated by CAPABILITY, could provide a rational evidence-based method to assist countries identify their health needs for the care and prevention of congenital disorders to enable them to plan appropriate services, sensitive to specific national and local contexts.

1.3.5 Germany – Public Private Partnerships

(report available on CAPABILITY website: http://www.capabilitynet.eu/Documents/presentation/Public%20Private%20Partnership.pdf)

The project "Public Private Partnerships" addressed the problem that health care systems in developing countries, willing to set up the scope of clinical genetic services, in particular genetic testing and screening, all face the problem of very limited resources that can be made available for these novel technologies. In addition health workers' migration and brain drain from developing to developed countries are severely affecting health care in developing countries.

How to attract private funds in order to satisfy the public need for genetic services has so far not been effectively considered in these economies. The project explored such possibilities by collecting background information on Private-Public-Partnership models with the intention to encourage the establishment of genetic services in developing countries built on such principles.

1.3.6 Evaluation approaches for the application of genetic testing in health care services

CAPABILITY conducted an overview of national and international frameworks for the evaluation of the clinical validity and utility of genetic testing. Over the last decade questions have been raised specifically in Europe and the United States about appropriate evidentiary standards for the translation of new genetic tests/genetic

knowledge into health practice and prevention. In Europe the NoE *EuroGentest* addressed these questions (Unit 3, http://www.eurogentest.org/unit3/). In the United States the ACCE Project piloted an evidence evaluation framework of 44 questions and addressed the components of evaluation: Analytic and Clinical Validity, Clinical Utility and Ethical, Legal and Social Issues. In total the ACCE project examined available evidence for five genetic testing applications. In 2005 the "Evaluation of Genomic Applications in Practice and Prevention (EGAPP) Working Group" was established by the National Office of Public Health Genomics at the Centres for Disease Control and Prevention "to develop a systematic process for evidence-based assessment" for genetic test applications[7].

In 2007 the PHG Foundation published recommendations for an expanded framework for genetic test evaluation ("Moving beyond ACCE") which was endorsed by the United Kingdom Genetic Testing Network (UKGTN)[8].

In 2008 the NoE *EuroGentest* published its recommendations for genetic test evaluation[9].

The CAPABILITY report "A comparison of criteria for clinical validity and utility in various national and international frameworks" provides a comparison of the various national and international frameworks developed so far (available on CAPABILITY website). The comparison shows that genetic tests evaluation is difficult because a) many genetic disorders are rare and b) number and quality of studies providing evidence are limited. Most studies do not fit very well within the accepted standards for systematic evidence review for clinical utility such as randomised controlled trials or cohort or case-control studies[7]. However, most evaluation approaches have adopted the basic aspects of the ACCE analytic framework and use or recommend the questions developed by ACCE to organise the collection of information (i.e. UKGTN, *EuroGentest* approach) for conditions with relatively high prevalence.

[7] Teutsch, Steven M. et al.: The Evaluation of Genomic Applications in Practice and Prevention (EGAPP) initiative: methods of the EGAPP Working Group. Genet Med. 2009 Jan;11(1):3-14.
[8] Wylie Burke and Ron Zimmern: Moving Beyond ACCE: An Expanded Framework for Genetic Test Evaluation. A paper for the United Kingdom Genetic Testing Network, 2007,(PHG Foundation: www.phgfoundation.org)
[9] Javaher P, Kääriäinen H, Kristoffersson U, Nippert I, Sequeiros J, Zimmern R, Schmidtke J. EUROGENTEST: DNA-based testing for inheritable disorders in Europe. Community Genetics 2008;11:75.120

For rare conditions a pragmatic decision tree has been developed by *EuroGentest* Unit 3 and published[9]. This decision tree aims at providing a founding that allows decision making in the absence of available data. It follows a constructed line of "evidence" using a series of key questions.

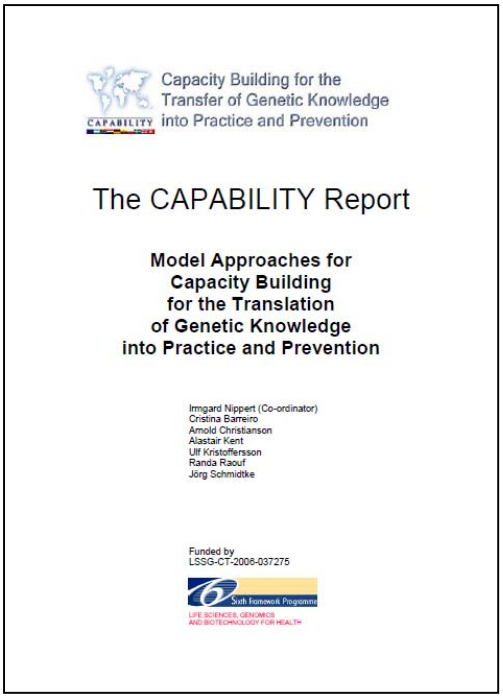
The CAPABILITY comparison also shows that evidence collection and assessment - due to the lack of available data – is a time consuming, costly procedure that requires the involvement of multidisciplinary experts representing areas such as: clinical genetics, laboratory expertise, epidemiology, evidence-based medicine, ELSI and economics.

