Research priorities in integrating community genetic services in primary care settings

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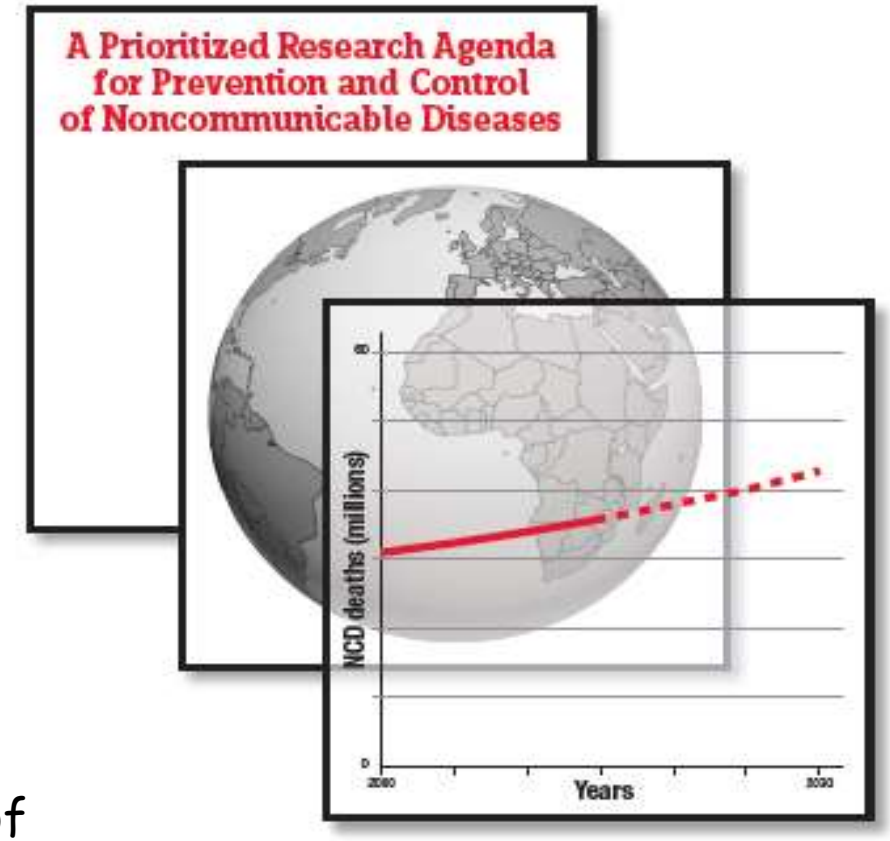

Globally at least 7.6 million children are born annually with severe genetic or congenital disorders. Nearly 90% of these infants are born in low- and middle-income countries.

World Health Organization

Develop methods and instruments for assessing the health and economic burden of congenital disorders

- Data on the prevalence of congenital disorders in the country (carriers, affecteds).
- Data on available genetic services, both clinical and diagnostic laboratory services.
- Data on available human resources in clinical genetics and genetic laboratory personnel.
- Data on number and capacities of special schools for the care and rehabilitation of affected.
- Outcomes for affected children in terms of survival.
- Data on college curricula related to genetics and genetic services.

Develop approaches applicable in primary health care for genetic risk assessment, information and counselling

- Develop approaches to facilitate equitable access to low-cost genetic testing and treatment of genetic disorders.
- Adapt medical record systems to include genetic and family history information.
- Develop informatics approaches for delivering information for health decision-makers, and instruments for professional education and information for patients and the community.

Develop community genetics services, suitable for the country

- Develop locally appropriate approaches for carrier screening of haemoglobinopathies.
- Develop locally appropriate information about reproductive risk and availability and acceptability of options for reducing it.
- Investigate requirements for patient care and cost-effective models of care to address birth defects.

Implement and evaluate preventive approaches for birth defects at:

- Before pregnancy (e.g. folic acid food fortification, detection and counselling for carriers, such as sickle cell anaemia or thalassaemia)
- During pregnancy (e.g. fetal anomaly scanning and other forms of prenatal diagnosis)
- At the end of pregnancy (anti-D to prevent rhesus haemolytic disease in subsequent pregnancies)
- In the newborn period (neonatal screening)
- Assess changes in affected birth prevalence and patient outcome in response to interventions addressing birth defects

Education

Develop cost-effective tools for delivering information on congenital disorders for health decision-makers, for patients and the community and for professional education

Gaps

- There is no agreed primary health care genetics curriculum (good educational material is available on the internet, but most is adapted to a high-income setting, and presented at too sophisticated a level).
- There is no agreement on the requirements for the verbal and graphical content of genetic information materials for practitioners, patients and the public.
- No validated instruments for assessing local epidemiology and costs, establishing surveillance, or providing information, education and training, are available to help decision-makers with planning genetics services.

Prerequisites for the organization of community genetic services program

- Reliable epidemiologic and burden of disease data collection, and needs assessment analysis are needed to focus on priorities in planning care and prevention programs for congenital disorders and genetic diseases.
- Data on the extent and availability of community genetic services, manpower and laboratory facilities in a country should be obtained.
- Political will and commitment and financial resources are needed for funding and planning programs for the care and prevention of congenital disorders.
- Creation of national committees in genetics and health care with a focal person in charge, in collaboration with the WHO Country Office, is essential for planning a community based care and prevention program for congenital disorders and genetic diseases.
- Knowledge and technology is available for treatment and/or prevention of specific genetic conditions.
- Health professionals are capable and interested in leading the actions.

Examples of research questions

Research questions related to consanguinity

Will diminishing the consanguinity rates diminish the prevalence of congenital disorders in the community?

Will diminishing the consanguinity rates diminish stability and solidarity of the family structure in the community?

Will diminishing the consanguinity rates diminish the infant mortality rate in the community?

What can we tell consanguineous couples requesting premarital and preconception counseling?

What are the safe, scientific and evidence-based messages that can be delivered through the media regarding consanguinity?

Questions related to the ethical, legal and religious issues for selective pregnancy termination

Guidelines for termination of pregnancy when fetus is affected depend on?

- Severity of disease?
- Survival?
- Impact on family?
- Impact on affected?
- Impact on community?

Recommendations

- Conduct epidemiological studies to determine the most prevalent congenital disorders and genetic diseases and the resulting health care needs.
- **Standardized methodology should be used.**
- Development of approaches applicable in primary health care, for genetic risk assessment, information and counselling.

- **Standardized methodology should be used for all research on consanguinity**
  - Inbreeding coefficient is the most accurate determinant of consanguinity in the population studied

Recommendations

- Increasing genetic and genomic literacy among the public, health care providers and policymakers through evidence-based WHO guidelines focusing on low and middle income countries.
- Development of internet-based systems for delivering community genetic resources for health decision makers, for professional education, and for patient information.