Summary report on the

Expert meeting on the prevention of congenital and genetic disorders in the Eastern Mediterranean Region

WHO-EM/WRH/101/E

London, United Kingdom of Great Britain and Northern Ireland
29–31 July 2016

World Health Organization
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1. Introduction

The WHO Regional Office for the Eastern Mediterranean held an expert meeting on the prevention of congenital and genetic disorders (CGDs) in the Eastern Mediterranean Region in London, United Kingdom of Great Britain and Northern Ireland, from 29 to 31 July 2016. The meeting was attended by experts from American University of Beirut, Geneva University, London School of Hygiene and Tropical Medicine, and University College London (UCL), as well as representatives from the March of Dimes Foundation and the PHG Foundation.

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 a programme at national level and requirements in terms of capacity to strengthen the prevention and control of CGDs.

In his opening remarks, Dr Ala Alwan, WHO Regional Director for the Eastern Mediterranean, noted that maternal and child health is one of five public health priorities in the WHO Eastern Mediterranean Region, as agreed with Member States. He said that CGDs remain an integral part of the unfinished agenda of maternal and child health, communicable diseases, noncommunicable diseases and emergency health care, within the framework of health system strengthening.

The Regional Director referred to the work done over the years to highlight the importance of CGDs and emphasized the need for action, particularly on haemoglobinopathies, noting that a number of CGDs

are of high prevalence in the Region. A publication on community control of genetics and congenital disorders had been published by the Regional Office in 1997 and disseminated to Member States with the aim of supporting national programmes to adopt new approaches towards reducing the burden of CGDs. However, due to competing priorities at the time, efforts had been slow to identify and target national CGD priorities.

Dr Alwan emphasized that with the focus on health within the Sustainable Development Goals (SDGs) and the need to ensure that “no one is left behind”, we need to package interventions to reduce CGDs, and their contribution to neonatal mortality and lifelong disabilities, within the framework of universal health coverage. Interventions should ideally accommodate the three principles of universal health coverage, namely: coverage of services, financial protection and access to quality essential health services. Preventive, curative and rehabilitative essential CGDs services should be delivered within the basic package of essential services at the primary health care level.

Moreover, the Regional Director highlighted the importance of strengthening our partnerships with United Nations Children's Fund (UNICEF) and United Nations Population Fund (UNFPA) in order to link our activities closely to existing initiatives to accelerate progress towards achieving maternal, newborn and child health indicators through existing mechanisms, such as the pre-conception package of services, the every newborn action plan (ENAP) and maternal, newborn, child and adolescent health plans.

2. Summary of discussions

Participants focused on the burden of CGDs in the Eastern Mediterranean Region, highlighting that the Region has a higher burden compared to other regions, and discussing tools to collect data to

estimate the prevalence of CGDs at birth and their distribution within the Region. There was consensus on the importance of adopting innovative technologies and standardized methods to collect accurate data on CGDs and enable comparative studies and research between countries. Moreover, participants highlighted surveillance of newborns as one of the main tools that can help to detect birth defects and assess their contribution to newborn and infant mortality and disability.

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. Congenital disorders and birth defects are defined as either environmental or constitutional.

Discrepancies in prevalence depend on the method of estimation. Differences exist between estimates using the global burden of disease and 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. Comparing the highest income countries of the Region to countries in western Europe, it is clear that the Region has higher CGD prevalence rates. Most 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.

The high consanguinity rate of 20–50% in most countries of the Region was highlighted as a main predictor of autosomal recessive

genetic disorders and haemoglobinopathies, including thalassemia and sickle-cell disease. 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 foetal surgery and termination of pregnancy.

Many of the disorders that are commonly diagnosed in the Region are preventable using low cost, high impact interventions such as food fortification and genetic counselling. Policies are needed to ensure that CGDs are prioritized and included in basic health services packages at the primary health care level. Facilities to properly diagnose and manage CGDs are few and do not provide all needed services. Richer families have the opportunity to outsource laboratory tests to Europe or selected countries in Asia. The Region does not have a laboratory to provide such services for early detection of congenital disorders.

Interventions to reduce the burden of CGDs in the Region are not available to all families. They are costly and couples may not 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 noted that disability at five years of age is increased with the introduction of interventions that reduce mortality due to CGDs. This will require improving the coverage and access to health services for people with disability.

The participants agreed that the main pillars for developing a national CGD programme include: the commitment of policy-makers, both politically and financially by allocating necessary resources; strengthening human resources; integration into primary health care settings; increasing public awareness and literacy; respecting ethical,

legal and religious norms; surveillance and national registries; population screening programmes; laboratory facilities, including DNA diagnosis; and genetic centres.

They also agreed that the essential genetic services required in a national programme should include: prevention and screening programmes; laboratory diagnostic facilities, including DNA; counselling in primary care; outreach and education to the public, including in schools; specialized genetic service facilities for diagnosis, training and research; and basic needs in human resources, including education and training of health professionals.

