



# National congenital anomaly registers in the world: historical and operational aspects

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## Abstract

**Objective:** To identify registers of congenital anomalies with national coverage currently available around the world, highlighting their main historical and operational characteristics. **Methods:** This was a documentary study by means of a Medline database search (via PubMed) and searches involving reports, official documents and websites. Studies reporting at least one national registry were included. **Results:** 40 registers of national congenital anomalies were identified in 39 different countries. All registers included in the study were concentrated in upper-middle or high-income countries located in Europe. Most of the registers were population-based, compulsory notification and with a time limit for notification of up to 1 year of age. The Brazilian register showed the highest annual coverage. **Conclusion:** The registers analyzed showed different characteristics, related to the reality of each country. The results presented provide support for the theme of congenital anomalies surveillance, especially in places where such activity is intended to be implemented.

**Keywords:** Congenital anomalies; Birth Declaration; Epidemiological Surveillance; Review; International Cooperation; Health Services; Disease Records.

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## Introduction

Congenital anomalies are structural or functional changes that occur during intrauterine life.<sup>1</sup> In addition to being important causes of perinatal and neonatal deaths, they can contribute to long-term disability, which may have significant impacts on individuals, their families, healthcare systems, and societies.<sup>1,2</sup> According to the World Health Organization, about 295,000 babies die within the first four weeks of birth due to congenital anomalies every year.<sup>1</sup>

Systematic and continuous collection of information on cases of congenital anomalies in a well-defined population characterizes a register.<sup>3</sup> In the world, different registers of congenital anomalies were developed in the second half of the twentieth century, after the episode known as "the thalidomide tragedy", when more than 10 thousand children in 46 countries were born with severe congenital anomalies due to the use of this drug during pregnancy.<sup>4</sup> Registers of congenital anomalies are useful for monitoring temporal or geographic differences in the frequencies of these disorders and assisting in the identification of risk factors. They can also contribute in delineating vulnerable populations, plan and evaluate health care offers, among others.<sup>3,5</sup>

Understanding the epidemiological profile of congenital anomalies, by means of information systems and monitoring with national coverage, provides countries with an opportunity to perceive their impact on the population and health systems. This knowledge also produces useful information that can promote prevention measures and adequate health care aimed at the specific scenario of each location.<sup>6-8</sup>

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Registers of congenital anomalies can cover entire countries or a representative sample of the national

population (national registers), or specific locations in a country (local or subnational registers). In addition, they can cover all births of a given geographical area (population-based) or cover only births in a single hospital or selected hospitals (hospital-based).<sup>9,10</sup> However, there are no data in the scientific literature on the global and updated perspective of national registers of congenital anomalies and their peculiarities.

The objective of this study was to identify registers of congenital anomalies with national coverage currently available around the world, highlighting their main historical and operational characteristics.

## Methods

This was a documentary study, which its methodological strategy was detailed in a narrative review on international congenital anomaly surveillance collaboration networks, previously published.<sup>6</sup> In general, to map and document the registers of congenital anomalies with national coverage currently available around the world, we searched for bibliographic references indexed in Medical Literature Analysis and Retrieval System Online (Medline)/PubMed database (made available by the National Library of Medicine of the United States) on January 10, 2020, through the search key specified in Figure 1. In addition to this search, relevant information related to this research theme was obtained from reports, official documents and websites made available by networks, records and institutions that work with the surveillance of congenital anomalies.

The main methodological steps, as well as the information extracted from each of the studies or documents that were found, are detailed in Figure 1. The information extracted was analyzed by two reviewers (Cardoso-dos-Santos AC and Alves SRM), in an independent manner. Information on the gross national income per capita of each participating country was obtained from the World Economic Situation and Prospects report, produced by the United Nations.<sup>7</sup>

## Results

A total of 40 registers of congenital anomalies with national coverage were identified in 39 countries. Their main characteristics are presented in Box 1. With the exception of Africa, all continents have submitted