The most important outcome of the comparison is that the way the evidence assessment and the evaluation process are structured and how the outcome is derived from evidence needs to be transparent and accountable. There are ongoing efforts to optimise existing evidence review methods and to maintain a consistent nomenclature such as the *EuroGentest* Clinical Utility Gene Cards (CUGC) for Europe and EGAPP project for the United States.

The comparison reveals that genetic test evaluation is a constant "work in progress" and that the challenge lies in achieving more rapid and less expensive reviews for genetic test applications despite limited available information and without sacrificing the quality of the recommendations.

The outcomes of the demonstration project and of the comparison on national/international approaches to evaluate the clinical validity and utility of genetic tests were submitted to international experts and observers for discussion during two international workshops and a mid-term evaluation meeting.

Based upon the outcome of the meetings CAPABILITY drafted the following recommendations for capacity building. The complete report "Model Approaches for Capacity Building for the Translation of Genetic Knowledge into Practice and Prevention" is available on the CAPABILITY website.


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

The CAPABILITY Report

Model Approaches for Capacity Building for the Translation of Genetic Knowledge into Practice and Prevention

Irmgard Nippert (Co-ordinator)
Cristina Barreiro
Arnold Christianson
Alastair Kent
Ulf Kristoffersson
Randa Raouf
Jörg Schmidtke

Funded by
LSSG-CT-2006-037275

Sixth Framework Programme

LIFE SCIENCES, GENOMICS AND BIOTECHNOLOGY FOR HEALTH

2. Recommendations: Moving from evidence to decisions for capacity building for medical genetic services in middle- and low-income countries

The CAPABILITY model approach for capacity building supports the development and implementation of a systematic evidence-based process for developing medical genetic services and the applications of genetic tests and/or other applications of genetic knowledge for clinical and public health practice.

Many developing countries have yet to confront the issues of developing policies for the provision of medical genetic services and medical genetic testing. Given their resource limitations they need practical approaches that have been proven to work in real settings. Approaches that will result in the development of appropriate needs-based services, that are sensitive to specific country contexts, health service patterns, available resources and capacities as well as to legislative factors and cultural and societal norms.

2.1 Health needs assessment as a prerequisite for evidence-based service development

The CAPABILITY model recommends a step-wise approach for capacity building. Its application could significantly assist countries to develop their medical genetic services:

- During the initial phase a systematic HNA – as defined and outlined in the CAPABILITY report "Model Approaches for Capacity Building for the Translation of Genetic Knowledge into Practice and Prevention" – should be undertaken by a multidisciplinary team preferably with a mandate from national and/or regional departments of health. The team should ensure it has the required resources to complete the HNA process. It can then delineate its strategic aims, according to its mandate, before undertaking a situational analysis. This comprises an assessment of the internal environment, an objective profile of existing health, including health care services. A baseline knowledge of these services enables an understanding of the current situation in order to consider what to change and how. This is supported by an assessment of the external environment, an appraisal of those issues that mould the internal environment.

- Comparing the internal and external environment and the strategic aims will enable an estimation of the ‘gap’ between them leading into the next phase. This is the collection of information on health needs, professional and public opinion regarding these and constraints that could affect proposed future services and interventions to meet these needs.

- Analysis of all this information should enable the prioritisation of health needs and services and interventions to meet them and planning for their implementation.

