

2. Recommendations: Moving from evidence to decisions for capacity building for medical genetic services

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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 chapter 3 – 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.

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 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 recommended¹⁰.

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 CAPABILITY: Networking achievements and impacts

- 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.

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 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 National Health Laboratory Services Division (NHLS) of Human Genetics at the University of the Witwatersrand (WITS) has decided to implement the tool.

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