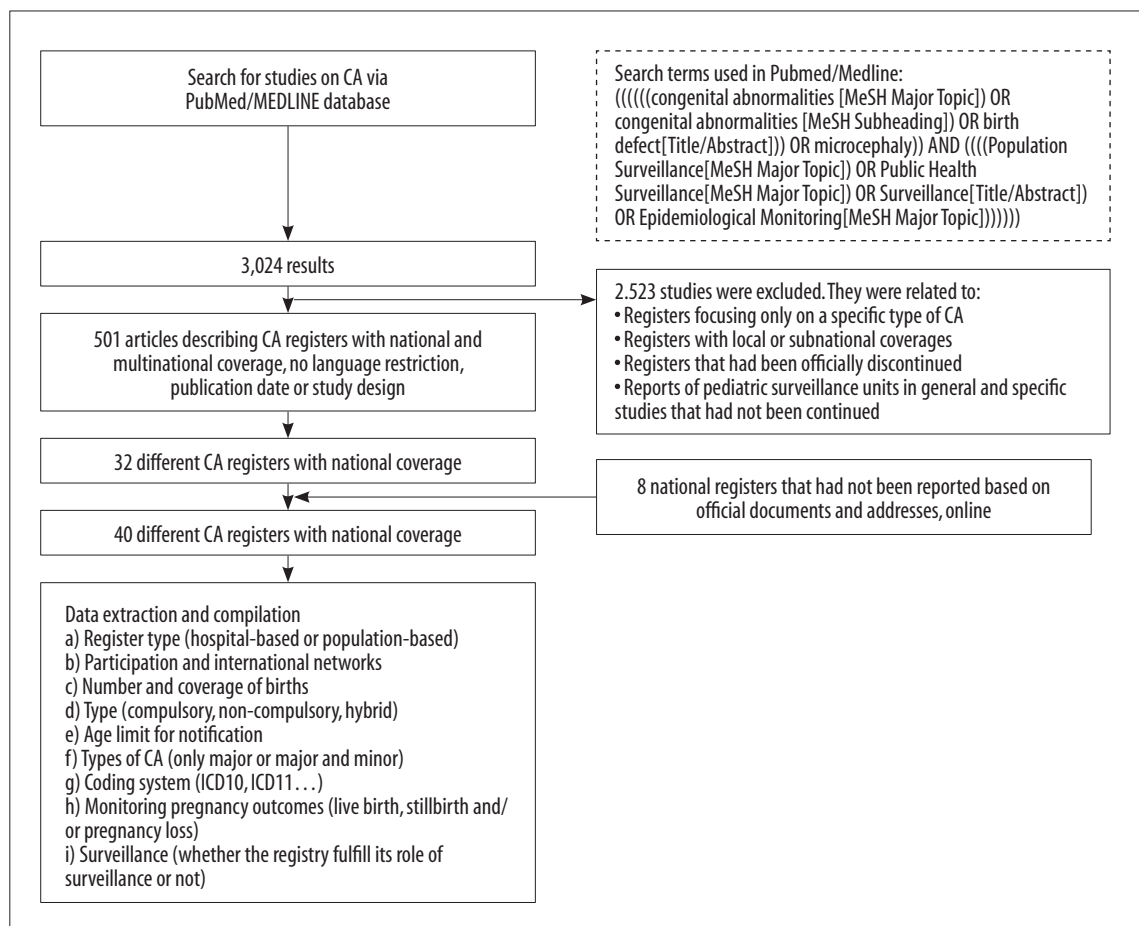
at least one national register of congenital anomalies. Most of these countries were located on the European continent, all of them were in the upper-middle (12) and high income (28) categories as shown in Figure 2. There were 26 population-based and 12 hospital-based registers; no registers were found for Chile and Panama.

The Hungarian Congenital Abnormality Registry was the oldest registry. It was established in 1962, while the most recent was the Scottish, implemented in 2018. The majority of the national registries (29/40) were included in at least one international congenital anomaly surveillance collaboration network. The Brazilian registry was the one with the highest number of births annually - around 3 million - and covered 100% of births in the country. In total, 19 registers had more than 98% coverage. Almost all of them were population-based: two of them, Cuba and

the Dominican Republic, in Central America, were hospital-based registries.

Fourteen national registers were compulsory, of these, 12 were population-based notifications; and of the 11 voluntary registers, only five were population-based notifications. The chronological age limit to notify an individual with congenital anomaly was also very different among the registers, ranging from hospital discharge (6) to one month (5) or greater than or equal to 1 year of age (23).

A total of 16 national registers reported only major congenital anomalies, and 14 reported major and minor congenital anomalies. The International Statistical Classification of Diseases and Related Health Problems (ICD, 9<sup>th</sup> and 10<sup>th</sup> revisions) was the main coding system used (32); however, ten national registers modified the ICD (especially the British Pediatric Association Classification of Diseases).



**Figure 1 – Flowchart of the main methodological steps for the identification and selection of registers of congenital anomalies with national coverage currently available around the world**

Box 1 – Main characteristics of registers of congenital anomalies with national coverage around the world, January 10, 2020