Participants further agreed that health professionals involved in CGD interventions should include: geneticists; trained primary health care providers (for identification of cases and recognizing risks); community-based genetic counsellors; laboratory personnel; specialists in obstetrics and paediatrics; family medicine practitioners; and specialist nurses.

The participants discussed the role of preconception care in promoting women's and couples' health before the occurrence of conception in order to improve pregnancy-related outcomes. Optimizing a woman's physical, mental and social well-being in preconception care includes obtaining accurate information about her health status, general lifestyle, nutrition and genetic conditions in the family followed by the appropriate counselling, prevention and management interventions as needed. The group reviewed the regional preconception care package, including core and expanded packages of recommendations and interventions targeting women/couples (married, planning for marriage or planning for pregnancy). They agreed on the importance of adopting the four preconception care areas at regional level: history, medical assessment, counselling and education, and prevention and management.

The participants debated the results of the regional preconception care assessment survey that highlighted the variation in coverage of the four areas of preconception care within the Region. The survey clearly indicated that collecting data on family history and counselling are the two components with the highest coverage, and that medical assessment, prevention and management are the components with the least coverage in the Region. It was also noted that the preconception care intervention coverage targeting CGDs varied between countries, with folic acid, complete blood picture and HIV test being the most frequently provided services in the Region. Genetic typing and immunization are the least commonly provided services in the Region.

Participants acknowledged the crucial role of strong partnerships to advance work on CGDs, including with United Nations agencies, nongovernmental organizations and community genetic services. Country experiences were discussed to show the impact of adopting cost-benefit interventions, including those on strengthening the prevention of congenital disorders and empowering women, their families and communities to reduce the risk of the CGDs by improving lifestyle and nutrition, preventing infection and adopting contraception care. Country experience in mobile health (m-health) showed its efficiency in identifying the carrier and managing CGDs cases in Punjab, Pakistan. The m-health staff was trained on thalassemia prevention and management, and services at district level included counselling, blood collection, result feedback and awareness.

The group agreed on the importance of having three kinds of tool to collect information on the epidemiology of CGDs, including: (i) tools for data collection to assess CGD status at country level and to monitor the effect and progress of CGD-specific interventions; (ii) tools for technical guidance to provide information on how to implement interventions and on know-how; and (iii) tools for communication and

dissemination of genetic information. A toolkit was presented and discussed during the meeting and the adoption of a CGD surveillance system, including innovative tools such as m-health, was proposed. Some applications used by the UCL Centre for Health Informatics and Multiprofessional Education were presented. The Geneva Foundation for Medical Education and Research shared tools integrated in training courses on sexual reproductive health research based on e-learning and training courses on genetic risk and drawing pedigrees.

3. Recommended interventions

The participants recommended the following interventions that, if adopted, would contribute to the reduction of the prevalence of CGDs.

Interventions during the preconception (including premarital) period, targeting all women of childbearing age, including:

- immunization for rubella and hepatitis b virus;
- fortification with folic acid and iron;
- fortification with vitamin B12 (although the evidence is still not conclusive) and iodine;
- screening for carriers of common autosomal recessive disorders in the Region, including beta thalassemia and sickle cell disease;
- triage: assessing maternal age distribution, genetic family history, referral of high risk families;
- screening and treatment of infections: syphilis, HIV/AIDS, others;
- screening, diagnosis and management of diabetes, anaemia and hypertension;
- blood grouping, including Rh;
- 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:

- management of maternal conditions such as diabetes;
- treatment of infection;
- 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 foetus for conditions such as Rh incompatibility;
- avoidance of tobacco use and exposure to pollution; and
- supplement 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;
- management: general newborn management and of identified conditions, and paediatric surgery to correct malformations;
- rehabilitation and palliative care;
- family support, including bereavement;
- diagnosis of the CGDs, with management and counselling families on future reproductive options; and
- extended family screening testing and counselling.

4. Conclusions

CGDs constitute a burden in the Eastern Mediterranean Region 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.

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%). 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.

There exist evidence-based interventions to prevent CGDs and reduce the burden of disability among children in the Region. 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.

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.

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.

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 a public health programme. Services should consider targeting every woman, every time.

5. Next steps

WHO

- Prepare country CGD profiles based on existing data.
- Convene an intercountry meeting to verify and refine the profiles and advocate for the inclusion of high-impact interventions in the basic package of health services.
- Adapt the surveillance tool and implementation guidelines to support Member States to conduct a CGD situation analysis and identify gaps related to health services.
- Support Members States to establish a system for regular data collection on CGDs.

- Update the regional survey tool that was used to assess the adoption of pre-conception care interventions.
- Update the WHO publication, Community control of genetic and congenital disorders (1997).

Member States

- Review and verify the country profile of CGDs.
- Establish national expert committees to support country-level activities to address the burden of CGDs.
- Develop the necessary policies, strategies and key interventions to support the reduction of CGDs.
- Support the establishment of a regional laboratory hub to reduce the costs and time taken for genetic testing.

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