The establishment of congenital malformations surveillance system in Albania – A national necessity

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Abstract

Background: Congenital malformations (CMs) represent an important public health problem worldwide. Until now, only spina bifida and congenital hip dislocation were officially reported to the Ministry of Health (MoH). Poor health statistics, lack of a CMs' national registry and surveillance system as well as relying only on hospital data have contributed to systematically underestimated prevalence of CMs in Albania. Our aim was to establish the congenital malformation surveillance system in order to highlight the detailed structure of CMs in Albania and to identify their potential risk factors.
Methods: In 2009, a multi-disciplinary team under the leadership of the Institute of Public Health worked for the conception of a new official reporting form and national registry of CMs. All related health professionals were trained on the necessity to report CMs and how to fill in the official form.
Results: The new reporting form is now made obligatory for both public and private system clinics through a dedicated MoH order. Two years after these efforts, around 80% of districts are using the new reporting form whereas 50% report electronically to IPH. The information on CMs can now be classified according to various diagnoses and national and regional analyses by the type of system affected and socio-demographic characteristics are possible.
Conclusion: The establishment of the new surveillance system of CMs in Albania shows very promising and will contribute to highlight the structure and risk factors of CMs, a very under researched area in this Eastern European country.

Keywords: *Congenital malformations, Albania, Surveillance, Epidemiology.*

Background

Worldwide situation

Congenital malformations (CMs) are a diverse group of disorders of prenatal origin which can be caused by single gene defects, chromosomal disorders, multifactorial inheritance, environmental teratogens and micronutrient deficiencies. Even though there are currently no sound estimates of the number of children born with serious congenital disorder(s) attributable to genetic or environmental causes (1,2) some risk factors have been studied extensively. For example, maternal infectious diseases such as syphilis and rubella are a significant cause of CMs in low and middle income countries. Maternal illnesses like diabetes mellitus, conditions such as iodine and folic acid deficiency, exposure to medicines and recreational drugs including alcohol and tobacco, certain environmental chemicals, and high doses of radiation are other independent risk factors for CMs (2).

CMs represent an increasing public health concern (1). As stated in the "Global Report on Birth Defects 2006" issued by the March of Dimes (MOD), every year there are 7.9 million children born with a serious birth defect of genetic or partially genetic origin accounting for 6% of all births worldwide (1). CMs can cause spontaneous abortions and stillbirths and are a significant but under recognized cause of mortality and disability among infants and children under five years of age. They can be life-threatening, result in long-term disability or cause mental, physical, auditory and/or visual impairment, and negatively affect individuals, families, health-care systems and societies(2)- Worldwide, at least 3.3 million children up to five years of age die from birth defects each year and an additional 3.2 million experience different permanent degrees of disabilities (1). While birth defects are recognized as a global problem (1-7), it is an especially challenging problem for the developing countries where more than 94% of serious birth defects and 95% of deaths from such defects occur (1). However, CMs are most prominent as a cause of death in settings where overall mortality rates are lower, for example in the European Region, where as many as 25% of neonatal deaths are due to congenital malformations (2,8,9).

The most common serious birth defects worldwide were: congenital heart defects, neural tube defects, hemoglobin disorders (thalassemia and sickle cell disease), Down syndrome and glucose-6-phosphate dehydrogenase deficiency (1). In many cases, genes and environment contribute together to cause a birth defect (3,5,10). There is little doubt that birth defects can cause enormous harm where elevated risk factors are present and health care is limited (11). Today, there are many opportunities to prevent a large number of birth defects and to reduce their negative consequences for the individual and the society. This can be done at a reasonable cost by simple public health methods and interventions (11).