Country	Income	Registry	Year created	Type of registry	Network	Number of births (thousands) annually (% of coverage)	Compulsory	Time limit	Type of congenital anomaly	Coding	Pregnancy outcomes	Surveillance	Information source
Saudi Arabia	High	Medical Service Department- Birth Defect Registry (MSD-BDR)	2010	Hospital-based	ICBDSR	–	Yes	2 years	Major	ICD-BPA	–	–	<a href="https://bit.ly/2XHWSNm">https://bit.ly/2XHWSNm</a>
Argentina	Upper-middle	National Registry of Congenital Anomalies of Argentina (RENAC)	2009	Hospital-based	ICBDSR, ReLAMC	300 (40%)	No	Hospital discharge	Major	ICD-BPA	Live birth and Stillborn	Yes	<a href="https://bit.ly/3bCBph4">https://bit.ly/3bCBph4</a> <a href="https://dx.doi.org/10.5546/aap.2013">https://dx.doi.org/10.5546/aap.2013</a>
Australia	High	Australian Congenital Anomalies Monitoring System (ACAMS)	1981	Population-based	–	–	Hybrid	Vary among different collections	Major	ICD-BPA and AM	Live birth and Stillborn	Yes	<a href="https://bit.ly/2LqbnT0">https://bit.ly/2LqbnT0</a> <a href="https://bit.ly/2Ke5ImZ">https://bit.ly/2Ke5ImZ</a>
Brazil	Upper-middle	Sistema de Informações sobre Nascidos Vivos (Sinasc)	1999	Population-based	ReLAMC	3.000 (100%)	Yes	Hospital discharge	Major	ICD	Live birth	No	<a href="https://bit.ly/3nld7VA">https://bit.ly/3nld7VA</a>
Canada	High	Canadian Congenital Anomalies Surveillance System (CCASS)	1966	Population-based	ICBDSR	330 (98%)	–	1 month	Major	ICD-CA	Live birth and Stillborn	Yes	<a href="https://bit.ly/3bEVm6U">https://bit.ly/3bEVm6U</a> <a href="https://bit.ly/2LMLUDV">https://bit.ly/2LMLUDV</a>
Chile	High	Registro Nacional de Anomalías Congénitas de Chile (RENACH)	2016	–	ReLAMC	220	–	–	–	–	–	–	<a href="https://bit.ly/38Gs1Hq">https://bit.ly/38Gs1Hq</a> <a href="https://bit.ly/3qyz7iq">https://bit.ly/3qyz7iq</a>
China	Upper-middle	Birth Defects Surveillance System for the Collaborative Project-China (BDSS-China)	1992	Population-based	–	260	–	42 days	Major	ICD	Live birth and Stillborn	Yes	<a href="https://doi.org/10.1002/ajmg.c.31690">https://doi.org/10.1002/ajmg.c.31690</a> <a href="https://doi.org/10.1007/s12519-011-0326-0">https://doi.org/10.1007/s12519-011-0326-0</a>
China	Upper-middle	Chinese Birth Defects Monitoring Network (CBDMN)	1988	Hospital-based	–	1.380 (8.51%)	No	7 days	Major	ICD	Live birth, Stillborn and Termination of pregnancy	Yes	<a href="https://doi.org/10.1007/s12519-011-0326-0">https://doi.org/10.1007/s12519-011-0326-0</a> <a href="https://doi.org/10.1046/j.1365-3016.2003.00478.x">https://doi.org/10.1046/j.1365-3016.2003.00478.x</a>
Colombia	Upper-middle	Vigilancia de los Defectos Congénitos	2010	Population-based	ReLAMC	661 (100%)	–	1 year	Major	ICD	–	–	<a href="https://doi.org/10.26633/RPSP.2019.44">https://doi.org/10.26633/RPSP.2019.44</a>

a) NBDPN is a volunteer-based organization in the United States; (b) the information is related to the United Kingdom; (c) covers all live births delivered or treated at a public hospital in New Zealand; (d) Down syndrome, neural tube defects, cleft lip and palate, limb abnormalities and Duchenne muscular dystrophy. Legend: BINOCCAR: British and Irish Network of Congenital Anomaly Researchers; BPA: British Paediatric Association Classification of Diseases; CDC: Centers for Disease Control and Prevention of the United States; ICD: International Classification of Diseases and Related Health Problems; EUROCAT: European surveillance of congenital anomalies; ICBDSR: International Clearinghouse for Birth Defects, Surveillance and Research; ReLAMC: Red Latinoamericana de Malformaciones Congénitas; SEAR-NBBD: South-East Asia Region New-born and Birth Defects.

To be continued

**Box 1 – Main characteristics of registers of congenital anomalies with national coverage around the world, January 10, 2020**

