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WHO events addressing public health priorities

[1] This report is extracted from the Summary report on the Expert meeting on the prevention of congenital and genetic disorders in the Eastern Mediterranean Region, London, United Kingdom, 29-31 July 2016 (http://applications.emro.who.int/docs/IC_Meet_Rep_2016_EN_18989.pdf?ua=1, accessed 30 March 2017).

Maternal and child health is one of five public health priorities in the WHO Eastern Mediterranean Region, as agreed with Member States (1), and congenital and genetic disorders (CGDs) remain important public health problems related to maternal and child health, communicable diseases, non-communicable diseases and emergency health care, within the framework of health system strengthening. In order to support work on CGDs, the WHO Regional Office for the Eastern Mediterranean (WHO-EMRO) held an expert meeting on the prevention of CGDs in the Eastern Mediterranean Region (EMR) in London, United Kingdom of Great Britain and Northern Ireland (UK), from 29 to 31 July 2016 (2). The meeting was attended by experts from American University of Beirut, Lebanon; Geneva University, Switzerland; London School of Hygiene and Tropical Medicine, UK; and University College London, UK; as well as representatives from the March of Dimes Foundation, United States of America, and the PHG Foundation, UK. The objectives of the meeting were to:

review the magnitude of CGDs, including their main causes, in Member States of the Region;

agree on key interventions that are evidence-based, high impact, cost-effective and feasible to implement by national health systems; and

identify basic requirements for national programmes, including capacity to strengthen the prevention and control of CGDs.

WHO defines congenital disorders, also known as birth defects, as structural or functional anomalies that occur during intrauterine life and can be identified prenatally, at birth or later in life (3). Discrepancies in prevalence depend on the method of estimation. Differences exist between estimates using the global burden of disease versus national data. These can be reduced by adopting accepted definitions for congenital anomalies and disorders, by comparing observational data on well-known congenital disorders collected by countries with those computed via global burden of disease, or by involving countries in the process of computation to assess the accuracy of CGD estimates.

The Region has a higher prevalence of CGDs compared to other regions in the world. Even when comparing the highest income countries of the Region to countries in western Europe, it is clear that the Region has higher CGD prevalence rates (4). Various countries of the Region have social practices that contribute to the increase in the incidence and prevalence of CGDs. These include the preference and support for consanguineous marriage, and the lack of laws to support termination of pregnancy where the congenital disorder is diagnosed early enough in pregnancy (4).

The high consanguinity rate of 20–50% in most countries of the Region has been highlighted as a main predictor of autosomal recessive genetic disorders and haemoglobinopathies, including thalassemia and sickle-cell disease (4). The prevalence of CGDs has been shown to be affected by: the presence of early detection mechanisms; genetic counselling; and allowing for interventions to reduce the number of newborns with defects, which range from food fortification to fetal surgery and termination of pregnancy.

Policies are needed to ensure that CGDs are prioritized and included in basic health services packages at the primary health care level. (2). Moreover, interventions to reduce the burden of CGDs in the Region are not easily available to all families since they are costly and couples may not necessarily be aware of their existence. Hence, universal coverage is one of the main challenges for services aiming to reduce CGDs. Paediatric surgery is a key intervention to prevent avoidable newborn and infant death and in gaining years of life cured or without disability.

It was recommended that the following primary and secondary interventions, if adopted, would contribute to the reduction of the CGD occurrence. Interventions during the preconception (including premarital) period, targeting all women of childbearing age, including:

immunization for Rubella and Hepatitis B virus;

fortification with vitamin B12, folic acid, iodine and iron;

screening for carriers of common autosomal recessive disorders in the Region, including beta thalassemia and sickle cell anaemia;

assessing maternal age distribution, genetic family history, and referral of high-risk families;

screening and treatment of infections such as syphilis and HIV/AIDS, among others;

screening, diagnosis and management of diabetes, anaemia and hypertension;

avoidance of tobacco use; and

increasing public awareness of risk factors, such as unhealthy diet, sedentary lifestyle and common genetic risk factors.

Interventions during pregnancy, including:

early detection and management of maternal conditions such as diabetes;

early detection and management of infections;

avoidance of teratogens (infections such as toxoplasmosis, drugs);

prenatal screening by maternal serum markers in first trimester and by ultrasonography;

prenatal diagnosis with/without termination of pregnancy;

care of fetus for conditions such as Rh incompatibility;

avoidance of tobacco use and exposure to pollution; and

supplementation with iron and folate.

