Epidemiology of congenital malformations in Albania during 2011-2012

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Abstract

Aim: The aim of this study was to present an overview of the regional distribution of congenital malformation (CMs) in Albania in order to provide researchers and policymakers with scientific information for implementing appropriate CMs prevention policies and consider local health care implications
Methods: This is a cross-sectional study implemented on national level. The data available for this study were extracted from the CM surveillance system of the National Institute of Public Health, during 2011-2012. Absolute numbers and frequencies of CMs according to mothers' and birth-related characteristics were reported. Chi square test was used to check for any statistically significant difference.
Results: The total reported rate of CMs per 1000 live births was 15.2 in 2011 and 14.3 in 2012. In both years, the majority of CMs cases were males. The most frequent CM affected the cardiovascular system. There was a significant decline in the proportion of reported CMs of central nervous system and chromosomal defects between 2011 and 2012 (9.6% vs. 5.8%, P=0.026 and 10.6% vs. 6.6%, P=0.025, respectively). No significant associations were observed with mother and birth-related characteristics. A not straight-forward trend could be offered in regional level.
Conclusions: The present survey offered for the first time a more detailed overview of CMs distribution in Albania. Despite the novelty of information, the issues related to under-reporting continue to be a considerable problem towards correct estimation of CMs rates, prevalence and risk factors.

Introduction

More than 20 million infants are diagnosed with congenital malformations (CMs)every year worldwide of which approximately 8 million experience serious ones (1). Despite efforts to control and possibly to treat these conditions, nearly 3.2 million of them will have to cope with disabilities for the rest of their lives. These children will suffer from various degrees of mental, physical, visual and/ or other types of disabilities. Certain congenital malformations can be lethal. Congenital malformations are the leading cause of infant mortality in the United States. Notwithstanding the significant medical progress in understanding the origin of CMs, in around 50% of cases the respective putative agents remain still unknown. Globally, the most prevalent congenital malformations comprise cardiovascular and neural tube defects as well as the Down syndrome (1,2). Despite being a widespread and global problem affecting virtually all human communities, CMs' impact affects disproportionally the middle and low-income countries. Indeed, over 90% of CMs and CMs related deaths occur in these countries (3). In the Unites States, the prevalence of severe CMs is around 3% and their treatment and care swallow huge amount of money every year (4). The European surveillance of congenital anomalies (EUROCAT) shows that, in Europe, approximately 2% of infants develop a congenital malformation that can affect their ability to survive or function normally. About 2% of these infants will die spontaneously during their growth, and approximately 14% will be terminated by choice.

Among all CMs, those affecting the limbs, heart and spinal cord represent together about 50%. Other frequently observed congenital malformations comprise face, gastro-intestinal tract and sexual organs' defects. About three infants in every 2000 are affected by major chromosomal malformations, such as Down syndrome (5,6). In Europe the prevalence of major CMs was 23.9 per 1,000 live births during 2003-2007 and 80% of them were observed among live born babies. Congenital malformations of heart were the most common non-chromosomal defects (7).The prevalence of congenital malformations from multiple births doubled from 2004 to 2007, an outcome from a registry-based study in fourteen European countries (8).

Numerous recent studies on trends in the prevalence of CMs in Asia, USA, Canada and Europe have shown that pregnancy terminations and prenatal diagnosis rates have steadily increased over the last two decades even though the overall total prevalence of major congenital malformations has been unchanged (9-11). However, different trends were observed by other studies that showed a decline in the prevalence of non-chromosomal CMs and an increase of chromosomal ones (12,13). A higher overall rate of congenital malformations is reported in males and black infants (1,14).

In Albania the rate of congenital malformations was 14.0 per 1000 live births in 2010. In Tirana, the capital of Albania, the rate of congenital malformations for the years 2009 and 2010 was 23.7 and 24.1 per 1000 live births, respectively. In 2010 the most common reported CMs in Albania were those affecting the cardiovascular system, the musculoskeletal system and the mouth with the digestive system (15). Despite some information available (only recently, as shown above), there is no research work referring to the distribution and regional prevalence of CMs in Albania. To our best knowledge, this is the first study assessing the distribution of CMs by different socio-demographic and birth related characteristics in Albania. In this context, the aim of this study was to present an overview of the regional distribution of CMs in Albania in order to provide researchers and policymakers with scientific information for implementing appropriate congenital malformations prevention policies and consider local health care implications.

