Latin American Network on Congenital Anomalies

Mariela Larrandaburu, MSc PhD

16th May 2024

Regional Meeting

Human Genomics for Health:
Enhancing the Impact of Effective Research

Latin America is a vast region of the earth

- Extension more 21 millons of Km^2,
- Population: 657 million people live
- Countries: More than 20-30 countries
- **Language: Spanish is the principal followed by Portuguese**. in Central America and the Caribbean are spoken: English, French, Papiamento, Dutch, among others.
- Brazil, Mexico, Argentina, Colombia, and Peru concentrate approximately 2/3 of its population.

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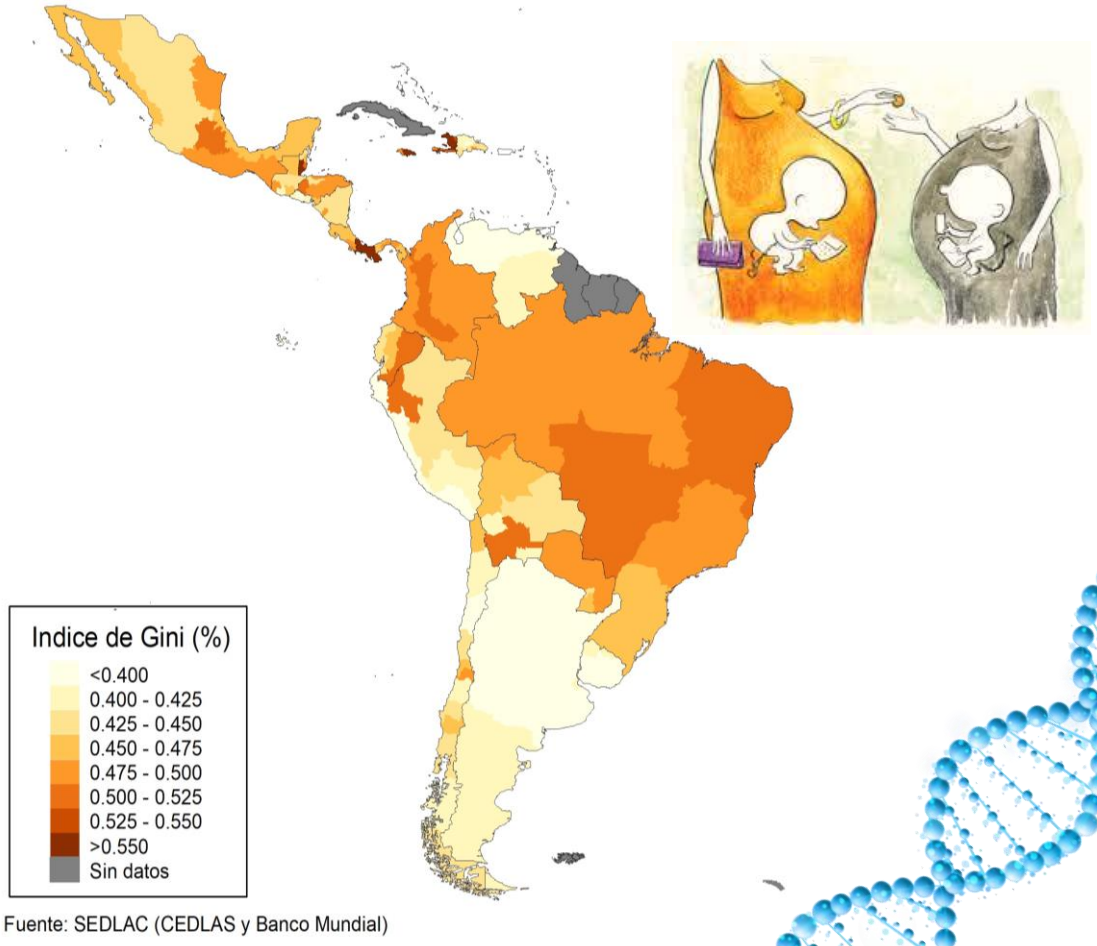
Inequality in Latin America and the Caribbean

Latin America is a highly diversified region both genetically and in its geography, demography, ethnic origin, economic, sociocultural and health aspects.