Country	Income	Registry	Year created	Type of registry	Network	Number of births (thousands) annually (% of coverage)	Compulsory	Time limit	Type of congenital anomaly	Coding	Pregnancy outcomes	Surveillance	Information source
Costa Rica	Upper-middle	Costa Rican Birth Defects Register Centre (CREC)	1985	Population-based	ICBDSR, ReLAMC	70 (100%)	Yes	1 year	Major and minor	ICD-BPA	Live Birth and Stillborn	Yes	<a href="https://bit.ly/3bPXdWLE">https://bit.ly/3bPXdWLE</a> <a href="https://doi.org/10.26633/RPSP.2019.44">https://doi.org/10.26633/RPSP.2019.44</a> <a href="https://doi.org/10.1007/s10995-014-1542-8">https://doi.org/10.1007/s10995-014-1542-8</a>
Cuba	Upper-middle	Cuban Register of Congenital Malformation (RECUMAC)	1985	Hospital-based	ICBDSR, ReLAMC	120 (100%)	No	Hospital discharge	Major and minor	ICD-BPA	Live birth, Stillborn and Termination of pregnancy	–	<a href="https://doi.org/10.26633/RPSP.2019.44">https://doi.org/10.26633/RPSP.2019.44</a> <a href="https://doi.org/10.26633/RPSP.2017.174">https://doi.org/10.26633/RPSP.2017.174</a>
Denmark	High	Danish Medical Birth Registry	1973	Population-based	–	–	–	1 year	–	ICD	Live birth, Stillborn and Termination of pregnancy	–	<a href="https://bit.ly/3quYJ4D">https://bit.ly/3quYJ4D</a> <a href="https://doi.org/10.1080/14034940210134194">https://doi.org/10.1080/14034940210134194</a> <a href="https://doi.org/10.3402/ijch.v68i5.17376">https://doi.org/10.3402/ijch.v68i5.17376</a>
The United Arab Emirates	High	National Congenital Anomalies Register	1999	Population-based	–	63 (100%)	–	1 year	Major	ICD with modification	Live Birth and Stillborn	Yes	<a href="https://bit.ly/39Lkphk">https://bit.ly/39Lkphk</a>
Scotland	High <sup>b</sup>	The Scottish Linked Routine Data Congenital Anomaly Register	2018	Population-based	EUROCAT	50-55 (100%)	Hybrid	1 year	Major	ICD	Live birth, Stillborn and Termination of pregnancy	–	<a href="https://bit.ly/3oUJOY1">https://bit.ly/3oUJOY1</a> <a href="https://bit.ly/39FY05">https://bit.ly/39FY05</a>
Slovakia	High	Teratologic Information Centre Slovak Medical University	1964	Population-based	ICBDSR	55 (100%)	Yes	Hospital-based	–	–	Live birth, Stillborn and Termination of pregnancy	–	<a href="https://bit.ly/2XHWSNm">https://bit.ly/2XHWSNm</a> <a href="https://dx.doi.org/10.1136/bmjopen-2018-028139">https://dx.doi.org/10.1136/bmjopen-2018-028139</a>
Spain	High	Spanish Collaborative Study of Congenital Malformations (ECEMC)	1976	Hospital-based	ICBDSR, EUROCAT	90 (20%)	No	3 days	Major and minor	ICD	Live birth and Stillborn (Termination of pregnancy in some hospitals)	Yes	<a href="https://bit.ly/3imztz6">https://bit.ly/3imztz6</a> <a href="https://doi.org/10.2165/00002018-200831060-00008">https://doi.org/10.2165/00002018-200831060-00008</a> <a href="https://doi.org/10.1016/j.jpedsurg.2008.07.002">https://doi.org/10.1016/j.jpedsurg.2008.07.002</a>
The United States	High	National Birth Defects Prevention Network (NBDPN)	1997	Population-based	–	–	No	–	Major	ICD e CDC/BPA	Live birth, Stillborn and Termination of pregnancy	Yes	<a href="https://bit.ly/3sskMG">https://bit.ly/3sskMG</a> <a href="https://doi.org/10.1002/bdr2.1607">https://doi.org/10.1002/bdr2.1607</a> <a href="https://doi.org/10.1155/2014/212874">https://doi.org/10.1155/2014/212874</a>

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Box 1 – Main characteristics of registers of congenital anomalies with national coverage around the world, January 10, 2020

