

RESEARCH ARTICLE

Information on congenital malformations obtained from hospital information flow data

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Abstract: Congenital malformations represent a relevant cause of fetal, neonatal and infant mortality, and morbidity. In consideration of the few resources available, various studies have been conducted to evaluate the different methods of data collection using different information sources. This study aims to improve knowledge on congenital malformations in the Calabria Region in 2017 through the development of a formal procedure to connect and integrate the current SDO and CeDAP flows with the regional register of congenital malformations (RRMC) to enhance different sources information and create a single database. Hospitalization flow of the Calabria Regional Health System have been selected for all those born in the Region in 2017 and their hospitalizations within the first year of life. There were 1,307 cases with at least one ICD-9-CM code in the range 740-759. Cases deemed “minor” were excluded and the “potentially malformed” case search procedures were performed. Following the application of the assessment criteria, 449 cases were considered “validated”, 81 cases with malformation in the birth flow and 215 in the Malformation registrer were detected. The various flows contributed to the construction of the integrated regional malformed registre, the integration of the RRMC flow with the hospital discharge cards (SDO) and with the Certificate of Childbirth Assistance (CedAP), has allowed to obtain complete coverage of the birth points of the Calabria Region, for a total of 16,282 births supervised. The birth rate recorded among the supervised births was 4.3 per 1,000. The separate use of information from the registers and from the different health flows, which normally do not communicate with each other, has however shown limits. The records contain detailed descriptions of the malformative condition, but often lack information on surgical interventions or further instrumental clinical investigations. Instead, this information is present in the other streams. On the other hand, the importance of diagnosis and coding of correct malformations, often deficient in SDO and CeDAP, have important implications for research and evaluation purposes.

Keywords: congenital malformation, hospital admissions, information flows

1 Introduction

Congenital anomalies are errors of morphogenesis, determined in part by genetic factors^[1]. The exposure of the mother and fetus to known teratogenic factors (infectious, physical, chemical, maternal pathologies) causes about 9-10% of the defects, while 65% have an unknown etiology. Congenital malformations^[2], considered individually, are usually rare, but their overall prevalence at birth (diagnosed within the first week of life) is around

2-3%. Their frequency varies from one case out of about 150-200 births for congenital heart disease considered overall to one case every 11,000-12,000 births for gastroschisis. Taken together, they are numerically important, affecting about 5-6% of children within the year of life.

In the period 2004-2007, according to the data of the European Network for the surveillance of Eurocat congenital malformations^[2] (referring to the Campania^[3], Emilia Romagna and Tuscany Registers), 7,894 cases were recorded with congenital malformations on 512,867 supervised births (live births + dead births), with a total prevalence of 153.62 per 10,000.

The congenital defects detected refer to 5,628 live births, 2,217 induced abortions and 49 fetal deaths (fetal deaths include both dead births and fetal deaths subsequent to the 20th gestational week).

Overall, chromosomal anomalies (1,327) represent around 17% of the overall series, with a total prevalence of 25.82 per 10,000 inhabitants. The most frequent chromosomal pathology is Down syndrome (total prevalence

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16.31).

From the analysis of the aggregated cases, according to the 14 subgroups of congenital defects defined by Eurocat^[3], it appears that the malformations of the cardiovascular system are the most frequent (total prevalence 44.95), followed by chromosomal anomalies (total prevalence 25.82), from the limb defects (total prevalence 22.09), urinary tract (total prevalence 18.35), nervous system (total prevalence 16.81) and genitals (total prevalence 15.76). All other groups have total headings of less than 10 per 10,000.

Although serious deficiencies in the etiopathogenesis of multifactorial congenital malformations persist^[2], a strategy for primary prevention uses some evidence-based points:

- (1) the promotion of folic acid;
- (2) the promotion of antirubeolic vaccination and the prevention of toxoplasmosis in pregnancy;
- (3) the correct use of drugs in the fertile woman, with particular regard to anti-epileptic, anticancer and endocrine therapies, and the knowledge, by the operators of the NHS, of substitute drugs;
- (4) the promotion of healthy and responsible food and lifestyle styles, with particular attention to the prevention of cigarette smoking, excessive alcohol consumption, diabetes and obesity;
- (5) protection of working conditions, in particular in the presence of exposure to particular toxic substances (eg work in intensive agriculture).