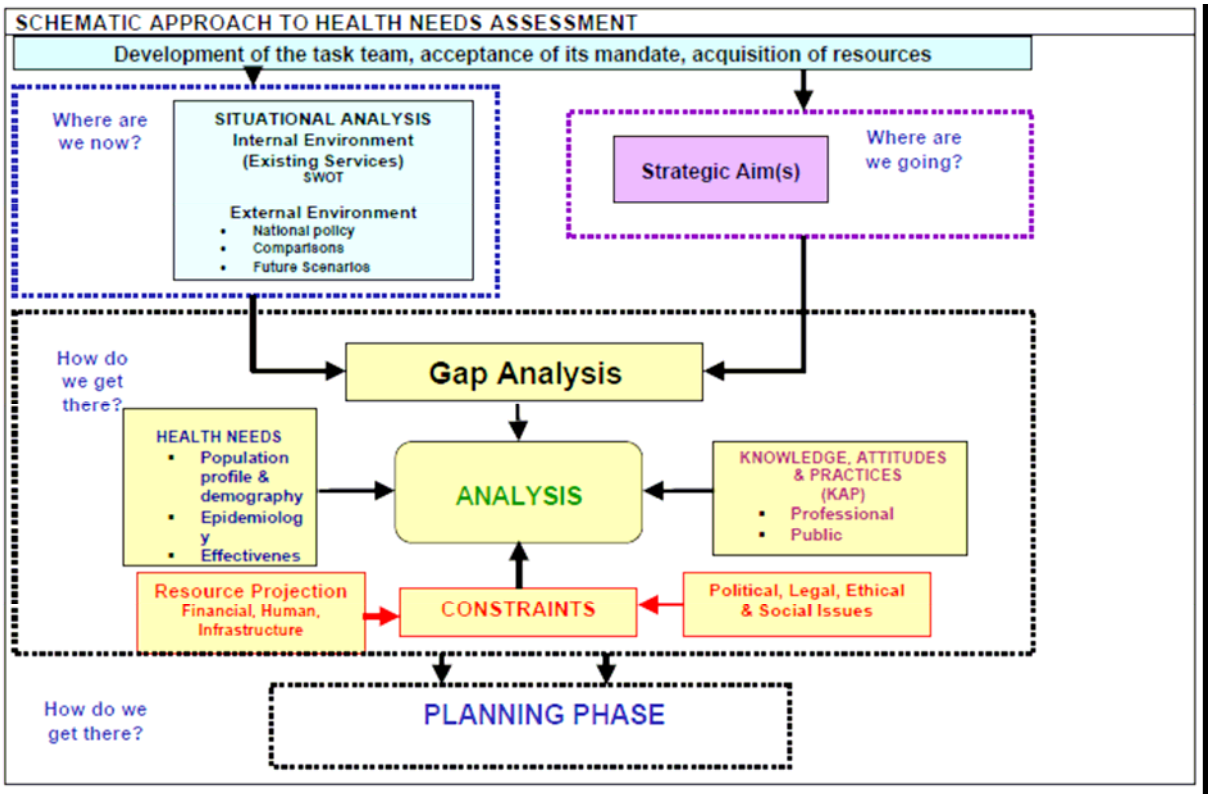
- In the absence of adequate epidemiological data, plans and actions should be based upon the best available evidence for initiating services.

- The outcomes of the HNA process, assumptions made in determining these outcomes and the relative weight attached to them should be made available in the public domain.

- Successful implementation of a health service plan for medical genetic services derived by HNA is not an endpoint. It will change the internal environment for these services whilst epidemiological transition, technological advances, changing public expectations and demands, resource limitations and the rising cost of health care remain, continuing to influence the ever changing external environment.

- If HNA is to have a purpose the successful achievement of its strategic aim(s) should be seen as the first step in an on-going and cyclical process of HNA for continuing health gain.

- Genetic services need to be accepted by governments as a legitimate part of the primary health care system and funded to the level necessary to ensure that they are able to meet population needs, in particular the needs of women and families hoping to gain control over their reproductive options in order to maximise their opportunity for a successful outcome to a wanted pregnancy.



Source: A. Christianson, R. Zimmern on behalf of the CAPABILITY Consortium: "HNA for Medical Genetic Services in Middle- and Low-Income Countries" (available on CAPABILITY website *http://www.capabilitynet.eu/Documents/Documents_09022010/HNA%20for%20Medical%20Genetic%20Services%20in%20Middle-%20and%20Low-Income-205.pdf*)

2.2 Genetic test evaluation

Developing countries have yet to confront the issues of developing policies for the provision of medical genetic testing.

Ideally, the following components should be considered when the clinical utility of a genetic test needs to be assessed:

- The natural history of the disease, if known, should be considered so that testing and intervention can be properly timed.
- Interventions that might follow a positive test result should be effective and available.
- Qualified pre-test, test, and post-test measures, including appropriate consent processes and genetic counselling, should be in place when needed.

- Health risks associated with testing and interventions following positive and negative test results as well as with not testing should be considered.
- Financial costs and benefits of testing as well as refraining from testing should be evaluated.
- Testing services should provide educational materials, access to genetic counselling, and maintain surveillance over their activities.

