

Genetic Testing in Emerging Economies (GenTEE) project

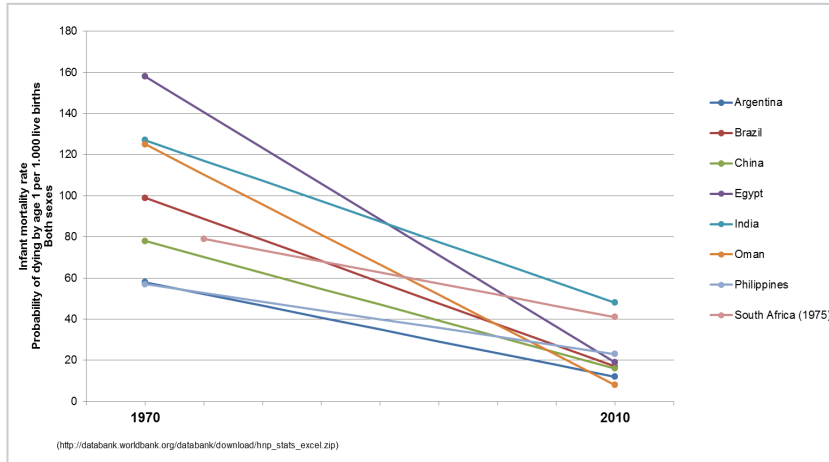


Due to the epidemiological transition in the emerging economies of China, East Asia, India, Latin America, the Middle East and South Africa

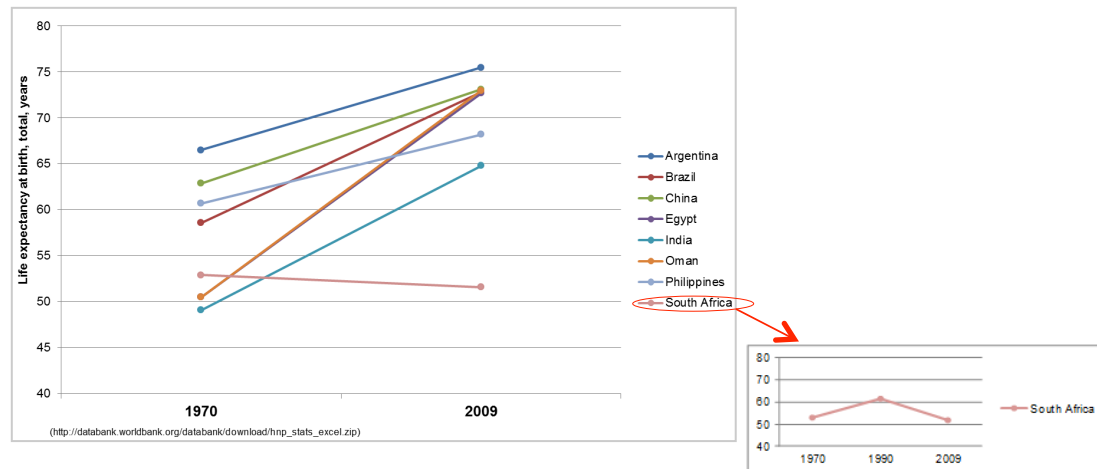


Epidemiological transition GenTEE countries 1970 - 2010

Infant mortality rate, probability of dying by age 1 per 1 000 live births



Life expectancy at birth, total, years



These economies are facing

- **an increasing proportion of infant morbidity and mortality due to congenital and genetic disorders**
- **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 risk 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 in these countries.

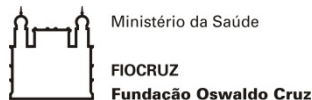
The challenges the emerging economies are facing are manifold:

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

GenTEE (Genetic Testing in Emerging Economies) is an international network that closely links leading academic/health/government institutions from eight emerging economies



Argentina Centre of Genetics and Public Health. Department of Health Sciences, Universidad Nacional de La Matanza, Buenos Aires



Brazil Centro de Genética Médica - Instituto Nacional de Saúde da Mulher, da Criança e do Adolescente Fernandes Figueira (IFF/Fiocruz), Rio de Janeiro



China Peking University Center of Medical Genetics, Beijing



Egypt Institute of Post Graduate Childhood Studies (IPGCS)/Ain-Shams University, Cairo



India Centre of Medical Genetics, Sir Ganga Ram Hospital, Rajender Nagar, New Delhi



Oman Genetic Unit, Directorate General of Health Affairs, Ministry of Health, Muscat



Philippines Department of Pediatrics, College of Medicine and Philippine General Hospital; Institute of Human Genetics, National Institutes of Health, University of the Philippines Manila



South Africa Division of Human Genetics, National Health Laboratory Service & University of the Witwatersrand

with European programmes and institutions that are tasked to develop, harmonize, validate and standardize genetic testing services in the 27 EU Member States

EuroGenetest

HARMONIZING GENETIC
TESTING ACROSS EUROPE



<http://www.eurogentest.org> EuroGentest2 Work Package (WP) 8:
“Best practice guidelines for the provision of genetic services”



JOINT RESEARCH CENTRE

Institute for Health and Consumer Protection (IHCP)

<http://ihcp.jrc.ec.europa.eu/>

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MHH
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Westfälische
Wilhelms-Universität Münster