Fuente: SEDLAC (CEDLAS y Banco Mundial)

(%) Analytical tool that measures the concentration of income among the inhabitants of a region, in a given period of time.

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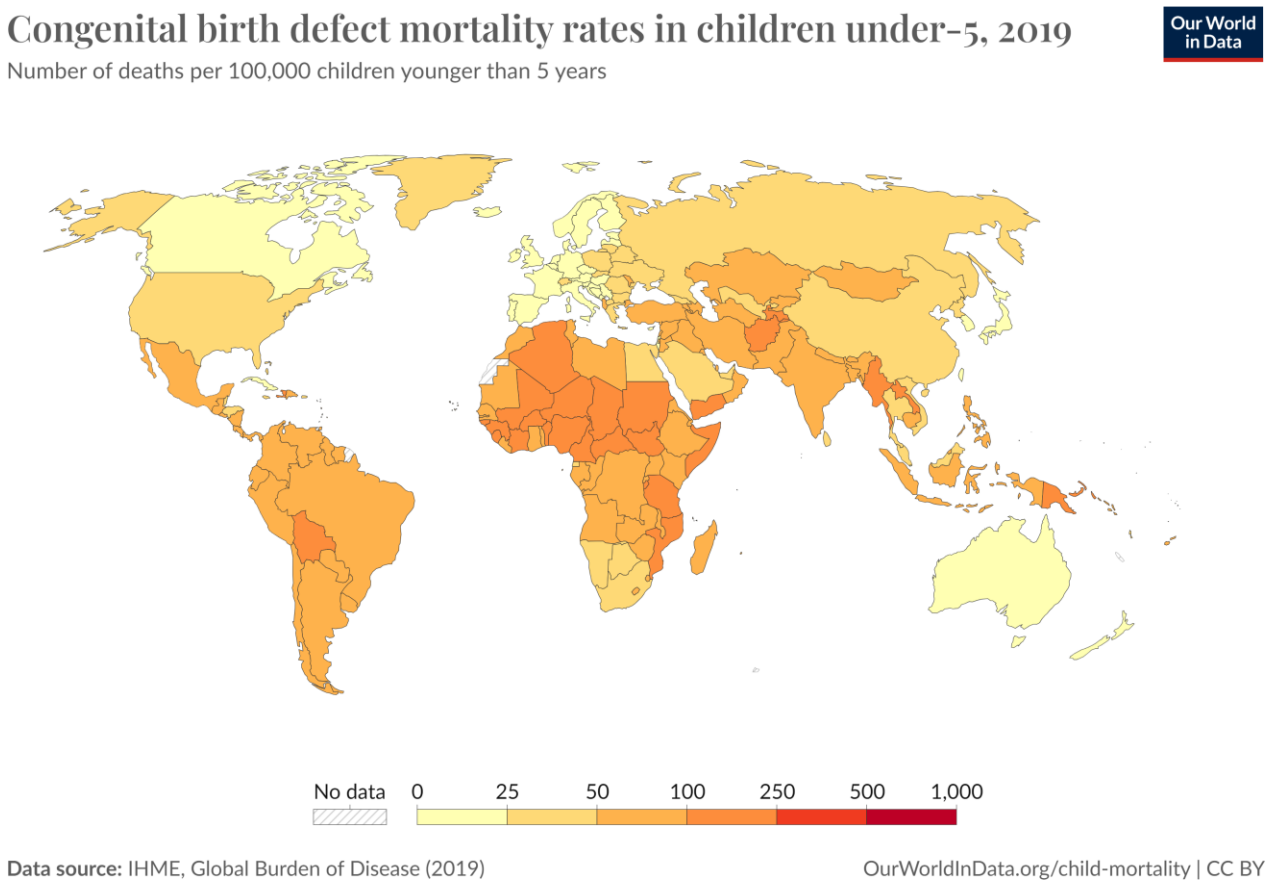
Impact of Rare Diseases and Congenital Anomaly