Given the resource limitations of developing countries they would be well be served by accepting and if necessary adapting approaches described in the CAPABILITY report "Model Approaches for Capacity Building for the Translation of Genetic Knowledge into Practice and Prevention". When considering the introduction of a new test into the publicly funded health care system a rational framework such as the *EuroGentest's* "Points to consider" documents and the *EuroGentest* CUGCs are recommended[10].

2.3 CAPABILITY: Achievements, Impact and Sustainability

CAPABILITY's overall objectives are to contribute to the efforts to establish and sustain a worldwide harmonisation process for developing genetic services and 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.

[10] Currently CUGCs can be found for 33 conditions on the *EuroGentest* website (http://www.eurogentest.org/web/info/public/unit3/geneCards.xhtml).
From 2010 onwards CUGCs will be published in the European Journal of Human Genetics. While the CUGCs can claim to represent the state of the art at the time of publication, the rapid medico-scientific progress in this field will require regular updates, a service that is intended to be provided by European funds (*EuroGentest2*). Furthermore, the collection of CUGCs is intended to be much expanded for disease conditions not yet covered.

2.3.1 Achievements

- **CAPABILITY has successfully established an international multidisciplinary working group that will continue to collaborate.**
  Over the three years funding period CAPABILITY has evolved into an expanding international network that includes representatives from Argentina, China (People's Republic of China and Hong Kong), Egypt, Germany, Philippines, South Africa, Sweden, United Kingdom and the USA. The network is linked to NoE *EuroGentest*.

The CAPABILITY network promotes the following long-term goals:

- enable health care systems to integrate genetic knowledge appropriately, based upon local needs and priorities;
- strengthen the kinds of basic capacities that will allow participating developing countries to more easily incorporate the benefits of genetic/genomics research as they unfold;
- help to reduce inequalities in genetics health care between developed and developing countries;
- serve as a model project for successful, sustainable collaboration between EU Member States and developing countries.

The CAPABILITY network will continue to collaborate and foster cooperation and partnerships beyond the EC funding period (2007-2009) in order to improve the translation of genetic knowledge into healthcare, education and health policy at an international level.

The CAPABILITY network will continue collaboration via the following projects:

- a joint survey on "Genetic Services in Emerging Economies" (GenTEE) in cooperation with the EC's Joint Research Centre (JRC), Institute for Consumer Health and Protection (IHCP), Ispra, Italy. The survey starts in 2010. The GenTEE project is part of the proposed coordination and support action "*EuroGentest2*" (2010-2013);
  The GenTEE project will include new partners from India and the Middle East.

- an international project for developing a HNA "toolkit" for middle- and low-income countries. This is a five years multi-partner collaboration to improve services via a systematic HNA approach and reduce morbidity, mortality and other impacts associated with congenital/genetic disorders in low and middle income countries which bear the burden of the disorders; the project has been submitted for funding in 2009 to the UBS Optimum Foundation and has gone through the first stage of selection. The project has been designed and will be coordinated by the PHG Foundation, Cambridge, U.K. (R. Zimmern, chairman of the PHG Foundation, is a member of the CAPABILITY Working Group).

- **CAPABILITY has developed a model approach for capacity building that is sensitive to specific country contexts** including in particular addressing the assessed magnitude of health needs, health service patterns, available resources and capacities, gaps in service provision, professional and expert knowledge, cultural and social attitudes.

Evidence suggests that developing countries have not used formal HNA to inform the planning and development of genetic services before CAPABILITY. This is possibly due to limited cogent literature on HNA and available examples of its application to medical genetic prior to CAPABILITY. The CAPABILITY approach to HNA is a significant step towards alleviating this situation and its application could significantly assist countries to develop their medical genetic services.

2.3.2 Impact

- The CAPABILITY capacity model approach has been implemented into the health service plan for medical genetic services by the Argentinean Ministry of Health (MoH). The MoH - based upon the results of the Chaco outreach project - has decided to implement and support medical genetic care programmes, including new laboratory services, in four provinces in North East Argentina.

The education material developed by the demonstration project has been distributed nationwide in book and CD format throughout the country by the Garahan Hospital Foundation.

- The CAPABILITY project in Egypt has provided for the Egyptian Ministry of Health and Population (MOHP) evidence-based data and a planning tool how to overcome barriers to the implementation of medical genetic services.