Country	Income	Registry	Year created	Type of registry	Network	Number of births (thousands) annually (% of coverage)	Compulsory	Time limit	Type of congenital anomaly	Coding	Pregnancy outcomes	Surveillance	Information source
Finland	Alta	Register of Congenital Malformations	1963	Population-based	ICBDSR, EUROCAT	60 (100%)	Yes	1 year	Major	ICD	Live birth, Stillborn and Termination of pregnancy	Yes	<a href="https://bit.ly/3nUTRUH">https://bit.ly/3nUTRUH</a> <a href="https://bit.ly/2XHWSNm">https://bit.ly/2XHWSNm</a> <a href="https://doi.org/10.3402/ijch.v68i5.17376">https://doi.org/10.3402/ijch.v68i5.17376</a> <a href="https://doi.org/10.1093/ije/11.3.239">https://doi.org/10.1093/ije/11.3.239</a>
Guatemala	Upper-middle	Protocolo de Vigilancia de Anomalías Congénitas	2017	Hospital-based	–	155 (40%)	–	1 month	Major	ICD	–	Yes	<a href="https://doi.org/10.26633/RPSP.2019.44">https://doi.org/10.26633/RPSP.2019.44</a>
Hungary	High	Hungarian Congenital Abnormality Registry (HCAR)	1962	Population-based	ICBDSR, EUROCAT	100 (100%)	Yes	1 year	Major and minor	ICD with modification	Live birth, Stillborn and Termination of Pregnancy	–	<a href="https://bit.ly/2XHWSNm">https://bit.ly/2XHWSNm</a> <a href="https://doi.org/10.1111/cga.12025">https://doi.org/10.1111/cga.12025</a>
England	High <sup>a</sup>	National Congenital Anomaly and Rare Disease Registration Service (NCARDRS)	2015	Population-based	BINOCAR, ICBDSR	610 (100%)	No	–	Major and minor	ICD	Live birth, Stillborn and Termination of pregnancy	–	<a href="https://bit.ly/38UJEB">https://bit.ly/38UJEB</a> <a href="https://dx.doi.org/10.1136/archdischild-2017-312833">https://dx.doi.org/10.1136/archdischild-2017-312833</a>
Iceland	High	Icelandic Register of Births	–	Base populacional	–	–	–	–	–	ICD	Live birth and Stillborn	–	<a href="https://doi.org/10.3402/ijch.v68i5.17376">https://doi.org/10.3402/ijch.v68i5.17376</a>
Italy	High	Italian Multicenter Register for Congenital Malformations (IPIMC)	1977	Hospital-based	–	116 (20%)	No	5 days	Major	–	Live birth and Stillborn	Yes	<a href="http://doi.wiley.com/10.1002/ajmg.1320460425">http://doi.wiley.com/10.1002/ajmg.1320460425</a> <a href="https://doi.org/10.1007/BF00162315">https://doi.org/10.1007/BF00162315</a>
Japan	High	Japan Association of Obstetricians and Gynaecologists (JAOG)	1972	Hospital-based	ICBDSR	100 (9%)	–	7 days	–	–	Live birth and Stillborn	–	<a href="https://bit.ly/2XTwSYV">https://bit.ly/2XTwSYV</a> <a href="https://bit.ly/2XHWSNm">https://bit.ly/2XHWSNm</a>
Latvia	High	Register of Patients Suffering from Certain Diseases	1987	Population-based	EUROCAT	19.2 (100%)	Yes	18 years	–	–	Live birth, Stillborn and Termination of pregnancy	–	<a href="https://bit.ly/3oUJOY1">https://bit.ly/3oUJOY1</a> <a href="https://doi.org/10.1186/s13023-014-0147-z">https://doi.org/10.1186/s13023-014-0147-z</a>

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Continuation

**Box 1 – Main characteristics of registers of congenital anomalies with national coverage around the world, January 10, 2020**

Country	Income	Registry	Year created	Type of registry	Network	Number of births (thousands annually (% of coverage)	Compulsory	Time limit	Type of congenital anomaly	Coding	Pregnancy outcomes	Surveillance	Information source
Malta	High	Malta Congenital Anomalies Register (MCAR)	1985	Population-based	ICBDSR, EUROCAT	4 (100%)	No	1 year	Major and minor	ICD	Live birth and Stillborn	-	<a href="https://bit.ly/2XHWSNm">https://bit.ly/2XHWSNm</a> <a href="https://bit.ly/3oUJOY1">https://bit.ly/3oUJOY1</a>
Mexico	Upper-middle	Registro y Vigilancia Epidemiológica de Malformaciones Congénitas (RYVEMCE)	1978	Hospital-based	ICBDSR	62 (3.5%)	No	Hospital discharge	Major and minor	-	Live birth and Stillborn	Yes	<a href="https://doi.org/10.26633/RPSP2019.44">https://doi.org/10.26633/RPSP2019.44</a> <a href="https://bit.ly/2XHWSNm">https://bit.ly/2XHWSNm</a> <a href="https://doi.org/10.1016/j.bmhimx.2017.02.003">https://doi.org/10.1016/j.bmhimx.2017.02.003</a> <a href="https://bit.ly/3qzZ7f6">https://bit.ly/3qzZ7f6</a>
Norway	High	Medical Birth Registry of Norway (MBRN)	1967	Population-based	ICBDSR, EUROCAT	60 (100%)	Yes	1 year	-	ICD-BPA	Live birth, Stillborn and Termination of pregnancy	Yes	<a href="https://bit.ly/350TzVM">https://bit.ly/350TzVM</a> <a href="https://doi.wiley.com/10.1034/j.1600-0412.2000.079006435.x">https://doi.wiley.com/10.1034/j.1600-0412.2000.079006435.x</a> <a href="https://doi.org/10.1111/dmnc.13552">https://doi.org/10.1111/dmnc.13552</a>
New Zealand	High	New Zealand Birth Defects Registry (NZBDR)	1975	Population-based	ICBDSR	58 (100%) <sup>c</sup>	Yes	No time limit	Major and minor	-	Live birth, Stillborn and Termination of pregnancy	-	<a href="https://bit.ly/2XRvHk4">https://bit.ly/2XRvHk4</a> <a href="https://bit.ly/2XHWSNm">https://bit.ly/2XHWSNm</a>
Wales	High <sup>b</sup>	Congenital Anomaly Register & Information Service for Wales (CARIS)	1998	Population-based	BINOCAR, ICBDSR, EUROCAT	35 (100%)	No	1 year	Major and minor	ICD	Live birth, Stillborn and Termination of pregnancy	-	<a href="https://bit.ly/3nUENXh">https://bit.ly/3nUENXh</a> <a href="https://bit.ly/2XHWSNm">https://bit.ly/2XHWSNm</a> <a href="https://doi.wiley.com/10.1002/bdra.23336">https://doi.wiley.com/10.1002/bdra.23336</a>
Panama	High	Programa Nacional de Malformaciones Congénitas de Panama (PNMC)	-	-	ReLAMC	-	-	-	-	-	-	-	<a href="https://bit.ly/3qyZ7iq">https://bit.ly/3qyZ7iq</a>
Paraguay	Upper-middle	Programa Nacional de Prevención de Defectos Congénitos del Ministerio de Salud Pública del Paraguay (PNPDC)	2016	Hospital-based	ReLAMC	-	Yes	1 year	Major and minor	ICD	-	-	<a href="https://bit.ly/3qww9LG">https://bit.ly/3qww9LG</a> <a href="https://bit.ly/3qyZ7iq">https://bit.ly/3qyZ7iq</a>
Poland	High	Polish Registry of Congenital Malformations (PRCM)	1997	Population-based	EUROCAT	300 (85%)	Yes	2 years	Major and minor	ICD	Live birth, Stillborn and Termination of pregnancy	Yes	<a href="https://bit.ly/3oVZp2G">https://bit.ly/3oVZp2G</a> <a href="https://bit.ly/3oUJOY1">https://bit.ly/3oUJOY1</a> <a href="https://bit.ly/3oWRSAL">https://bit.ly/3oWRSAL</a>