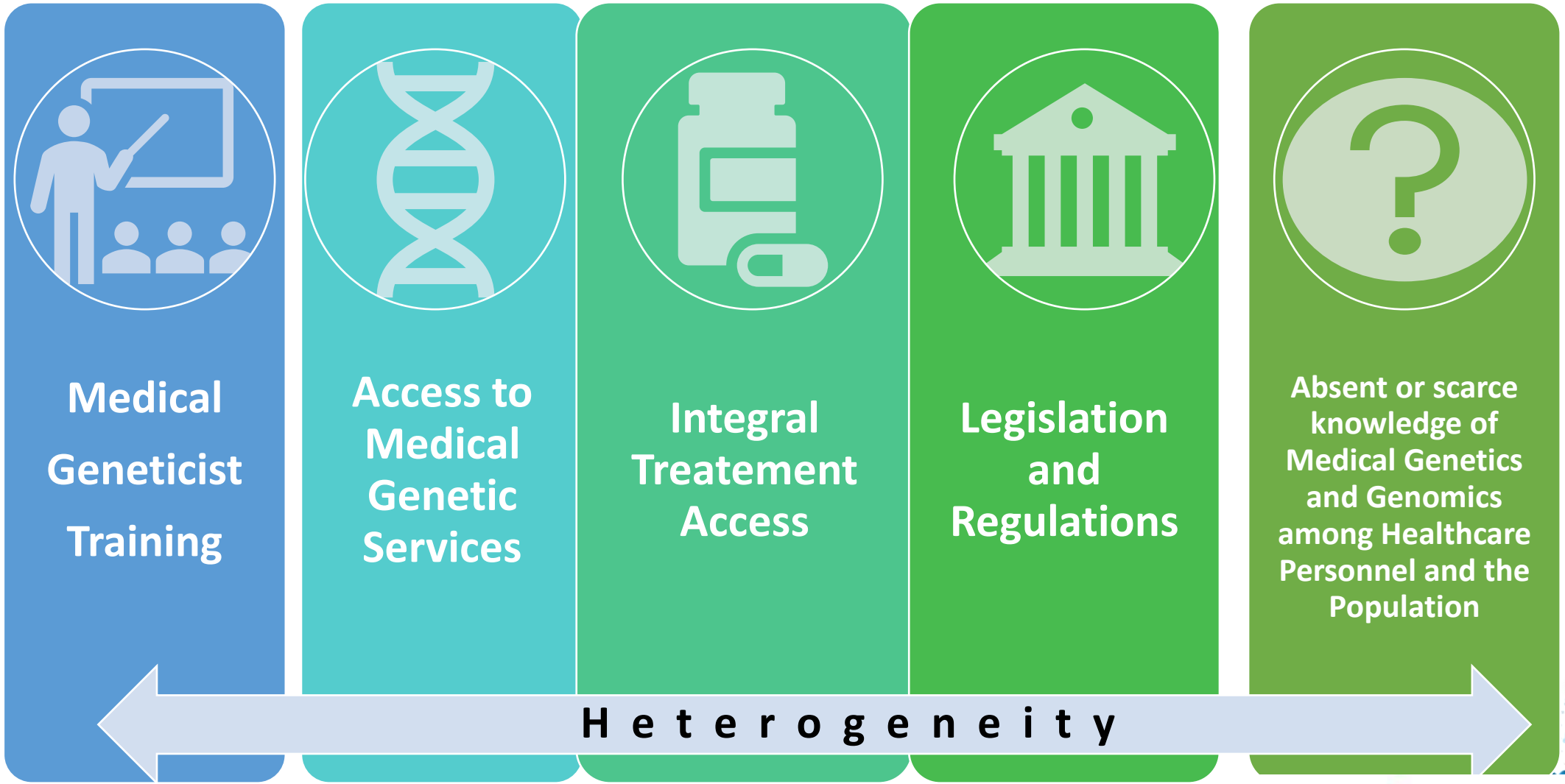


https://ourworldindata.org/grapher/congenital-birth-defect-mortality-rates-in-children-under-5