- The knowledge and experience gained from the South African demonstration project has serious implications for the development of genetic services in South Africa, showing how severely affected are primary and secondary care services by staff shortages (migration/brain drain) and the HIV/AIDS and TB epidemics. Developing appropriate medical genetic services is difficult in these circumstances. Based upon the outcome CAPABILITY has recommended during a meeting with the National Department of Health that HNA would be an objective way to clarify matters and plan future genetic services. Based upon this recommendations the Department of Health is seeking funds to undertake an HNA in more provinces.

- Based upon the evaluation of the clinical utility of QF-PCR for diagnostic services, the NHLS Division of Human Genetics at WITS has decided to implement the tool.

2.3.3 Sustainability

The CAPABILITY network will be sustained via projects initiated during the reporting period (see above) and the CAPABILITY website will be sustained for the next three years.

3. Dissemination and Use

The CAPABILITY model approach - which is currently distributed - has gained momentum and is being adopted by participating developing countries (in Argentina for medical genetic service development, in South Africa to assess the magnitude of

needs and to identify service gaps via HNA in defined regions). This comes at a time when the WHO's Executive Board has recommended in 2009 the priorisation of genetic services and genetic in developing countries for the management and prevention of genetic/congenital disorders to assist them to attain their MDG4[11].

One of the great strength that the CAPABILITY Consortium brings to the dissemination strategy is its unique network of functions and contacts the participants have on national, European and global levels.
From its very start CAPABILITY has aimed at transparency and open access to information. Therefore, CAPABILITY website has been accessible to the public from the start of the project in 2007 and will be maintained until 2013.

The overall objective of CAPABILITY's dissemination plan is to reach out to defined international target audiences in order to:

- promote the concept of a worldwide harmonisation process to ensure a sufficient level of quality, safety and efficacy of genetic service provision and testing,
- provide key stakeholders with a validated model approach for developing capacity building strategies including a systematic HNA and recommendations for evaluation of genetic tests that are considered for application into general clinical practice and for developing capacity building strategies,
- to share information on agreed quality standards for genetic test evaluation.

CAPABILITY project dissemination plan puts special emphasis on disseminating CAPABILITY activities and outcomes to middle- and low-income countries in Latin America, Central and Eastern Europe, Middle East, Asia and Africa.

The CAPABILITY model approach is disseminated to the following target audiences:

- The scientific community
- Health policy makers
- Global non-government organisations
- Health care providers, especially in primary care
- Parent/patient organisations and the public
- The private sector

[11] WHO Executive Board, May 14th, 2009

Dissemination overview table

Planned/ actual Dates	Type	Type of audience	Countries addressed	Size of audience	Partner responsible /involved
	Conferences:	**oral presentations**			
17-21 June 2007	"3rd International Conference on Birth Defects and Disabilities in the Developing World", Rio de Janeiro, Brazil	Research, Parent/ Patient Organi-sations	Brazil and other developing Latin American countries	appr. 1.000	participant 1 (I. Nippert, WWU) on behalf of CAPABILITY Consortium
25-26 September 2007	Birth Defects Conference, Lhasa, Tibet/China	Medical Practitioners and Health Care Providers	China	appr. 300	participant 1 (I. Nippert, WWU) on behalf of CAPABILITY Consortium
22-23 November 2007	Conference: Annual EuroGentest General Assembly	Research	EU Member States	appr. 200	participant 1 (I. Nippert, WWU) on behalf of CAPABILITY Consortium
17.-19 February 2008	First International Symposium on Genetics, Health and Disease. Amritsar, India	Research	India	appr. 1.000	participant 1 (I. Nippert, WWU) on behalf of CAPABILITY Consortium
August 2008	Multidisciplinary Medical Symposium, Buenos Aires, Argentina	Health Policy Makers, Health Care Providers, MDs	Argentina	511	participant 7 (C. Barreiro)
October 2008	CAPABILITY Argentina	Health Policy Makers, Health Care Providers, MDs	Argentina	missing information	participant 7 (C. Barreiro, SAMIC)
29-31 October 2008	IPTS Workshop "Genetic Testing in Emerging Economies: Development of an International Survey", Seville, Spain	Research		20	participants 1 (I. Nippert, WWU), 2 (U. Kristoffersson, LU), 5 (A. Christianson, WITS), 6 (R. Raouf, MOHP), Advisors: C. Padilla, N. Zhong, R. Zimmern
20-21 November 2008	Conference: Annual EuroGentest General Assembly	Research	EU Member States	appr. 200	participant 1 (I. Nippert, WWU) on behalf of CAPABILITY Consortium

