“CAPABILITY” and “Genetic testing in emerging economies” (GenTEE)

I Nippert

Received: 24 June 2013 / Accepted: 26 June 2013
© Springer-Verlag Berlin Heidelberg 2013

CAPABILITY was a 3-year model project (2007–2009) that linked participants (A Kent, UK; U Kristofferson, Sweden; I Nippert and J Schmidtke, Germany) of the EuroGentest unit “Clinical Genetics, Community Genetics and Public Health” and unit “Education” with leading experts from Argentina (C Barreiro, Garrahan Hospital, Buenos Aires), Egypt (R Kamal Raouf, Ministry of Health&Population, Cairo) and South Africa (A Christianson, National Health Laboratory Service and University of the Witwatersrand, Johannesburg). The experts were chosen because they were engaged in national development projects to integrate genetic services into primary care services in their respective countries. Together, the EuroGentest participants and the experts formed the CAPABILITY consortium. The consortium shared a commonality of interests to:

- Promote an internationally shared set of basic quality standards for genetic services in middle- and low-income countries
- Assess genetic service needs in middle- and low-income countries
- Identify priorities for medical genetic service development and priorities for capacity building via a systematic health needs assessment (HNA)
- Develop a validated model capacity building approach for genetic services via demonstration projects.

The model approach for capacity building developed by CAPABILITY is sensitive to specific country contexts including in particular the assessed magnitude of needs, health service patterns, available resources and capacities, gaps in

service provision, professional and expert knowledge and cultural and social attitudes.

The CAPABILITY consortium successfully established the multidisciplinary international network GenTEE (2010–2013). This network closely links leading academic/health/government institutions from eight emerging economies: Argentina, Brazil, China, Egypt, India, Oman, the Philippines and South Africa, with the EuroGentest2 workpackage 8 “Best practice guidelines for the provision of genetic services,” the European Commission Joint Research Centre Institute for Health and Consumer Protection (IHCP), European academic institutions from Germany and Sweden and international parent and patient organisations. The main objectives of the GenTEE network are to document and compare current practices and the state of genetic service provision in the participating eight emerging economies via a standardised global survey (GenTEE survey) that allows comparison of services internationally across a number of key dimensions by using a core set of indicators selected by the GenTEE participants for their relevance and comparability. In addition, the GenTEE survey identifies current knowledge gaps and unmet service needs.

The outline of the special issue

Presented are four papers from the CAPABILITY project; two of them addressing conceptual approaches developed by the CAPABILITY consortium and two papers describing the outcomes of capacity building demonstration projects in Argentina and South Africa. Six papers are provided by GenTEE participants describing in a condensed way outcomes of the GenTEE survey in Argentina, Brazil, China, Oman, the Philippines and South Africa. The IHCP will publish a comprehensive report on the outcome of the GenTEE survey later this year. This report will include the outcomes for the surveys conducted in Egypt and India which could not be included in this special issue.

I. Nippert (✉)
Women’s Health Research Unit, Department of Human Genetics,
Medical School, Westfaelische Wilhelms-University,
Muenster, Germany
e-mail: nippert@uni-muenster.de

In their paper “Health needs assessment for medical genetic services for congenital disorders in middle- and low-income nations,” Arnold Christianson et al. describe the CAPABILITY HNA approach. CAPABILITY HNA is an epidemiological-assisted systematic approach for providing health policy makers with data for informed decision making in order to plan, introduce and beneficially change health care services (genetic services) to improve both individual and population health.

Florian Meier et al. explore ways how middle- and low-income countries could acquire the necessary capital to strengthen health care services/genetic services via public–private partnerships (PPPs). So far, PPPs have been used exclusively in other health areas. In their paper “Public–private partnerships as a solution for integrating genetic services into health care of countries with low and middle incomes,” a first attempt is made to discuss the feasibility of transferring the concept of PPPs to genetic services and explore how the PPP model could be applied for infrastructure building services.

The success of the CAPABILITY Argentina demonstration project is described by Cristina Barreiro et al. in “CHACO outreach project: The development of a primary health care based medical genetic service in an Argentinean province.” Based upon a systematic HNA for Argentina, the outreach project was conducted in one (Chaco) of the 10 Argentine provinces that lacked genetic services. Chaco is a remote under-resourced province that lies in the Gran Chaco plains, also known as the “Green Hell” for its extreme weather conditions and restricted accessibility. The Chaco demonstration outreach project successfully trained non-genetic health professionals to include genetics in their daily practice at primary and secondary health care facilities. The training not only enabled professionals to provide appropriate interventions to a population that lacked infrastructure and economic resources but also demonstrated and encouraged government action to generalise the programme to a number of other provinces.