Number of afected people:
3-5% Total birth

- 13.000.000 live in Brazil
- 7.5.000.000- 10.000.000 in Mexico
- 3.000.000 in Colombia and Argentina
- 1.000.000 - 1.5,000.000 million in Chile, Peru, Bolivia, Paraguay, Venezuela
- 60,000- 210,000 in Uruguay

- **2/3 of those affected live in Brazil, Mexico, Colombia, Peru, and Argentina**



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MILESTONES Surveillance Systems in BD

1960 | 2010 | 2015 | 2019 | 2020

63° AWH-WHO

Birth Defect

Sustainable Development Goals

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ANOMALIA CONGENITA	CIE-10	N° ORPHANET
Síndrome de Down	Q090	870
Anencefalia	Q00	1048
Espina Bífida	Q05	823
Encefalocele	Q01	199647
Labio Leporino y paladar Hendido	Q37	199306
Hipospadias	Q54	440
Focomelia de MS	Q71.1	294975
Ectrodactilia	Q71.6;Q72.7	2440
Hidranencefalia	Q04.3	2177
Gastrosquisis	Q79.3	2368
Onfalocele	Q79.2	660
Hernia Diafragmática	Q79.0	2140
Tetralogia de Fallot	Q21.3	3303

orphanet

DESAFÍOS: Codificación, Clasificación de estas entidades

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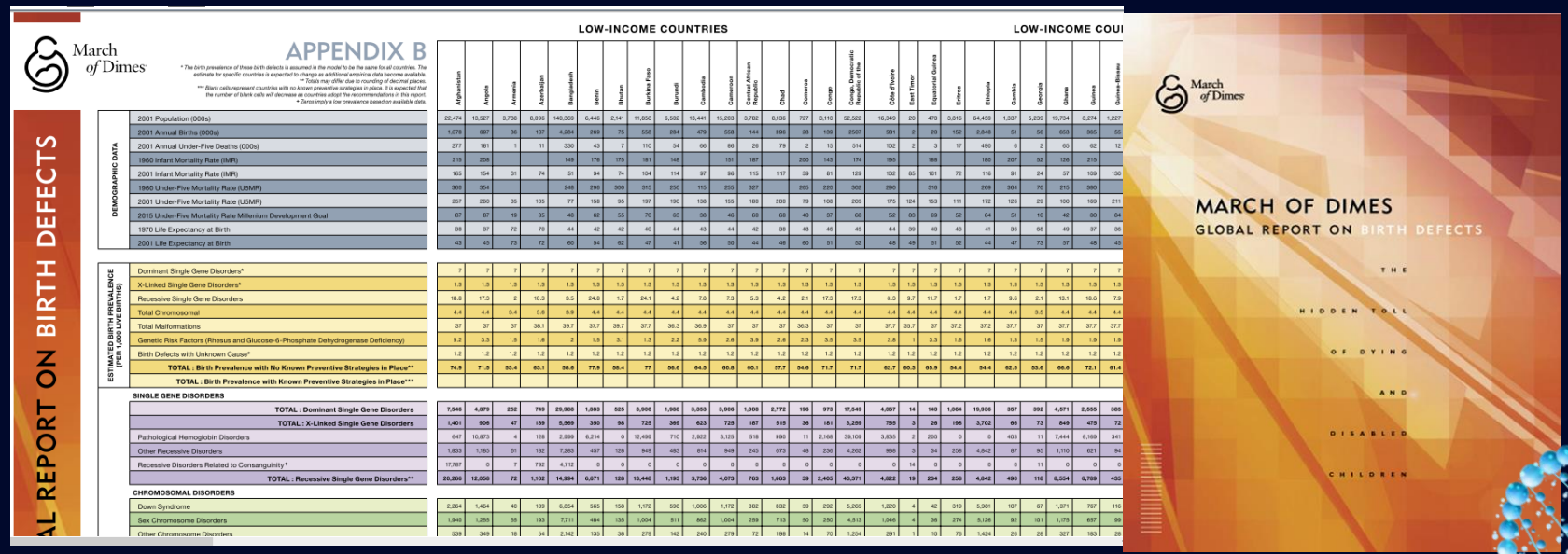
March of Dimes Global Report on Birth Defects 2006

- Global Report 2006 Christianson A, Howson CP, Modell B
- Estimates developed by Modell & Christianson (2004)
- Modell Global Database (MGDb)
- Estimate the number of births affected by subset of congenital disorders & outcomes of these births, in no-care & current care scenarios.


