Community genetics services

Report of a WHO Consultation on community genetics in low- and middle-income countries

Geneva, Switzerland,
13–14 September 2010

World Health Organization

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Dpwfs; Denis Meissner WHO Graphics

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Executive summary

Community genetics has been defined as "the art and science of the responsible and realistic application of health and disease-related genetics and genomics knowledge and technologies in human populations (communities) to the benefit of individual persons." The objective of this Consultation was to develop an evidence-based report on community genetics services to provide guidance to low- and middle-income countries (LMIC) in accordance with the *2008–2013 Action Plan for the Global Strategy for the Prevention and Control of Noncommunicable Diseases* (NCDs). The goal of community genetics in LMIC is to prevent congenital disorders and genetic diseases at population level and, at the same time, to provide genetics services (diagnosis and counselling) in the community for individuals and families. The term "prevention" is used in this report to indicate actions implemented with the indivisible objectives of reducing the birth prevalence and health impact of congenital disorders and genetic diseases, while respecting voluntary reproductive decisions.

A Group of experts (listed at the end of this report), all internationally-recognized specialists in the field of community genetics in LMIC, was convened for the purposes of the Consultation. The Group agreed that availability of community genetics services in LMIC is less than adequate. This may be due to a number of reasons: paucity of resources; genetic conditions not being considered priorities; misconceptions that the control of common congenital disorders is too expensive and linked with sophisticated technology; low genetics literacy; cultural, legal and religious limitations such as the fear of stigmatization within the community and the legal or religious restrictions to selective abortion of affected fetuses; an insufficient number of trained health professionals; and inadequate data on the true magnitude and economic burden of congenital disorders.

Congenital disorders (birth defects) are defined as structural or functional abnormalities which are present from birth, whether recognized at birth or later. They constitute a major health problem worldwide especially in LMIC. Factors that may contribute to the high rates of congenital disorders in LMIC include the general low availability of public health measures directed at the care and prevention of these disorders, the high frequency of haemoglobinopathies in Africa, Middle-East and South-East Asia, the high consanguinity rates in the Eastern Mediterranean and South-East Asia regions that can increase the occurrence of recessively inherited diseases, advanced maternal age at conception in many low- and middle-income countries, which increases the predisposition to chromosomal trisomies such as Down syndrome, and large family size that may increase the number of affected children in families with autosomal recessive conditions.

It is worth noting, however, that prevention programmes have been successfully implemented in some low- and middle-income countries, thus reducing the burden of congenital disorders or genetic diseases. Examples of countries adopting such programmes are, among others, Bahrain Cyprus and Iran.

The services required for the prevention and care of congenital disorders and genetic diseases include prevention strategies at primary, secondary and tertiary health-care levels. The Group emphasized specifically the need for education in genetics to be provided to all health professionals, policy-makers, and the general public. This also includes the sensitization to ethical, legal and social issues (ELSI) which are of key concern in the context of congenital disorders and

genetic diseases. WHO regional offices, international experts, academic institutions, professional organizations, philanthropic organizations, parent–patient associations, private sectors and donors need to work together for the implementation of community genetics services in LMIC.

In conclusion, the Group recommended all appropriate stakeholders convene to carry out the following:

- conduct epidemiological studies to determine the most prevalent congenital disorders and genetic diseases and the resulting health-care needs, through the establishment of registers for these conditions;
- establish clear and measurable objectives and goals;
- determine the most efficient interventions to achieve the specific goals;
- implement the interventions identified, following the ethical, legal and social guidelines, and evaluate outcomes.

In relation to determining the most efficient interventions, experience and examples from different countries indicate that the most common genetics services at community level (i.e. "minimum package of interventions") are:

i) training health professionals in basic concepts of genetics and their application to community genetics services;

ii) use of family history as an instrument to detect genetic risks;

iii) pre-conception and prenatal care, including folic acid fortification or supplementation, addressing the genetic risks of maternal age and consanguinity, genetic counselling when needed, prenatal ultrasound screening to detect fetal abnormalities, and referring detected genetic problems to a tertiary centre for proper management;

iv) public education to avoid alcohol, tobacco and potential teratogens in gestation;

v) carrier detection for haemoglobinopathies in countries where these conditions are a significant burden to public health, followed by genetic counselling and the option of prenatal diagnosis where voluntary termination of affected pregnancies is acceptable;

vi) clinical detection of congenital disorders in newborns and appropriate referrals to higher levels of care for diagnosis and treatment;

vii) newborn screening for congenital conditions in which early intervention is effective, such as congenital hypothyroidism, followed by treatment and other services for life. Newborn screening for haemoglobinopathies should be considered in countries where hereditary anaemias constitute a significant burden of disease.

From the perspective of WHO Human Genetics, it would be desirable to start by supporting the development of community genetics services in selected countries as demonstration projects.

1. Objective of the Consultation

The objective of the WHO Consultation was to develop an evidence-based report on community genetics services to provide guidance to low- and middle-income countries in accordance with the 2008–2013 NCD Action Plan. The Group of experts noted that the World Health Organization (WHO) has dealt with issues related to prevention and control of genetic diseases and congenital disorders and the organization of genetics services at community level on several occasions in past years (Alwan & Modell, 1997; WHO 1999; WHO 2006a; WHO 2006b).

2. Definitions

2.1. Community genetics

The Group endorsed the definition of community genetics as being "the art and science of the responsible and realistic application of health and disease-related genetics and genomics knowledge and technologies in human populations (communities) to the benefit of individual persons." (ten Kate et al., 2010). They noted, however, that in low- and medium-income countries, genetics services at community level are usually planned and implemented by the public health sector of the state. The dual and indivisible goal is to prevent congenital disorders and genetic diseases at population level and, at the same time, provide genetics services (diagnosis and counselling) in the community for individuals and families. The public health component of this indivisible goal of population health promotion and reduction of the prevalence of congenital disorders and genetic diseases, is not in contradiction with the genetics services component for individuals and families. Both should respect the right of individuals to make autonomous decisions in health and reproductive matters without coercion of any kind and from any source.