Methodology

This is a cross-sectional study implemented on national level. The data available for this study were extracted from the congenital malformations surveillance system of the Department of Epidemiology and Health Systems in the National Institute of Public Health. The congenital malformations surveillance system includes all congenital malformations identified and reported among live born babies and also congenital malformations identified among stillborn babies for two years, 2011 and 2012. Fetal deaths prior to 20 weeks of gestational age are not included. The congenital malformations

surveillance system only includes reportable congenital malformations (registered in the ICD9 international statistical classification of diseases). The live births data for two years, 2011 and 2012, were extracted from the Statistical Department of the Ministry of Health. The reporting rate for congenital malformations is different across regions and the Department of Epidemiology and Health Systems estimates that the surveillance captures over 80% of all congenital malformations diagnosed at the time of birth. The establishment of this surveillance system, the way it works and the type of data that it collects has been described elsewhere (16).

The data were analyzed using the SPSS "The Statistical Package for Social Science 15th Edition (SPSS Inc.) software. Analysis was performed as follows: frequencies were run for all variables to identify missing data and outlier values. The congenital malformations data were analyzed by the specific socio-demographic characteristics (region, district, locality of mother's residence, mother's age, mother's education and infant's gender). Also the data analyzes was performed about congenital malformations birth related characteristics (infant's birth weight and vital status).

Results

In Albania, there were 521 and 499 cases of CMs reported for the years 2011 and 2012, respectively. For the year 2011 and 2012 there are 34,297 and 34,974 live births reported.

Approximately 53% of CM cases and 54% of CM cases for the year 2011 and 2012 respectively are rural residents. For the year 2011, 57.2% of CM cases were males and for the year 2012, 63.3% of CM cases were males. In both years approximately 98% of CM cases belong to the white ethnicity. In 2011, the unemployment rate of mothers who gave birth to babies with congenital malformations was 61% in 2012 was 63% (data not shown).

In these two years, the most frequent reported congenital malformations are those affecting the cardiovascular system, gastrointestinal system including oral cavity, musculoskeletal system and those affecting the genital system (Table 1).

Table 1. The distribution of congenital malformations according to the body system affected for the years 2011 and 2012 in Albania

Variable	Years 2011		Year 2012		P-value
	Number	Percentage	Number	Percentage	
Body systems					
The central nervous system	50	9.6	29	5.8	0.026*
(CNS)	16	3.1	19	3.8	0.607
The eye, ear, face and neck	123	23.6	133	26.7	0.279
The cardio-vascular system	6	1.2	6	1.2	1.000
The respiratory system	71	13.6	84	16.8	0.163
The gastro-intestinal system and oral cavity	65	12.5	76	15.2	0.206
The genital system	15	2.9	7	1.4	0.132
The urinary system	98	18.8	84	16.8	0.415
The musculoskeletal system	3	0.6	2	0.4	1.000
The defects of integument	55	10.6	33	6.6	0.025
The chromosomal defects	19	3.6	26	5.2	0.286
The unspecified defects	*521*	*100.0*	*499*	*100.0*	-
Total					

* P-value according to Fischer's exact test.

In 2012 in comparison with the year 2011, there is a slight increase of CMs affecting the cardiovascular system, the gastrointestinal system including the oral cavity and of those affecting the genital system but there is a slight decrease of CMs affecting the musculoskeletal system. All these differences were

not significant (Table 1). In 2012, there is a significant decline of chromosomal defects and of CM cases affecting the central nervous system in comparison with the year 2011.

A description of the most common CMs reported in 2011 and 2012 is presented in Table 2. The most prevalent CM in 2011 was the Down Syndrome (8.8%) whereas in 2012 the most frequent CM was hypospadias with 11.6% of cases.