Based upon the CAPABILITY Chaco capacity building model, the Garrahan Hospital, Buenos Aires, the CAPABILITY partner institution, decided to provide financial support from within Argentina to build a cytogenetic laboratory in the capital of the Province of Chaco. Training of the technicians for this laboratory was also sponsored. The training project in Chaco exponentially increased the number of consultations and samples being analysed in the laboratory, overwhelming the laboratory capacity and creating the need for the installation of additional laboratories in Chaco and other provinces of the country. This resulted in the creation of a network of new cytogenetic laboratories sponsored by the Garrahan Foundation. The Garrahan Foundation continues sponsoring the training model for genetics in the provinces as well as the training of personnel for the new laboratories within the network of cytogenetic laboratories.

Based on the CAPABILITY Argentina project in Chaco, the National Ministry of Health decided to implement a genetic programme in other provinces in northeast Argentina. The education material developed by CAPABILITY Argentina is now used nationwide and is continually updated.

Greater Sekhukhune is one of six health districts in Limpopo province, South Africa. It is a rural area with one regional hospital as the major centre of care for a population of one million; although there are molecular and cytogenetic laboratories in South Africa, distance and a lack of resources means that they are not easily accessible to those away from the main centres of population.

The knowledge and experience gained from the South African “Greater Sekhukhune CAPABILITY outreach project” described by Arnold Christianson et al. has serious implications for the development of genetic services in Limpopo Province, and by extension in South Africa, showing how severely affected are the primary and secondary care services by staff shortages (migration/brain drain) and the HIV/AIDS and TB epidemics. Developing appropriate genetic services in these circumstances is difficult. The paper recommends HNA as an objective way to clarify matters as they stand in South Africa today and to plan future medical services for the care and prevention of congenital and genetic disorders.

Undertaking these outreach demonstration projects in different health care systems has demonstrated that genetics services need not be a minority service for the wealthy (or for those living in developed health care economies). Simple, affordable interventions can extend the reach of genetic services into communities that are economically poor, where the infrastructure is underdeveloped and where the levels of genetic literacy might lead one to the assumption that genetic concepts and ideas would be difficult to grasp. Given the opportunity, genetic services are utilised and community support for services is widespread. CAPABILITY has shown the development of services, and support for those at risk must be incorporated into the priorities of the national health care systems if it is to be sustainable and able to reach its intended target groups on an on-going basis. The Chaco project achieved this, and the Argentinean government in partnership with the country’s leading paediatric hospital was able to reach its extent to other underserved provinces. In South Africa, other pressures prevented on-going support.

Due to the epidemiological transition in the emerging economies of China, East Asia, India, Latin America and South Africa, these economies are facing an increasing proportion of infant morbidity and mortality due to congenital and genetic disorders and an increasing exposure of their adult population to risks for non-communicable chronic diseases such as: heart disease, stroke, cancer and diabetes—diseases that all have subgroups with significant genetic components. The changes of risk factors involved in the

epidemiological transition result in a rising need for genetic services to improve both individual patient outcomes and overall population health. The challenges emerging economy countries are facing are manifold:

- To develop a service delivery infrastructure, including health workforce training, quality guidelines and procedures leading to equitable and affordable access to high-quality genetic services;
- To enable their health care systems to reap the potential benefits that the rapid development of genetic/genomic technologies and knowledge brings and ensure the successful translation of genetics/genomics laboratory and academic research into quality assured pathways.

The GenTEE international network initiative responded to these challenges by facilitating inter- and intra-country comparison on the current state of genetic service testing development with the help of a systematic survey conducted in eight countries selected for their capability and readiness to conduct such a survey. The GenTEE survey is the first survey worldwide that systematically assesses the current state of medical genetic services in emerging economies. The survey is based upon a common method/framework for data ascertainment, allowing examination and comparison of service development in the context of a broader view of the existing health care systems, given service resources and service delivery, governance, national health and research policies and genetic testing development and civil society engagement.