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To be continued



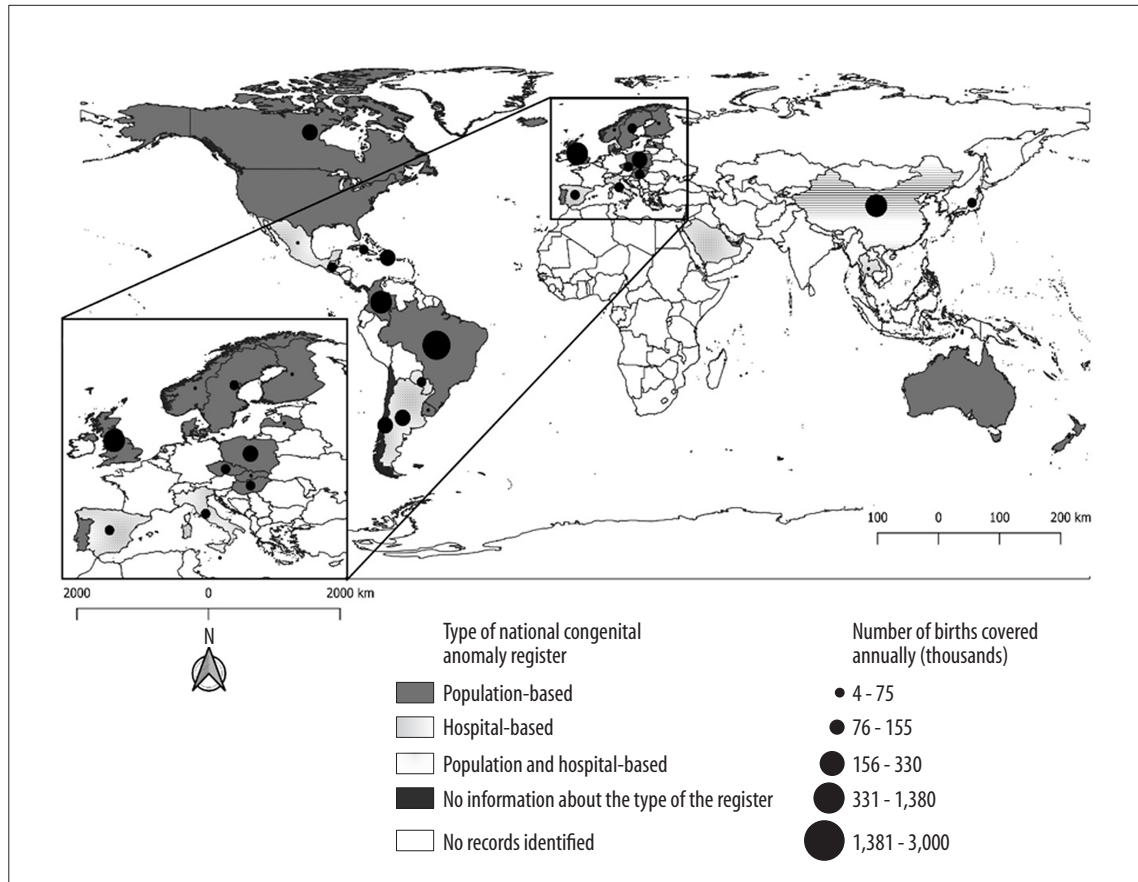
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Box 1 – Main characteristics of registers of congenital anomalies with national coverage around the world, January 10, 2020