Table 2. The most common congenital malformations reported in 2011 and 2012 in Albania

Congenital malformations diagnosis in 2011	Year 2011	Year 2012	Congenital malformations diagnosis in 2012
Down syndrome	46 (8.8)	58 (11.6)	Hypospadia
Unspecified anomalies of genital organs	46 (8.8)	32 (6.4)	Polydactyly
Atresia and stenosis of large intestine, rectum and anal canal	25 (4.8)	30 (6.0)	Down syndrome
Ostium secundum type atrial septal defect	24 (4.6)	28 (5.6)	Ventricular septal defect
Polydactyly	22 (4.2)	28 (5.6)	Atresia and stenosis of large intestine, rectum and anal canal
Ventricular septal defect	20 (3.8)	25 (5.0)	Ostium secundum type atrial septal defect
Other specified anomalies of heart	19 (3.6)	23 (4.6)	Unspecified anomalies of heart
Hypospadia	19 (3.6)	18 (3.6)	Unspecified anomalies of genital organs
Congenital hydrocephalus	18 (3.5)	14 (2.8)	Anomalies of ear causing impairment of hearing
Varus deformities of feet	14 (2.7)	14 (2.8)	Varus deformities of feet
Common truncus	12 (2.3)	13 (2.6)	Cheilopalatoschisis
Anomalies of ear causing impairment of hearing	12 (2.3)	10 (2.0)	Transposition of great vessels

The distribution of CM cases by year of study and mother's and birth-related characteristics were not statistically significant (Table 3). The distribution of CM cases by mother's and birth-related characteristics and by year of study is displayed in Table 3. In both years, approximately 45% of mothers who gave birth to babies with congenital malformations had completed only the secondary education. Among all diagnosed cases with congenital malformations, most of their mothers belong to the reproductive age group. In 2012 there is a lower rate in reporting mother's age rather than in 2011. Regarding the distribution of CM by vital status, in both years the vast majority of CMs was diagnosed in live born babies. In 2012, there is an increase in the proportion of babies born with congenital malformations more than 2500 gram and there is a decrease in that of premature babies (less than 37 weeks) born with congenital malformations, compared with the year 2011 (Table 3).

Table 3. Distribution of CM cases by socio-demographic and birth-related characteristics.

Variable	Year 2011		Year 2012		P-value
	Number	Percentage	Number	Percentage	
Mothers education					
No education	2	0.4	7	1.4	
Primary	14	2.7	4	0.8	
Secondary	242	46.4	213	42.7	0.916 *
High school	136	26.1	131	26.3	
University	71	13.6	61	12.2	
Missing	*56*	*10.7*	*83*	*16.6*	
Age-group					
15-19 years	19	3.6	17	3.4	
20-24 years	128	24.6	118	23.6	
25-29 years	171	32.8	143	28.7	
30-34 years	128	24.6	80	16.0	0.246
35-39 years	59	11.3	42	8.4	
40-44 years	15	2.9	14	2.8	
>45 years	1	0.2	3	0.6	
Missing	*0*	*0.0*	*82*	*16.4*	
Vital status					
Fetal death	21	4.0	36	7.2	
Early neonatal death	34	6.5	19	3.8	0.448
Live birth	464	89.1	436	87.4	
Induced abortion	2	0.4	8	1.6	
Birth weight of CMs cases					
<1500 grams	26	5.0	23	4.6	
1500-2500 grams	94	18.0	73	14.6	0.173
>2500 grams	385	73.9	395	79.2	
Missing	*16*	*3.1*	*8*	*1.6*	
Pregnancy age					
<37 weeks	108	20.7	86	17.2	0.175 †
≥37 weeks	389	74.7	399	80.0	
Missing	*24*	*4.6*	*14*	*2.8*	

* P-value according to chi-square test (the missing category was not included for this analysis).

† P-value according to Fischer's exact test.

The distribution of CMs by country's regions in 2011-2012 are presented in Table 4. The highest rate of CMs in 2011 was observed in Gjirokaster region (27.4 per 1000 live births), whereas in 2012 the highest rate was registered in Tirana (26.0 per 1000 live births). The lowest rate of CMs in 2011 and 2012 was registered in Shkoder region (3.2 and 1.7 per 1000 live births, respectively).

Table 4. The rate of congenital malformations per 1000 live births by place of residence for the years 2011 and 2012 in Albania.