March of Dimes

APPENDIX B

LOW-INCOME COUNTRIES

LOW-INCOME COUI

AL REPORT ON BIRTH DEFECTS

DEMOGRAPHIC DATA

2001 Population (000s)

2001 Annual Births (000s)

2001 Annual Under-Five Deaths (000s)

1960 Infant Mortality Rate (IMR)

2001 Infant Mortality Rate (IMR)

1960 Under-Five Mortality Rate (U5MR)

2001 Under-Five Mortality Rate (U5MR)

2015 Under-Five Mortality Rate Millenium Development Goal

1970 Life Expectancy at Birth

2001 Life Expectancy at Birth

ESTIMATED BIRTH PREVALENCE (PER 1,000 LIVE BIRTHS)

Dominant Single Gene Disorders*

X-Linked Single Gene Disorders*

Recessive Single Gene Disorders

Total Chromosomal

Total Malformations

Genetic Risk Factors (Rhesus and Glucose-6-Phosphate Dehydrogenase Deficiency)

Birth Defects with Unknown Cause*

TOTAL : Birth Prevalence with No Known Preventive Strategies in Place**

TOTAL : Birth Prevalence with Known Preventive Strategies in Place***

SINGLE GENE DISORDERS

TOTAL : Dominant Single Gene Disorders

TOTAL : X-Linked Single Gene Disorders

Pathological Hemoglobin Disorders

Other Recessive Disorders

Recessive Disorders Related to Consanguinity*

TOTAL : Recessive Single Gene Disorders**

CHROMOSOMAL DISORDERS

Down Syndrome

Sex Chromosome Disorders

Accions

WHO Technical Working Group on Burden of Birth Defects (BBDTWG)

In 2022, WHO formed a Burden of Birth Defects Technical Working Group (BBD-TWG) composed of **27 technical experts** to review the previous estimationn work, identify available data and advise on optmal approaches to estimation. **5 Priority conditions were identified by the BBD-TWG for which prevention and care strategies exist:**

1. Structural anomalies that can be seen at birth or PN as NTDs, Orofacial Clefts, Abdominal Wall Anomalies
2. Trisomies and Syndromic Conditions
3. Congenital Heart Defects
4. Congenital Hypothyroidism and Congenital Syphilis
5. Haemoglobinopathies

https://ishg2024.org/abstracts/Dr-Kathleen-Strong.pdf

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Opinion and analysis

Pan American Journal of Public Health

Zika virus infection and congenital anomalies in the Americas: opportunities for regional action

Mariela Larrandaburu,[1] Fernanda Sales Luiz Vianna,[1] André Anjos da Silva,[1] Luis Nacul,[2] Maria Teresa Vieira Sanseverino,[1] and Lavínia Schuler-Faccini[1]

Suggested citation Larrandaburu M, Vianna FSL, Anjos-da-Silva A, Nacul L, Sanseverino MTV, and Schuler-Faccini L. Zika virus infection and congenital anomalies in the Americas: opportunities for regional action. Rev Panam Salud Publica. 2017;41:e174. doi: 10.26633/RPSP.2017.174.