Interventions after birth, including:

newborn biochemical screening for congenital hypothyroidism, phenylketonuria (PKU), galactosaemia, sickle cell disorder, glucose-6-phosphate dehydrogenase (G6PD) deficiency, congenital adrenal hyperplasia, methyl coenzyme dehydrogenase deficiency;

newborn screening for hearing impairment, congenital hip dislocation, isolated cleft palate, heart defects and other clinically identifiable congenital disorders;

stillborn examination and investigations;

general newborn management and of identified conditions, and paediatric surgery to correct malformations;

rehabilitation and palliative care;

family support, including bereavement;

diagnosis of CGDs, with management and counselling families on future reproductive options; and

extended family screening testing and counselling.

CGDs constitute a burden in the EMR and birth prevalence of congenital disorders remains highest in the Region compared to other WHO regions. Disability is the main problem presented by congenital disorders and single gene disorders remain the most difficult and expensive to treat (2). The observed rates of major congenital malformations vary across the Region and common autosomal recessive disorders in countries are alpha thalassemia (carrier rate ranges between 2–50%), beta thalassemia (carrier rate ranges between 2–7%) and sickle cell anaemia (carrier rate ranges between 0.3 and 30%) (3,4). Consanguinity is the highest risk factor in the Region and there is no availability of services to provide counselling for couples. Assessment tools are needed to conduct measurement of CGD birth prevalence in countries, adopting a standardized epidemiological approach to produce accurate CGDs estimates per country and to enable comparison (2,3).

There exist evidence-based interventions to prevent CGDs and reduce the burden of disability among children in the Region (5). Paediatric surgery has a significant effect on reducing the newborn and infant mortality burden, while termination of pregnancy is recognized as an effective intervention in reducing the number of newborns with haemoglobinopathies and other CGDs. It is imperative that all efforts to strengthen health systems and reach universal health coverage must include the prevention and management of CGDs in the package of services (2).

However, there are some encouraging initiatives in the Region, but these need validation and improvement. A regional initiative to support countries to develop their own country-specific strategy for control of CGDs is crucial for the reduction of newborn and infant mortality and morbidity. There is a need for countries to use scientific evidence to prioritize the prevention and

care of CGDs in order to improve newborn and child health outcomes (2).

Genetic counselling is essential to prevent CGDs by providing accurate and correct information; hence, comprehensive efforts are needed in medical genetics education and training including genetic counselling. Tools for providing information, education and counselling need to be standardized to be delivered by health providers at primary health care level. The use of innovative methods is crucial, as is strengthening health system components to be able to integrate CGD prevention activities, especially into basic and primary healthcare services. Meanwhile, advocacy remains crucial to ensure the commitment of policy-makers at country level and to integrate CGD preventive and management services into existing health care programmes, ranging from individual care services to public health programmes. Moreover, services should consider targeting every woman, every time.

References

1. World Health Organization. Shaping the future of health in the WHO Eastern Mediterranean Region: reinforcing the role of WHO. Cairo: WHO Regional Office for the Eastern Mediterranean; 2012 (http://applications.emro.who.int/dsaf/EMROPUB_2012_EN_742.pdf, accessed 30 March 2017).

2. World Health Organization. Expert meeting on the prevention of congenital and genetic disorders in the Eastern Mediterranean Region, London, United Kingdom of Great Britain and Northern Ireland 29-31 July 2016 (http://applications.emro.who.int/docs/IC_Meet_Rep_2016_EN_18989.pdf?ua=1, accessed 30 March 2017).

3. World Health Organization. Congenital anomalies fact sheet. Geneva: World Health Organization; 2016 (http://www.who.int/mediacentre/factsheets/fs370/en/, accessed 30 March 2017).

4. March of Dimes. Global report on birth defects: the hidden toll of dying and disabled children. New York: March of Dimes Birth Defects Foundation; 2006 (http://www.marchofdimes.org/materials/global-report-on-birth-defects-the-hidden-toll-of-dying-and-disabled-children-full-report.pdf, accessed 30 March 2017).

5. Hamamy H, Alwan A. Genetic disorders and congential abnormalities: strategies for reducing the burden in the Region. East Mediterr Health J. 1997;3(1):133–143.

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