Country	Income	Registry	Year created	Type of registry	Network	Number of births (thousands) annually (% of coverage)	Compulsory	Time limit	Type of congenital anomaly	Coding	Pregnancy outcomes	Surveillance	Information source
Portugal	High	Portuguese national registry of congenital anomalies (RENAC)	1995	Population-based	EUROCAT	–	No	At the end of the neonatal period	Major	ICD	Live birth, Stillborn and Termination of pregnancy	Yes	<a href="https://bit.ly/2NgJ7DC">https://bit.ly/2NgJ7DC</a> <a href="http://doi.wiley.com/10.1002/bdra.23530">http://doi.wiley.com/10.1002/bdra.23530</a>
Czech Republic	High	National Registry of Congenital Anomalies (NRC) of the Czech Republic	1964	Population-based	ICBDSR, EUROCAT	110 (100%)	Yes	15 years	–	ICD	Live birth, Stillborn and Termination of pregnancy	Yes	<a href="https://bit.ly/35QVI6U">https://bit.ly/35QVI6U</a> <a href="https://bit.ly/2XHWSNm">https://bit.ly/2XHWSNm</a> <a href="https://doi.org/10.21101/cejph.a4201">https://doi.org/10.21101/cejph.a4201</a>
Dominican Republic	Upper-middle	Sistema Nacional de Vigilancia Epidemiológica	2016	Hospital-based	–	193 (100%)	–	Hospital discharge	Major and minor	ICD	–	–	<a href="https://doi.org/10.26633/RPSP.2019.44">https://doi.org/10.26633/RPSP.2019.44</a>
Singapore	High	National Birth Defects Registry	1992	Population-based	–	–	–	18 years	Major and minor	ICD	Live birth, Stillborn and Termination of pregnancy	–	<a href="http://doi.org/10.3389/fped.2014.00060">http://doi.org/10.3389/fped.2014.00060</a> <a href="https://bit.ly/38S57Nk">https://bit.ly/38S57Nk</a>
Sweden	High	Swedish Medical Birth Registry (MBR)	1964	Population-based	ICBDSR, EUROCAT	100-120 (100%)	Yes	1 year	–	ICD	Live birth, Stillborn and Termination of pregnancy	Yes	<a href="https://doi.wiley.com/10.1080/00016340902934696">https://doi.wiley.com/10.1080/00016340902934696</a> <a href="http://doi.wiley.com/10.1111/j.1651-2227.1989.tb11122.x">http://doi.wiley.com/10.1111/j.1651-2227.1989.tb11122.x</a>
Thailand	Upper-middle	Thailand Birth Defects Registry	2014	Hospital-based	SEAR-NBBD	67 (8.3%)	–	1 year	5 priority types <sup>a</sup>	ICD	Live birth	–	<a href="https://bit.ly/2NgKfQq">https://bit.ly/2NgKfQq</a> <a href="https://doi.org/10.1002/ajmg.c.31690">https://doi.org/10.1002/ajmg.c.31690</a>
Uruguay	High	Registro Nacional de Defectos Congénitos y Enfermedades Raras de Uruguay (RNDCEUR)	2011	Population-based	ReLAMC	28 (58%)	Yes	6 years	Major and minor	ICD	Live birth, Stillborn and Termination of pregnancy	Yes	<a href="https://bit.ly/2KumORM">https://bit.ly/2KumORM</a> <a href="https://doi.org/10.26633/RPSP.2019.44">https://doi.org/10.26633/RPSP.2019.44</a> <a href="https://bit.ly/3qyZ7iq">https://bit.ly/3qyZ7iq</a>

a) NBBDPN is a volunteer-based organization in the United States; (b) the information is related to the United Kingdom; (c) covers all live births delivered or treated at a public hospital in New Zealand; (d) Down syndrome, neural tube defects, cleft lip and palate, limb abnormalities and Duchenne muscular dystrophy. Legend: BINO/CAR: British and Irish Network of Congenital Anomaly Researchers; BPA: British Paediatric Association Classification of Diseases; DC: Centers for Disease Control and Prevention of the United States; ICD: International Classification of Diseases and Related Health Problems; EUROCAT: European surveillance of congenital anomalies; ICBDSR: International Clearinghouse for Birth Defects, Surveillance and Research; ReLAMC: Red Latinoamericana de Malformaciones Congénitas; SEAR-NBBD: South-East Asia Region New-born and Birth Defects.