ABSTRACT

The Zika virus (ZIKV) was identified in 1947 in the Zika forest in Uganda, but recently it has emerged as a public health threat. The first evidence of human infection occurred in 1952, but only in April 2007 was the first outbreak in humans recognized. In the Americas, a ZIKV outbreak began in Brazil in 2015, and from the second half of 2015 We consider this to be a unique opportunity for countries in the Region of the Americas to develop, strengthen, and improve surveillance systems for congenital anomalies and teratogen information services. Creating health needs assessment tools for low- and middle-income countries may help them to develop effective policies to ensure primary, secondary, and tertiary prevention measures for congenital anomalies. Such initiatives will be useful for ZIKV congenital syndrome control and also for having a much wider impact on a significant proportion of preventable and manageable congenital conditions.

Keywords Zika virus; microcephaly; epidemiological surveillance; Americas.

On 1 February 2016, the World Health Organization (WHO) declared that the clusters of microcephaly cases and other neurological disorders such as Guillain-Barré syndrome in some areas affected by Zika virus (ZIKV) constituted a Public Health Emergency of International Concern (PHEIC) (1). "The increased prevalence of microcephaly at birth is particularly alarming, because it is a painful burden on the families and communities," pointed out WHO Director-General Margaret Chan (2).

The Zika virus was identified in the 1940s in the Zika forest in Uganda in monkeys. The first evidence of infection in humans occurred in 1952. Some five decades later, the international community recognized that the first ZIKV outbreak in humans had occurred in April 2007 in Yap, one of the states of the Federal States of Micronesia, in the Pacific Ocean. At that time, the transmission was reported in 10 other Pacific island countries and areas (3). In the Americas, the virus en-

https://iris.paho.org/handle/10665.2/34540

Review > Epidemiol Serv Saude. 2020;29(4):e2020093.

doi: 10.5123/s1679-49742020000400003. Epub 2020 Jul 29.

International collaboration networks for the surveillance of congenital anomalies: a narrative review

[Article in English, Portuguese]

Augusto César Cardoso-Dos-Santos [1], Vivyanne Santiago Magalhães [1], Ana Cláudia Medeiros-de-Souza [1], João Matheus Bremm [1], Ronaldo Fernandes Santos Alves [1], Valdelaine Etelvina Miranda de Araujo [1], Eduardo Marques Macario [1], Wanderson Kleber de Oliveira [1], Giovanny Vinícius Araújo de França [1]

Affiliations + expand

PMID: 32756830 DOI: 10.5123/s1679-49742020000400003

frontiers in Public Health

OPINION
published: 02 November 2021
doi: 10.3389/fpubh.2021.753342

Check for updates

Why are Birth Defects Surveillance Programs Important?

Débora Gusmão Melo [1], Maria Teresa Vieira Sanseverino [2,3], Thanyse de Oliveira Schmalfuss [4] and Mariela Larrandaburu [5]*

[1] Department of Medicine, Federal University of São Carlos (UFSCar), São Carlos, Brazil, [2] School of Medicine, Pontifical Catholic University of Rio Grande do Sul, Porto Alegre, Brazil, [3] Medical Genetics Service, Clinical Hospital of Porto Alegre, Porto Alegre, Brazil, [4] School of Medicine, Federal University of Rio Grande do Sul (UFRGS), Porto Alegre, Brazil, [5] Disability and Rehabilitation Program, Ministry of Public Health of Uruguay, Montevideo, Uruguay

Keywords: birth defects, congenital anomalies, public health surveillance, epidemiological monitoring, registries, teratogens, newborn screening

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PAHO Pan American Health Organization World Health Organization Americas | PAN AMERICAN JOURNAL OF PUBLIC HEALTH

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Systems for surveillance of birth defects in Latin America and the Caribbean: present and future

Durán et al.

Objectives.

To determine the availability of national systems for surveillance of birth defects in Latin America and the Caribbean and describe their characteristics.

Methods.

Cross-sectional study based on a semi-structured, self-administered online survey sent in 2017 by local representative offices of the Pan American Health Organization to authorities at the ministries of health of all countries in Latin America and the Caribbean. The survey obtained information on the availability and characteristics of national systems for surveillance of birth defects in each country.

Results.