Regions	Year 2011			Year 2012		
	Number of live births	Number of CMs	Rate (CM/1000 live births)	Number of live births	Number of CMs	Rate (CM/1000 live births)
Berat	1440	24	16.7	1522	15	9.9
Dibër	1487	7	4.7	1541	5	3.2
Durrës	3741	29	7.8	3468	35	10.1
Elbasan	3269	33	10.1	3412	24	7.0
Fier	3447	43	12.5	3611	38	10.5
Gjirokastër	547	15	27.4	511	10	19.6
Korçë	2238	23	10.3	2185	18	8.2
Kukës	1138	13	11.4	1143	10	8.7
Lezhë	1527	13	8.5	1542	8	5.2
Shkodër	2178	7	3.2	2368	4	1.7
Tiranë	11401	282	24.7	11725	305	26.0
Vlorë	1884	32	17.0	1946	27	13.9
Total	*34297*	*521*	*15.2*	*34974*	*499*	*14.3*

Discussion

This study was performed on secondary data extracted from the congenital malformations surveillance system dataset aimed to identify the rates of congenital malformations by regions and their characteristics. This study shows the overall picture of the distribution of all congenital malformations identified among live born infants, terminated pregnancies and fetal deaths for two years, 2011 and 2012. Congenital malformations were analyzed according to the distribution of socio demographic and birth related characteristics. In 2011 in Albania, the prevalence rate of congenital malformations was 15.2 per 1000 live births and it went slightly down to 14.3 per 1000 live births in 2012. These rates were lower than the rate reported by Global Report on Birth Defects, 2006 (52.9 per 1000 live births) (1). The rates of congenital malformations among regions were higher in 2011 than in 2012, except for Tirana and Durres. The differences in the rates of congenital malformations across regions can be explained somehow by the differences in the reporting of hospitals across regions, differences in diagnosis and use of ultrasound and also to some extent by differences in the population profile (e.g. according to religion, nutrition status) attended in the hospitals and also can partly be explained by the limitation of having only two years for analysis and not the additional years prior to 2011 (15,17). The regional variations in CM rates from 2011 to 2012 require further investigation. In both years the majority of CMs cases were males. In the present study, the higher proportion of CM among males is not mainly due to genital system malformations. In our survey male infants were over represented among all cases and similar findings were observed in a 10 year study conducted in Atlanta, Georgia, where more than half (54.1%) of congenital malformations were observed also among males. (18) In our study, we didn't have information regarding assisted reproduction technology among newborns, stillbirths and terminated pregnancies, but a previous study conducted in Israel, observed a higher prevalence rate of congenital malformations in multiple births than singletons born with assisted reproduction technology (19). The aim of this study was to show the overall picture of congenital malformations and especially their distribution to different socio demographic and birth related characteristics. But there is need for further investigation in order to see if mother's age, mother's education, ethnicity, birth weight and pregnancy weeks are linked with the onset of the congenital malformations (20,21). In our study the percentage of mothers who gave birth to babies

with congenital malformations that completed the university degree was lower than those who completed the secondary education and high school, and a possible explanation is that highly educated women tend to take care of their health and to visit the physician more often than less educated ones (22). This study included only cases diagnosed within 48 hours after delivery. However, age limit at time of diagnosis may have affected our reported prevalence of birth defects and thus our prevalence rates might have been underestimated because of this limitation. Certain regions are very problematic as they do not report about diagnosed CMs. Also, there is lack of reporting from health centers and this could also have contributed towards a further underestimation of CMs prevalence in Albania.

Cardiovascular malformations were the most frequent malformations in 2011 and 2012, although no significant differences were noticed in general.

Enriching the congenital malformations surveillance system by including congenital malformations among still births and terminated pregnancies (more and less than 20 weeks of gestational age), increasing the age at time of diagnosis of congenital malformations, utilizing active case findings surveillance systems (23) and including both inpatient and outpatient facilities in the surveillance (24), would contribute to the better completion of the database thus leaving little possibility for cases to be routinely missed. Congenital malformations are a significant public health problem since they are relatively common, they lead to disabilities and are a major reason for hospitalization in infancy and childhood (25). Although the data are still considerably incomplete, the variation of the regional disparity in congenital malformations should be of concern, and suggests the need for appropriate public health and medical interventions.

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