Congenital malformations represent an important cause of fetal, neonatal and infant mortality, and morbidity^[2]. In recent years, the evidence has grown that congenital disorders can be partially prevented or reduced if properly treated.

Surveillance systems based on congenital malformation registries (RMC) have existed for some time^[4], the data they provide are increasingly used for the evaluation of health services (laboratory diagnostic tests and ultrasound diagnostics in pregnancy) and for surveillance and prevention (vaccinations or administration of folic acid)^[4].

The monitoring over time and space of congenital malformations represents an important basis for identifying etiological factors^[2].

In consideration of the ever-decreasing resources available, various studies have been conducted to evaluate the different methods of data collection using different information sources. The integrated use between the various current information flows, such as birth attendance certificates (CeDAP), hospital discharge cards and malformation registers is of growing interest. Current flows are however different from each other for purposes and char-

acteristics, for accuracy and reliability. The evaluation of the quality of the collected data and their integration is a very delicate process.

The registers of congenital malformations represent in theory the best source, but there is a need to evaluate the quantitative relevance, relating to the inclusion of all cases, all variables and qualitative relevance, which concerns the ability to identify all the different malformative groups^[2].

This study aims to improve knowledge on congenital malformations in the Calabria Region in 2017 through the development of a formal procedure to connect and integrate the current SDO and CeDAP flows with the regional register of congenital malformations (RRMC) to enhance different sources information and create a single database^[5-8].

2 Materials and methods

2.1 Hospital discharge form (SDO)

The SDO is the tool for collecting information regarding each patient discharged from public and private hospitalization institutions throughout the national territory.

The diseases in the SDO are classified with the ICD-9-CM system, the regional SDO flow will be used to identify cases with congenital birth defects reported in hospitalizations within the first year of life.

2.2 Childbirth Assistance Certificate

Childbirth Assistance Certificate (CeDAP) provides health, epidemiological and socio-demographic information through the detection of birth events, birth-mortality ratio and births affected by malformations.

The certificate is drawn up no later than the tenth day after birth by the midwife or the doctor who assisted the birth or by the doctor in charge of the operating unit in which the birth occurred.

2.3 The Registry of Congenital Malformations in the Calabria Region (RRMC)

The Registry started the business in 2017, with Decree no. 3713 of 1/4/2014 "Regional Register of Congenital Malformations (RRMC) - integration of information sources", the new surveillance system was established^[4]. The notification network consisting of all birth points, TINs, neonatology, pediatrics and pediatric surgery departments has been reorganized. The survey is based on an IT platform via the web. The main strengths of the new system are the detection of cases diagnosed from birth to the first year of life, the possibility of integration with the regional information system (improvement of coverage)

and data entry via web mask (improvement of timeliness). The Registry participates in the coordination of the Italian registers of congenital malformations established at the Istituto Superiore di Sanità in order to share the experiences of various registers and propose common lines of research.

This study will be carried out by building an algorithm that performs a search and identification of malformed cases starting from the analysis of the individual SDOs. Subsequently, the algorithm verifies the presence of the cases identified also in the CeDAP and RRMC Register flows. The first step is the selection in the SDO flow of the codes included in the specific range of malformative pathologies (ICD-9-CM 740-759); codes outside this range will not be considered. Within the specified range, pathologies deemed minor in accordance with Eurocat indications will subsequently be excluded^[3].

The result of this first step will be the establishment of a set of cases defined as “potentially malformed”.

The next step will be to divide the “potentially malformed” cases into two categories: validated and to be evaluated.

The cases are “validated” according to 5 assessment criteria used in hierarchical order; the validation procedure stops when a criterion occurs in this order:

(1) in the SDO there is a specific surgical code for that malformation;

(2) the malformation falls within a predefined list of conditions (situs inversus, transposition of the large vessels, chromosomal anomalies, anencephaly, spina bifida, tetralogy of Fallot);

(3) the pathology is also confirmed in the CeDAP flow, which has the sole purpose here of confirming the presence of the malformation;

(4) the same pathology is repeated in several SDOs. This condition is not necessarily an evidence of correct diagnosis, but represents a high probability of the presence of the malformation. The case is unique, even if the malformation is repeated;

(5) the simultaneous presence in the SDO of multiple malformations in different apparatuses occurs (poly-deformed case);

