Latin American Network on Congenital Anomalies

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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 millions of Km²,
- 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.
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.

(%): Analytical tool that measures the concentration of income among the inhabitants of a region, in a given period of time.
Impact of Rare Diseases and Congenital Anomaly

Number of affected 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


Data source: IHME, Global Burden of Disease (2019)
Medical Geneticist Training

Access to Medical Genetic Services

Integral Treatment Access

Legislation and Regulations

Absent or scarce knowledge of Medical Genetics and Genomics among Healthcare Personnel and the Population

Heterogeneity
MILESTONES Surveillance Systems in BD


- 63º AWH-WHO Birth Defect
- Sustainable Development Goals
- Naciones Unidas

Defectos Congénitos
Objetivos de Desarrollo Sostenible

M Larrandaburu, 2024
<table>
<thead>
<tr>
<th>ANOMALIA CONGENITA</th>
<th>CIE-10</th>
<th>N° ORPHANET</th>
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<tbody>
<tr>
<td>Síndrome de Down</td>
<td>Q090</td>
<td>870</td>
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<td>Anencefalia</td>
<td>Q00</td>
<td>1048</td>
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<tr>
<td>Espina Bífida</td>
<td>Q05</td>
<td>823</td>
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<tr>
<td>Encefalocele</td>
<td>Q01</td>
<td>199647</td>
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<tr>
<td>Labio Leporino y paladar Hendido</td>
<td>Q37</td>
<td>199306</td>
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<td>Hipospadias</td>
<td>Q54</td>
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<td>Focomelia de MS</td>
<td>Q71.1</td>
<td>294975</td>
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<tr>
<td>Ectrodactilia</td>
<td>Q71.6;Q72.7</td>
<td>2440</td>
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<td>Hidranencefalia</td>
<td>Q04.3</td>
<td>2177</td>
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<td>Gastroquisis</td>
<td>Q79.3</td>
<td>2368</td>
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<td>Q79.2</td>
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<td>Hernia Diafragmática</td>
<td>Q79.0</td>
<td>2140</td>
</tr>
<tr>
<td>Tetralogía de Fallot</td>
<td>Q21.3</td>
<td>3303</td>
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</tbody>
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**DESAFÍOS: Codificación, Clasificación de estas entidades**
March of Dimes Global Report on Birth Defects 2006

- Global Report 2006 Christianson A, Howson CP, Modell B
- 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.
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 estimation work, identify available data and advise on optimal 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

M Larrandaburu, 2024
Zika virus infection and congenital anomalies in the Americas: opportunities for regional action

Mariela Larrandaburu, Fernanda Sales Luiz Vianna, André Anjos de Silva, Luis Nacul, Maria Teresa Vieira Sarceverino, and Lavinia Schuler-Faccini

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 teratogenic 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 resources 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 Federated 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 emi...
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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- Strengthening public health policies for childhood cancer: Peru’s achievements through the WHO Global Initiative for Childhood Cancer
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- A short list of high-priority indicators of health system responsiveness for aging: an eDelphi consensus study
  Special report

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

First published: 14 December 2020 | https://doi.org/10.1002/ajmg.c.31872 | Citations: 8

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https://www.relamc.org/es
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<th>PARAGUAY - RENADECOPY</th>
<th>NUEVO LEÓN - ReDeCon HU</th>
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Prevalencia por 10,000 nacimientos. Anomalías: TODAS - 2017 a 2023 - Registro: TODOS

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Accions
Thank you

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