Note: England, Scotland and Wales registries were jointly represented by the United Kingdom, and the annual coverage was represented by the sum of births in the three registries of congenital anomalies. Filled polygons represent countries that have at least one national registry of congenital anomalies of population-based, hospital-based or both. The size of the circle is proportional to the number (thousands) of births covered annually, in each registry.

**Figure 2 – Worldwide distribution and annual coverage of the registers included in this study (A), highlighting the European continent (B), January 10, 2020**

Regarding gestational outcomes, 33 information systems reported notifications of live births and stillbirths, and of these, 20 included termination of pregnancy in their scope. Brazilian and Thai registers were the only ones to include only live births. Among the registers searched, at least 19 also performed the surveillance of congenital anomalies in their coverage area.

## Discussion

Only 40 registers of congenital anomalies with national coverage were found in 39 different countries worldwide. The registers identified were concentrated exclusively in upper-middle and high-income countries, most of them located in Europe. Thus, the global estimates of congenital anomalies tend to overrepresent these places, to the detriment of others. However, the

production of a series of technical and scientific papers for guidance of the different stages of surveillance of congenital anomalies is an important contribution of the registers present in these countries.

The absence of representativeness in low and lower-middle-income countries is important because, in these contexts, the impact of anomalies on health may be greater, due to the low supply of adequate health services for the care of affected children.<sup>8</sup> In addition, in these countries, other causes of infant mortality, such as malnutrition, poor sanitation conditions and susceptibility to infections, difficulty accessing health care services, among others, are still common, which can help "mask" the real magnitude of congenital anomalies, in epidemiological aspects.<sup>8,11</sup>

Major events involving teratogenic effects, such as the thalidomide tragedy,<sup>4,12</sup> the Chernobyl disaster<sup>13</sup>

and the congenital syndrome associated with Zika virus infection,<sup>14</sup> have been mobilizing nations and their territories for the surveillance of congenital anomalies. Despite the Hungarian register being the oldest found in this study (1962), the Czech Republic National Registry began its activities in 1961 but regular monitoring started in 1964.<sup>15</sup>

The registers with national coverage presented different characteristics among themselves. Most of them were population-based, which makes the prevalence obtained from such systems less susceptible to biases, compared to those obtained from hospital records.<sup>5,16</sup> Hospital-based programs may be an interesting choice, especially in countries with incipient records and/or limited financial resources; in addition, there is a possibility of these same programs expanding into population-based programs in the future.<sup>5,17</sup> It is also important to mention that some registers have complemented their information through active search, such as review of hospital discharge reports (for example some States of the United States),<sup>18</sup> perinatal audit (Australia and New Zealand)<sup>19</sup> and linkage between databases (Scotland).<sup>20</sup>

What, when and how to notify congenital anomalies were also revised questions. Most registers showed compulsory notifications, especially related to major anomalies according to ICD, among live births, stillbirths and children aged equal to or less than 1 year.

Although the study investigated only registers with national coverage, the proportion of births covered annually varied widely. The Brazilian register – Live Birth Information System (Sinasc) - presented the highest absolute number of births covered annually. Sinasc uses the Declaration of Live Birth, a nationally standardized document, in which data on the presence and type of congenital anomaly have been recorded since 1999.<sup>21,22</sup>

Although hospital surveillance has been carried out in a few hospitals in Brazil through the Latin American

Collaborative Study of Congenital Malformations (ECLAMC), the surveillance of congenital anomalies has not been performed systematically yet, throughout the national territory. However, a national surveillance model has been structured within the Health Surveillance Secretariat of the Brazilian Ministry of Health, in articulation with other secretariats of the agency, members of the academic community and medical societies.<sup>23-25</sup>

This study presented some limitations, that is, some registers with national coverage may not have been retrieved through the methodological approach used, due to the lack of material published online. In addition, registers are dynamic and some characteristics, such as 'coverage' can change over time.

In conclusion, this study reviewed the main registers of congenital anomalies with national coverage around the world. A total of 40 registers were analyzed and presented different characteristics, constituting consultation material for professionals interested in the theme and, above all, providing subsidies for reflections on surveillance activities and characteristics that are desirable or possible to be implemented, taking into consideration the reality of each nation.

### Authors' contributions


Cardoso-dos-Santos AC, Alves RSM, Medeiros-de-Souza AC, Bremm JM, Gomes JA, Alves RFS, Araujo VEM and França GVA collaborated with the study conception and design, drafting or critical reviewing of the manuscript intellectual content. Cardoso-dos-Santos AC, Alves RSM and França GVA collaborated with data analysis and interpretation. All authors have approved the final version of the manuscript and declared themselves to be responsible for all aspects of the work, including ensuring its accuracy and integrity.

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