The cases “to be evaluated” are those that remain after the application of the assessment criteria and that require viewing of the medical record to be validated. To reduce the number, exclusion filters were used, based on the MONITER experience and for selected malformations. The filters allow to exclude cases in which the malformation being selected is present only in the SDO at birth. The filters used are:

(F1): gestation duration of less than 37 weeks (reference is made to the ICD-9-CM codes in the SDO 76521-

76528; for example, exclusion of the premature case with patent ductus arteriosus, ICD-9-CM 7470);

(F2): duration of hospitalization in days (for example, exclusion of unspecified cardiac pathologies, ICD-9-CM 7469, when the number of days of hospitalization at birth is less than the value established as an indicator of a possible clinical problem);

(F3): isolated malformation (for example, exclusion of macroglossia). The malformation is considered isolated when no other ICD-9-CM malformation codes are indicated.

The last procedure will be to compare the cases selected by the algorithm with the CeDAP flow and with the Register of congenital malformations. The procedure will allow to verify if the cases validated, excluded and to be evaluated clinically are possibly present in the CeDAP and RRMC flows, each validated case not present in the Register will complete the RRMC database.

The variables under study are:

(1) Date of birth dd / mm / yyyy;

(2) Structure of birth structure code;

(3) Gender m / f / undetermined;

(4) Single or multiple birth s / m;

(5) Type of event NV / NM / IVG;

(6) Duration of gestation n. weeks;

(7) Birth weight grams;

(8) Municipality of mother residence ISTAT code (6 digits);

(9) Maternal age years;

(10) Malformation (GR) group;

(11) Malformation (MC) code ICD9;

(12) Nati number.

3 Results

The SDOs of the Calabria Regional Health System have been selected for all those born in the Region in 2017 and their hospitalizations within the first year of life. There were 1,307 cases with at least one ICD-9-CM code in the range 740-759. Cases deemed “minor” were excluded and the “potentially malformed” case search procedures were performed.

Following the application of the assessment criteria, 449 cases were considered “validated”. 81 Cases with malformation in the CeDAP flow and 215 in the RRMC registry were detected. The various flows contributed to the construction of the integrated regional malformed register (Figure 1).

The integration of the RRMC flow with the hospital discharge cards (SDO) and with the Certificate of Child-birth Assistance (CedAP), has allowed to obtain complete coverage of the birth points of the Calabria Region, for a

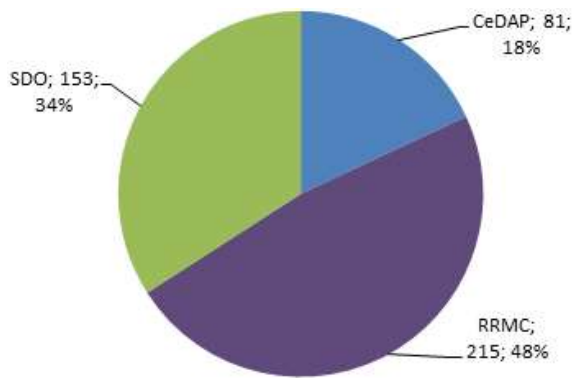


Figure 1 Contribution of flows

total of 16,282 births supervised.

The birth rate recorded among the supervised births was 4.3 per 1,000. (Table 1)

Table 1 Denominators 2017 (source CedAP)

Denominators (2017)	N
Live born	16,211
Born dead	71 (4.3‰)
Total born	16282

There were 449 cases with congenital malformations recorded in the various streams (250 males and 199 females) with a prevalence at birth of 27.6 per 1,000, if we also consider the terminations of pregnancy (n.98) the prevalence is 33, 6 per 1,000. The analysis by province of birth shows a maximum value (82.8 per 1,000) in Crotona and a minimum value (16.4 per 1,000) in Reggio Calabria.

Figure 2 presents the provincial prevalences in 2017 of cases born malformed with relative confidence limits (95%) with respect to the regional average prevalence of 27.6‰ (IC95% 25.1-30.1) in the same period.