Planned/ actual Dates	Type	Type of audience	Countries addressed	Size of audience	Partner responsible /involved
March 2009	Symposium fellows and residents, Buenos Aires, Argentina	Health Policy Makers, Health Care Providers, MDs	Argentina	appr. 250	participant 7 (C. Barreiro, SAMIC)
5-7 April 2009	Southern African Society of Human Genetics Conference, Stellenbosch, South Africa	Research		appr. 800	participants 1 (I. Nippert, WWU), 2 (U. Kristoffersson), 5 (A. Christianson, WITS)
23-26 May 2009	European Society of Human Genetics Annual Meeting, Vienna, Austria	Research	EU Member States	appr. 1.500	participants 1 (I. Nippert, WWU), 5 (A. Christianson, WITS), 7 (C. Barreiro, SAMIC)
September 2009	38 National Argentine Congress of Genetics, Tucumán, Argentina	Research	Argentina	appr. 300	participant 7 (C. Barreiro, SAMIC)
14 September 2009	Division of Noncommunicable Disease Prevention, WHO, Geneva, Switzerland	Research		internal meeting	participants 1 (I. Nippert, WWU), 5 (A. Christianson, WITS)
4-7 October 2009	4th Birth defects conference, New Delhi, India	Research	India and developing countries from Asia	appr. 2.000	participants 1 (I. Nippert, WWU), 4 (A. Kent, GIG), 5 (A. Christianson, WITS), 6 (R. Raouf, MOHP)
November 2009	V National Meeting of Hospitals, Buenos Aires, Argentina	Health Policy Makers, Health Care Providers, MDs	Argentina	appr. 500	participant 7 (C. Barreiro, SAMIC)
9-10 November 2009	IHCP "Workshop on Genetic Testing, Ispra, Italy	Research		10	participants 1 (I. Nippert, WWU), 2 (U. Kristoffersson), 3 (J. Schmidtke, MHH)

Planned/ actual Dates	Type	Type of audience	Countries addressed	Size of audience	Partner responsible /involved
	Posters:				
31 May -03 June 2008	European Society of Human Genetics Annual Meeting, Barcelona, Spain	Research	EU Member States	appr. 1.500	participant 1 (I. Nippert, WWU) on behalf of CAPABILITY Consortium
11-15 November 2008	American Society of Human Genetics Annual Meeting, Philadelphia, USA	Research	USA and international	appr. 5.000	participant 1 (I. Nippert, WWU) on behalf of CAPABILITY Consortium
1-3 April 2009	German Society of Human Genetics Annual Meeting, Aachen, Germany	Research	Germany, Austria, Switzerland	appr. 1.000	participant 1 (I. Nippert, WWU) on behalf of CAPABILITY Consortium
5-7 April 2009	Southern African Society of Human Genetics Conference, Stellenbosch, South Africa	Research		800	participant 5 (A. Christianson, WITS) on behalf of CAPABILITY Consortium
23-26 May 2009	European Society of Human Genetics Annual Meeting, Vienna, Austria	Research	EU Member States	appr. 1.500	participants 1 (I. Nippert, WWU), 7 (C. Barreiro, SAMIC)
4-7 October 2009	"4th International Conference on Birth Defects and Disabilities in the Developing World", New Delhi, India	Research, Parent/ Patient Organi-sations	India and developing countries from Asia	appr. 2.000	participant 5 (A. Christianson, WITS) on behalf of CAPABILITY Consortium
20-24 October 2009	American Society of Human Genetics Annual Meeting, Hawaii, USA	Research	USA and international	appr. 5.000	participant 1 (I. Nippert, WWU) on behalf of CAPABILITY Consortium
November 2009	V National Meeting of Hospitals, Buenos Aires, Argentina	Health Policy Makers, Health Care Providers, MDs	Argentina	appr. 500	participant 7 (C. Barreiro, SAMIC)

Planned/ actual Dates	Type	Type of audience	Countries addressed	Size of audience	Partner responsible /involved
	Project web-site:				
June 2007	http://www.capabilitynet.eu/	General Public	international	> 4.000 visitors per month	participant 1 (I. Nippert, WWU)/CAPABILITY Co-ordination centre
March 2008	www.capabilityargentina.com.ar	Health care professionals, restricted access through a password	Argentina	missing information	participant 7 (C. Barreiro, SAMIC)
	Press release(press/ radio/TV):				
6 March 2008	Newspaper "Diario Norte"	General public	Argentina	missing information	participant 7 (C. Barreiro, SAMIC)
16-18 April 2008	**Newspapers:** Newspaper “Diario Norte”. Local Magazine “Página 1” **Radio:** FM Libertad FM Milenium Radio Cordial **TV:** News: Breñas Cable Color News program	General public	Argentina	missing information	participant 7 (C. Barreiro, SAMIC)
	Newspapers: Newspaper “Diario Norte”, "El Reporte Portal de Noticias 24" and "Revista de La Federación médica del Chaco **Radio:** FM Joven **TV:** "Multimedios Chaco Roque Saenz Pena", "Video vision Castelli Canal 4", "Multicanal Resistencia canal	General public	Argentina	about 2.300.000	participant 7 (C. Barreiro, SAMIC)