Situation in Albania and the previous CMs reporting way

According to March of Dimes, from 192 countries included in the global report on birth defects, Albania ranks among the countries with a high prevalence of congenital malformations - 52.9/ 1000 live births (1). Among all diagnosed congenital malformations, District Public Health Directories (DPHD) report to the Ministry of Health (MoH) only spina bifida (8.52/1000 live births in 2006) and hip dislocation (1.23 /1000 live births in 2006). Until 2009 there was no information about major or minor congenital malformations of genetic or partially genetic origin (12,13). According to the International Classification of Diseases, 9th revision (ICD-9), the doctors and/ or nurses that assist in deliveries performed in all maternity wards and homes of Albania are requested to record all congenital malformations among *live born and dead born babies*. This is a mandatory process according to the Law of Statistic No. 7687 date 16.03.1993. Every DPHD in Albania routinely collects these data from the respective maternity wards and home deliveries. As mentioned, the DPHDs report every six months to MoH the total number of congenital malformations, the total numbers of spina bifida malformations and hip dislocations (among all other congenital malformations identified). They report these data as an aggregate number (including other data) by two official reporting forms, namely "Form 4/ Sh" and "Form 12/ Sh/ 1". The Form 4/ Sh includes data on obstetrical activity in all health institutions and delivery homes (if any). This form is reported to MoH every six months. The Form 12/Sh/1 includes data on infant mortality (less than 1 year of age) and it is reported on monthly basis to the MoH (12). Every child born alive with a congenital malformation has his/ her own clinical card kept in the statistical office of the maternity hospital. For the children with congenital malformations born dead or that die during the first year of life because of a congenital malformation, the death certificate is compiled. Congenital malformations are registered using these clinical cards and death certificates using ICD-9 codes by doctors or midwifes in all the maternity wards of the country.

However, without any obvious reason, the DPHDs detail on and report only spina bifida and hip dislocation among all malformations identified in Albania (12), since the two reporting forms require only these two categories and the total number of CMs. There was no information if the private clinics diagnosed any congenital malformations or recorded the diagnosed ones, nor did they report such information to the MoH. This way only the public sector reports them (12).

Rationale for the establishment of the CMs surveillance system

Constrained diagnostic capability, poor health statistics, lack of CMs surveillance and registries have led to a systematic underestimation of the incidence rates of congenital malformations in Albania.

This situation consequently raised the necessity of building up the congenital malformation surveillance system (CMSS). The aim of CMSS is the recording and reporting of CMs, through a newly conceived official reporting form, diagnosed during pregnancy, pregnancy interruption, births, fetal deaths and up to 2 years of age, in order to highlight the situation of congenital malformations in our country, to identify risk factors of genetic and/ or non-genetic origin and help policy makers to investigate and implement appropriate CMs' preventive policies. The new and improved way of reporting would, more specifically serve the following array of objectives (14,15):

- produce data for planning, advocacy, education and prevention;
- determine the annual prevalence of congenital malformations among all live births;
- determine the major congenital malformations diagnosed in our country according to geographical distribution and demographic characteristics of parents;
- investigate the increase in the prevalence of congenital malformations;
- conduct epidemiological studies on specific congenital malformations;
- provide useful information for maternal and child health programs;
- evaluate the effectiveness of various congenital malformations prevention programs;
- respond to the medical requirements related to congenital malformations;
- collaborate with other institutions that provide services for children living with congenital malformations.

Pathway for the establishment of the CM surveillance system

In 2009, the Institute of Public Health (IPH) in collaboration with MoH and with the support of UNFPA started the implementation of the CMSS as a first step towards identification of major birth defects in Albania and identifying potential genetic, environmental, nutritional and other factors potentially associated with congenital malformations. In early 2009, was established the Order of the Ministry of Health (dt.05. 03. 2009, NO. 157) on compulsory reporting of CMs identified in pregnancy, induced abortions, birth, fetal deaths and up to 2 years of age in public and private sectors of our country. This Ministerial Order outlines clear directives for a number of stakeholders in the process. The reporting centers comprise all maternity hospitals/pediatric wards and the primary health care centers in all districts and private hospitals licensed by the Ministry of Health that provide pediatric and obstetric-gynecological services. Each congenital malformation diagnosed is now reported through the newly conceived confidential official form (individual form 4/ 1/ ID-SH). Following these efforts, all obstetrician-gynecologists, neonatologists and pediatricians were trained on CMs reporting details, and also all reporting health institutions in the country were supplied with the registry of congenital malformations. All reporting centers are legally obliged to send the reporting forms to the DPHDs on monthly basis. The Reproductive Health Inspector of each DPHD is responsible for organizing, coordinating the work in that district for gathering CMs individual forms in public and private health institutions and subsequently for sending these reporting forms to the IPH every three months (12,13).