Eleven countries have a national system for surveillance of birth defects: Argentina, Colombia, Costa Rica, Cuba, Dominican Republic, Guatemala, Mexico, Panama, Paraguay, Uruguay, and Venezuela. These systems have heterogeneous features: six are hospital-based; 10 include both live births and stillbirths in their case definition. All the surveillance systems include cases with severe and minor defects, except in Argentina, Colombia, and Guatemala, where only severe birth defects

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https://doi.org/10.26633/RPSP.2019.44

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ReLAMC

RED LATINOAMERICANA de MALFORMACIONES CONGÉNITAS

Red de Registros que realiza de forma cooperativa la vigilancia epidemiológica de anomalías congénitas en América Latina

AJMG AMERICAN JOURNAL OF medical genetics PART C
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RESEARCH ARTICLE

The Latin American network for congenital malformation surveillance: ReLAMC

Iêda Maria Orioli, Helen Dolk, Jorge Lopez-Camelo, Boris Groisman, Adriana Benavides-Lara, Lucas Gabriel Gimenez, Daniel Mattos Correa, Marta Ascurra ... See all authors

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https://www.relamc.org/es

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RED LATINOAMERICANA de MALFORMACIONES CONGÉNITAS

Red de Registros que realiza de forma cooperativa la vigilancia epidemiológica de anomalías congénitas en América Latina

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- ARGENTINA - RENAC
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- COSTA RICA - CREC
- CHILE - RENACH
- MAULE - RRMC SSM
- SÃO PAULO - SINASC/SIM MSP
- BRASIL - SINASC/SIM BRS

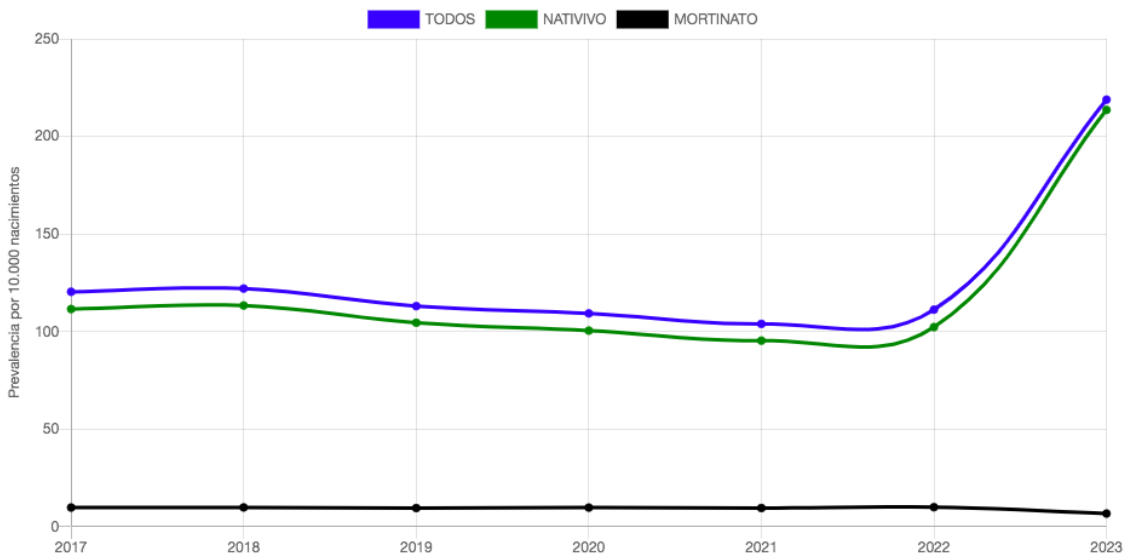
Todos los registros: TODOS

Anomalías: TODAS

Años: 2017 a 2023: 2017 – 2023

Borrar | Aplicar

Prevalencia por 10.000 nacimientos. Anomalías: TODAS - 2017 a 2023 - Registro: TODOS



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VISION

CONGENITAL ANOMALIES

RARE DISEASES

DISABILITY

INBORN METABOLISM ERRORS

COMMON DISEASES

COMMON DISEASES

ONCOLOGICAL PATHOLOGY

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Thank you

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