Planned/ actual Dates	Type	Type of audience	Countries addressed	Size of audience	Partner responsible /involved
	80"				
	2 radio interviews	General Public	Egypt	missing information	participant 6 (R. Raouf, MOHP)
	Educational Materials				
	Talks for parents and the general community and brochures for the community	General Public	Argentina	appr. 130	participant 7 (C. Barreiro, SAMIC)
	Distribution teaching material CAPABILITY Argentina (Syllabus and CDs) in areas other than Chaco Workshops	Scientific Community	Argentina	960	participant 7 (C. Barreiro, SAMIC)
	Education Material for parents and primary care providers	Parents, primary care providers	Egypt		participant 6 (R. Raouf, MOHP)
1 October-31 December 2009	4 Seminars	Physicians, nurses and COVs	Egypt	30 to 50/seminar	participant 6 (R. Raouf, MOHP)
December 2009	4 seminars for physicians in the Giza governorate	Physicians	Egypt	15 to 25/seminar	participant 6 (R. Raouf, MOHP)
	Publication				
2008	Bidondo, Maria Paz; Barreiro, Cristina Zulema: Genética para la Práctica en Atención	Health care professionals (primary care)	Argentina	missing information	participant 7 (C. Barreiro, SAMIC)
	direct emailing	**Start date December 2009 will continue 2010**			
	European Society of Human Genetics Professional and Public Policy Committee pppc@eshg.org	Membership European Society of Human Genetics	European Member States, Eastern and Central European States	appr. 1.500	participants 2 (U. Kristoffersson, LU), 3 (J. Schmidtke, MHH) on behalf of the CAPABILITY Consortium

Planned/ actual Dates	Type	Type of audience	Countries addressed	Size of audience	Partner responsible /involved
	Rede Latino Americana de Genética Humana relagh@ufrgs.br	Membership Rede Latino Americana de Genética Humana	Latin American	appr. 1.000	participant 7 (C. Barreiro, SAMIC) on behalf of the CAPABILITY Consortium
	The Southern African Society for Human Genetics (SASHG)rhillerm @sun.ac.za	South African health care professionals	South Africa	missing information	participant 5 (A. Christianson, WITS) on behalf of the CAPABILITY Consortium
	Egyptian Society of Human Genetics	Health care professionals	Egypt	missing information	participant 6 (R. Raouf, MOHP) on behalf of the CAPABILITY Consortium
	European Genetic Alliances' Network www.egaweb.org	European patient organisations	EU	missing information	participant 4 (A. Kent, GIG) on behalf of the CAPABILITY Consortium
	European Platform for Patients' Organisations, Science and Industry (EPPOSI) www.epposi.org	Science, industry and patient organisations	EU	1.000	participant 4 (A. Kent, GIG) on behalf of the CAPABILITY Consortium
	Genetic Interest Group www.gig.org.uk	Patient organisations	UK	1.000	participant 4 (A. Kent, GIG) on behalf of the CAPABILITY Consortium
	SAIDA - The South African Inherited Disorders Association http://www.saida.org.za/	Patient organisations	South Africa	missing information	participant 5 (A. Christianson, WITS) on behalf of the CAPABILITY Consortium
	International Genetic Alliance (IGA) http://www.worldmuscleforum.org/demo/iga/index2.htm	International patient organisations	international	appr. 10.000	participant 4 (A. Kent, GIG) on behalf of the CAPABILITY Consortium