The IPH is the final center where the reporting forms are collected, analyzed and reports and recommendations are issued. It reports periodically to MoH.

Core elements of the new CM surveillance system

The three key aspects of the CMSS are: 1) case identification and ascertainment; 2) data collection;

and 3) data use and surveillance's evaluation (14,16). Regarding *the first key element*, criteria for CMs case ascertainment were established and agreed upon. Congenital malformations can be defined as structural or functional abnormalities, including metabolic disorders, which are present from birth. Therefore, specific health diagnoses (congenital malformations, deformations and chromosomal abnormalities) that fall under the chapter XIV of ICD-9 system, are going to be reported. All congenital malformations identified during pregnancy, induced abortions, birth, fetal deaths and up to 2 years of age in public and private sectors of the country shall be registered and reported (12,13). Congenital malformations like inborn errors of metabolism and blood disorders of prenatal origin that appear in other chapters of ICD-9 will not be part of CMSS. Regarding *the second key element* of CMSS, it was established that the reporting form would collect basic demographic and epidemiologic information on mother, father and fetus/ infant/ child with congenital malformations. Obstetrician-gynecologists, neonatologists and pediatricians shall fill in the individual reporting forms for each diagnosed case and then the reproductive health inspector of each public health district directory shall collect the reporting forms in each reporting center every three months and send them to IPH (12,13). Regarding *the third core element*, using CMSS data will be very important and beneficial for a range of stakeholders: public health experts, health professionals, policy makers and the public in order to fulfill all of its objectives and to inform on many of the issues described previously. The evaluation of CMSS is fundamental and needs to show improvement of currently available information on congenital malformations as well as improvements in the quantity and quality of data collected. It ensures that birth defect case reporting is complete, accurate, appropriate, and within the guidelines for timely reporting (14).

Results of the intervention

Now, in Albania all obstetrician-gynecologists, neonatologists and pediatricians (of all maternity wards, pediatric wards and obstetric-gynecologic wards) are trained for reporting congenital malformations by the official reporting form. All the congenital malformations reporting health centers in the country have the Congenital Malformation Registry. All reporting centers report congenital malformations forms to DPHDs. The results of such training are as follows: there are 354 medical doctors (pediatricians, neonatologists, obstetricians and gynecologists) trained for registering and reporting congenital malformations through the individual reporting form. There are 37 persons of the DPHDs' staff (reproductive health inspectors and other) trained for reporting the individual reporting form and monitoring the whole process. After two years into these efforts, around 80% of country districts are using the reporting system and in 50% of them the electronic way of reporting has been installed (13).

Now, the IPH is the final center which monitors the progress of the CMSS. It also collects the reporting forms, analyzes the data, produces reports and disseminates findings. The IPH in collaboration with the MoH provides appropriate recommendations for congenital malformations control, care and prevention.

Discussion

Congenital malformations are an important and very sensitive public health problem with a tremendous emotional and psychological impact on families. A number of risk factors for congenital malformations have been identified, including low socioeconomic status, advanced maternal age; however the factors that contribute to congenital malformations remain unknown. The CMSS activities have the potential to inform, complement and enhance existing mother and child health programs. Services and interventions for the prevention and care of birth defects should be part of existing health-care services, in particular those concerned with maternal and child health. They should combine the best possible patient care with a preventive strategy encompassing education, preconception care, population screening, genetic counseling, and the availability of diagnostic services (2,16). Effective delivery of services for the prevention and care of birth defects depends on the availability of a range of specialist clinical and diagnostic services, and a primary health-care system that is able to use them. Conventional laboratory services (hematological, microbiological, and biochemical) need to be supplemented with cytogenetic and DNA-based diagnostic services (2,15). Even though the reporting rate is at 80% there are still problems regarding the certainty of the diagnosis (suspect/ confirmed), filling the forms and reporting them, and also there is need for continuous training of the public health district directories staff due to the quick replacement of the staff from their job positions. Up to now, the major challenge of CMSS properly functioning is to convince medical specialists to fill in the reporting forms of congenital malformations (13), as they often are "too busy" to do that by themselves and leave this responsibility to nurses or statistical office employers

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