Presented here are summaries of the country reports from Argentina by Victor B. Penchazsadeh (*Genetic testing and services in Argentina*), Brazil by Dafne Dain Gandelman Horovitz et al. (*Genetic services and testing in Brazil*), China by Xinliang Zhao et al. (*Genetic services and testing in China*), Oman by Anna Rajab (*Genetic services and testing in Oman*), the Philippines by Carmencita David Padilla and Eva Maria Cutiongco de la Paz (*Genetic services and testing in the Philippines*) and South Africa by Jennifer G.R. Kromberg et al. (*Genetic services and testing in South Africa*).

Although these countries represent different population and country sizes, different health care systems and funding schemes, different health care capacities, different socioeconomic structures and different cultural backgrounds, they share, as the reports show, significant commonalities: congenital and genetic disorders have become a major disease “burden” and there is a need to adjust new demands for essential genetic testing services and for capacity building functions that strategically respond to the needs of those affected by or at risk for genetic disorders.

Development of services was/is often funded by research means depending on the priorities chosen by individual academics or institutions resulting in unplanned service “silo” development. The number of genetic units and genetic testing

services is increasing; however, services are dominantly available at tertiary care level, as commercial out-of-pocket services and situated in affluent urban areas. Social and private insurance plans rarely cover genetic conditions. The exception is Oman where out-of-pocket payment does not play a significant role due to universal coverage. Due to these financial (affordability) and geographical barriers (concentration in main cities and non-availability in particular areas), genetic services are highly inequitable. Genetic services are accessible for the educated, affluent upper- and upper middle classes; the less affluent rural population is underserved.

Services in the public health sector are fragmented, underfunded and understaffed leading to excessive waiting lists that implicitly lead to non-transparent prioritisation and rationing. Lack of expertise and skill gaps to recognise genetic disorders by primary care providers result in delayed (or no referral at all) in all countries. Routine points of entry to genetic services at primary care level are very limited. Community genetic services near to patients and their families throughout the country are rare and can only be found to a certain extent in Oman, yet with restricted scope of services.

The lack of certified medical geneticists is a ubiquitous problem but is especially acute in Brazil, China, the Philippines and South Africa. The limitation in available medical geneticists not only severely hampers the ability of these countries to diagnose and manage hereditary disorders but also their ability to incorporate the benefits of genetic/genomic research into mainstream medicine.

South Africa is the only GenTEE country currently that has non-MD genetic counsellors, educated and trained following the model developed in the UK and USA.

In contrast to largely underdeveloped, underfunded genetic services in the public domain, the governments of Brazil, China, the Philippines and South Africa (India is not reported here), have put substantial resources into genetic, mainly genomic, research. Especially Brazil and China have developed cutting edge capacity to undertake genetic/genomic research in a remarkably short period of time. However, due to the failure to recognise congenital and genetic disorders as a priority health problem and lack of investment into the development of public genetic services, there is a striking mismatch between highly developed research capacities and the availability of an adequate public service infrastructure.

Nevertheless—maybe with the possible exception of South Africa—in all GenTEE countries, represented in this special issue, positive development to improve genetic service structures can be observed. Strengthening genetic services is a gradual process that can be facilitated by international networking activities.

This special issue is dedicated to Rodney Harris CBE, Emeritus Professor of Medical Genetics, University of Manchester, formerly Chair of the Department Medical Genetics, St. Mary’s Hospital Manchester, UK, on the

occasion of his 80th birthday. Rodney Harris has been a pioneer in setting up an international network of senior clinical geneticists to investigate the structure, workloads and quality of genetic services in 31 European countries. His initiative for the Concerted Action on Genetic Services in Europe (CAGSE), funded by the European Commission in the early 1990s, provided vital data to encourage medical genetic services consistent with the special needs of each country and to promote international co-operation (Harris 1997). GenTEE stands in this tradition.

Acknowledgments The CAPABILITY Special Support Action is funded by the EC 6th Framework Programme, contract number: LSSG-CT-2006-037275. The GenTEE survey was supported by (1) the European

Commission Joint Research Centre Institute for Health and Consumer Protection (IHCP) Ispra, Italy; (2) the Department of Human Genetics, Hannover Medical School, Hannover, Germany; and (3) the Unit of Women's Health Research, Medical School, Westfälische Wilhelms-University Muenster, Muenster, Germany. CAPABILITY and GenTEE were workpackages integral with EuroGentest1 (an EC funded Network of Excellence FP6-512148) and EuroGentest2 (an EC funded Coordination Action FP7-HEALTH-F4-2010-261469) respectively.

Reference

Harris R, Reid M (1997) Medical genetic services in 31 countries: an overview. *Eur J Hum Genet* 5(Suppl 2):3–21