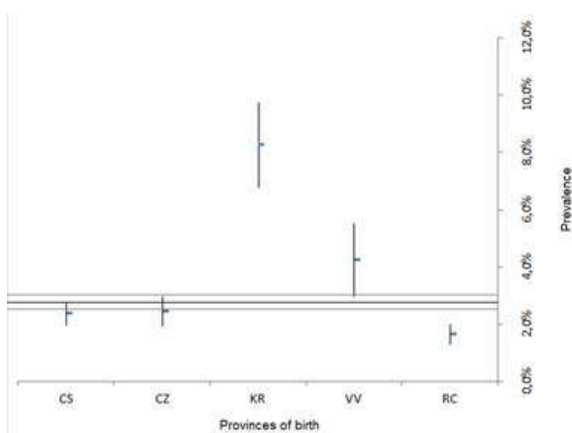


Figure 2 Comparison of prevalences in the provinces of birth and regional average

Figure 3 shows the prevalences in the provinces of mother’s residence with relative confidence limits (95%) with respect to the regional average prevalence.

The prevalence ranges from 19.3‰ (IC95% 15.3-23.2) for cases in the province of Reggio Calabria, to 75.0‰ (IC95% 61.8-88.1) in the province of Crotona.

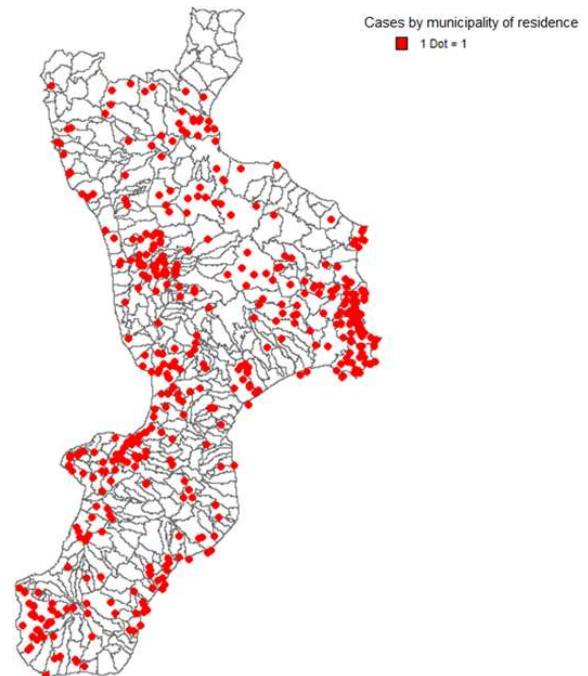


Figure 3 Identified cases, distribution by diagnosis (ICD9)

4 Conclusions

The need to have reliable data on congenital anomalies has led to an increasing use of the registers of congenital malformations for public health purposes. At the same time, the need emerged for the assessment of the completeness and reliability of the data collected and the need for integration of the various sources that collect health data at birth and in subsequent hospitalizations.

The results obtained showed that there is no perfect data collection model, but that one can and must take advantage of the integration of information collected from multiple sources. The percentage of coverage of the flows differs according to the birth points of the Region, this figure varies according to the health organization (personnel involved, changes in assistance, etc.).

There were 449 cases with congenital malformations recorded by the integration of the different flows, the prevalence in the Calabria region (27.6‰) is slightly higher than in the Emilia Romagna region (27.0‰) and Tuscany (26.0‰).

In the CeDAP flow, only 81 cases of infants born

with malformation were found out of 215 detected in the RRMC register, whose activity begins precisely in the year 2017.

By comparing the confidence intervals at 95% of the prevalences (per 1,000 births) in the provinces of residence of the mother with the regional average prevalence of 27.6‰ (95% CI 25.1-30.1), significantly higher than the average regional value in the province of Vibo Valentia (44.3; IC95% 32.8-55.7) and in the province of Crotona (75.0; IC95% 61.8-88.1).

On the other hand, the prevalence rates of mother cases residing in the provinces of Cosenza (23.9; IC95% 19.7-28.1), Catanzaro (20.1; IC95% 15.0-25.3) and Reggio Calabria (19.3; IC95% 15.3-23.2).

The separate use of information from the registers and from the different health flows, which normally do not communicate with each other, has however shown limits. The records contain detailed descriptions of the malformative condition, but often lack information on surgical interventions or further instrumental clinical investigations. Instead, this information is present in the other streams. On the other hand, the importance of diagnosis and coding of correct malformations, often deficient in SDOs and CeDAPs, have important implications for research and evaluation purposes.

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