European academic institutions & parent, patient organizations



**Department of Human Genetics,
Hannover, Germany** (*EuroGentest WP 10 Leader*)



**Women's Health Research,
University of Muenster,
School, Germany**



**Department of Clinical Genetics,
University Hospital, Lund University,
Sweden** (*EuroGentest WP 8 Leader*)



Genetic Alliance UK, UK (*EuroGentest WP 16 Leader*)

and other international networking activities



Global Network for Maternal and Infant Health



1995



2001



2010-

The GenTEE Global Network



Main GenTEE objectives

- to document and compare current practices and the state of genetic service provision in the participating GenTEE countries via a standardized survey (**GenTEE survey**)

Main GenTEE objectives

- to promote an internationally shared set of basic quality standards for genetic testing and the provision of appropriate genetics/genomics services that will facilitate future joint research, the exchange and transfer of knowledge and new technologies such as high throughput genome analyses via

GenTEE demonstration projects

Methods

The GenTEE survey

- **documents and compares current practices and the state of genetics service provision in participating GenTEE countries**
- **identifies current knowledge gaps**
- **identifies unmet service needs**

Methods

The GenTEE survey

validates a method/framework for ascertaining data on genetic services/testing development in emerging economies

that

allows comparison of services internationally across a number of key dimensions by using a core set of indicators, selected by the GenTEE Consortium for their relevance and comparability.

GenTEE Conceptual Framework

Survey

Key dimensions

INDICATORS OF CONGENITAL/GENETIC
„DISEASE BURDEN“

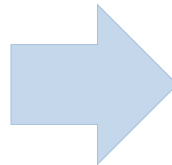
AVAILABILITY OF GENETIC SERVICES

ACCESS TO GENETIC SERVICES

GENETIC SERVICE DELIVERY

CIVIL SOCIETY ENGAGEMENT

GOVERNANCE/HEALTH & RESEARCH
POLICIES



Outcomes

GenTEE report

DATA/KNOWLEDGE GAPS

HEALTH SYSTEM RESPONSIVENESS

QUALITY OF ACCESS/COVERAGE

QUALITY OF CARE & SERVICE DELIVERY

NEED FOR SYSTEM STRENGTHENING
EFFORTS



DESIGN & FUND DEMONSTRATION
PROJECTS

The following health care context dimensions of genetic testing are targeted

- Prenatal testing and PGD
- Newborn screening
- Carrier screening (e.g. Haemoglobinopathies)
- Diagnostic testing for monogenic and congenital disorders
- Testing for common disorders with a major gene subgroup
- Pharmacogenetic testing
- Genetic susceptibility testing (e.g. for infectious diseases)

GenTEE survey data collection and report

- **Surveys have been completed end 2011**
- **Data are presented as "country reports"**
- **Each country report has been submitted to an external expert review for validation**

Publication dates:

Report: Summer 2012, by European Commission (IHCP)

Special issue Journal of Community Genetics: Early 2013

Results

Development of services: drivers (national)

- funded by research means
- dependent on the priorities chosen by individual academics acting in their country as early innovators and driving forces
- genetic services development fragmented, characterized by “enthusiasm based” decision-making by individuals or institutions, resulting in unplanned service “silo” development

Barriers: Availability of and access to services

Services are dominantly available

- at tertiary care level,
- as commercial/ out-of-pocket service¹ and
- in urban areas

in all countries

¹exception Oman where commercial services do not play a significant role due to universal coverage

Barriers: Availability of and access to services

No equitable access to genetic services mainly due to

- **financial barriers** (*affordability: out-of-pocket expenses in the private sector tend to be the norm in most GenTEE countries*)
- **geographical barriers** (*concentration of services in main cities*)
- **inadequacy of public services** (*fragmented, underfunded, understaffed*)
- **skill gaps** (*delayed or no referral*)

Genetic services are accessible for the educated, affluent upper, and upper middle classes, the less affluent, rural population is underserved.

Barriers: Lack of national data & health priority setting policies

- **failure to recognize congenital disorders/genetic diseases as a priority health problem**
 - **underfunded fragmented public services**
 - **lack of basic infrastructure facilities in rural areas**
 - **the private sector in several countries has stepped in during the last decade and increasingly provides genetic services not available in the public sector and services covered or not covered by private health insurance**

Barriers: Lack of national data & health priority setting policies

- **lack of national epidemiological data to demonstrate impact of congenital disorders/genetic diseases and need for services**
 - **lack of planned development**
- **translation gaps & lack of proven clinical validity/ utility of testing services**
 - **„enthusiasm-based“ decision-making**
 - **lack of SOPs and agreed QA processes for new technologies**

Opportunities

- **learning from the development in other countries**
- **avoiding implementation of expensive, obsolete „old“ technologies**
- **decreasing costs of DNA sequencing will lead to increased understanding of genetics/genomics**
- **use of new technologies with proven clinical utility will improve/refine diagnosis and management of diseases**
- **development of competencies and improved medical/professional training**
- **increasing availability of international/regional networks & activities to improve the provision of genetic/genomic services**
(Human Variome Project, HapMap project, WHO „63rd World Health Assembly (WHA) Executive Paper on Birth Defects, WHA63/17“, EuroGentest, ESHG, regional networks available in Arab countries & Gulf States, APSHG etc.)

Thank you for your attention



The GenTEE consortium