Planned/ actual Dates	Type	Type of audience	Countries addressed	Size of audience	Partner responsible /involved
	Ministry of Health (Ministerio de Salud y Ambiente) (Argentina)	Health policy makers	Argentina	missing information	participant 7 (C. Barreiro, SAMIC) on behalf of the CAPABILITY Consortium
	Women's Health and Genetics Unit, Directorate of Women, Maternal & Child Health, Department of National Health (South Africa)	Health policy makers	South Africa	missing information	participant 5 (A. Christianson, WITS) on behalf of the CAPABILITY Consortium
	World Health Organization (WHO)	Health policy makers	international	missing information	participants 1 (I. Nippert, WWU), 5 (A. Christianson, WITS) on behalf of the CAPABILITY Consortium
	Pan American Health Organization (PAHO)	Health policy makers	Latin America	missing information	Advisor V. Penchaszadeh on behalf of the CAPABILITY Consortium
	Organisation for Economic Co-operation and Development (OECD)	Health policy makers	international	missing information	participant 2 (U. Kristoffersson, LU) on behalf of the CAPABILITY Consortium
	Association for Medical Education in Europe (AMEE)	Health care providers	EU	missing information	CAPABILITY Consortium
	European Medicines Agency (EMEA)	Research	EU	missing information	participant 4 (A. Kent, GIG) on behalf of the CAPABILITY Consortium
	European Diagnostics Manufactures Association (EDMA) www.edma-ivd.be	Industry	EU	missing information	participant 4 (A. Kent, GIG) on behalf of the CAPABILITY Consortium

From: Heather Law
To: 'Prof. Dr. I. Nippert'
Sent: Tuesday, December 15, 2009 12:38 PM
Subject: Capability dissemination plan

Dear Irma

turning to the dissemination of the CAPABILITY report in the UK, we will place copies of the report on the GIG and Rare Disease UK website, with links to the Capability website for further information.

Copies will be sent to Regional Genetics Centres, the British Society for Human Genetics, the Joint Committee on Medical Genetics of the Royal Colleges and the National Institute of Medical Research. The MRC, the Wellcome Trust and other bodies will also be informed.

GIG's member organisations and the Association of Medical Research Charities will be notified and offered copies/the executive summary.

Articles will be placed in the GIG and RDUK newsletters and other similar publications.

Copies will be sent to the relevant minister in the Department of Health and DFID, and to the chairs of appropriate select committees in the Commons and the Lords.

A press release will be issued and key opinion leaders known to GIG will be contacted (MPs, journalists, Policy Think Tanks etc.)

Regards.

Alastair Kent
Director
Administrator
Genetic Interest Group (GIG)
Unit 4D, Leroy House
436 Essex Road
London
N1 3QP
heather@gig.org.uk
www.gig.org.uk
T: +44 (0) 20 7704 3141
F: +44 (0) 20 7359 1447

Annex:
CAPABILITY Working Group

Participants:

Participant 1
Irmgard Nippert

Women's Health Research
Institute of Human Genetics
Westfälische Wilhelms-Universität (WWU)
Münster, Germany

Participant 2
Ulf Kristoffersson

Head of the Department of Clinical Genetics
University Hospital in Lund University (LU)
Lund, Sweden

Participant 3
Jörg Schmidtke

Director of the Institute of Human Genetics
Medizinische Hochschule Hannover (MHH)
Hannover, Germany

Participant 4
Alastair Kent

Director of the Genetic Interest Group (GIG)
London, United Kingdom

Participant 5
Arnold Christianson

Medical Genetic Services
School of Pathology, Faculty of Health Sciences
University of Witwatersrand (WITS)
Johannesburg, South Africa

Participant 6
Randa Kamal Abdel-Raouf

Children with Special Needs Department
Ministry of Health and Population (MOHP)
Cairo, Egypt

Participant 7
Cristina Barreiro

Medical Genetic Service
Hospital de Pediatría S.A.M.I.C.
"Prof. Dr. Juan P. Garrahan" (SAMIC)
Buenos Aires, Argentina

Advisors:

Hillary Harris		Senior partner in a South Manchester general practice Manchester, United Kingdom
Stephen T. S. Lam		Director of the Clinical Genetic Service Centre Department of Health Hong Kong Special Administrative Region
Victor B. Penchaszadeh		Program on Genetics in Public Health and Human Rights, Pan American Health Organization (PAHO) Buenos Aires, Argentina
Ron Zimmern		Chairman PHG Foundation Cambridge, United Kingdom

Observers:

Nanbert Zhong

Director and Chairman, Peking University Center of Medical Genetics, Bejing, China; Director, New York State Office of Mental Retardation and Developmental Disabilities, Institute for Basic Research in Developmental Disabilities, Staten Island New York, USA

Carmencita Padilla

Director of the Institute of Human Genetics, National Institutes of Health, University of Philippines, Manila, Philippines

Ysbrand Poortmann

Vice President, World Alliance of Organizations for the Prevention and Treatment of genetic and congenital conditions, The Hague, Netherlands

Evaluator:

Christopher Howson

Vice president
March of Dimes Global Programs
White Plains, USA