



INSTITUTO DE CIENCIAS E INNOVACIÓN EN MEDICINA  
Facultad de Medicina  
Clínica Alemana - Universidad del Desarrollo



**HOSPITAL**  
**PADRE HURTADO**  
Creciendo con Ustedes

# Rare Diseases in Chile

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## “Macrozones”

North

Central

SouthCentral

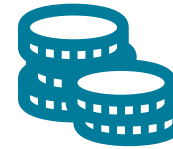
South

Austral

## Chile in figures, 2019



**Population** ~19MM  
40% Metropolitan Region of Santiago



**GDP per capita** ~ U\$27,000  
Median income ~ U\$6,700



**Health expenditure**  
9.1% GDP  
U\$1,450/per capita (33% out-of-pocket)



**Health care system:** mixed  
Public 80%, Private 15%, Armed Forces 3%



**R&D Expenditure** 0.35 % GDP  
U\$1.5 billion

# 1. Existing Legislation and Policies on RDs

Universal newborn screening program for PKU-since 1992

Pilot for other 20 IEM, cystic fibrosis (suspended 2019) and hearing loss

Explicit Health care guarantees Law (Health care reform-2004)

Guidelines and coverage for 80 **high-burden health care issues**

RDs: Hemophilias, Cystic Fibrosis, Pediatric cancers

**High cost medication and devices law** (2014-30 diseases)

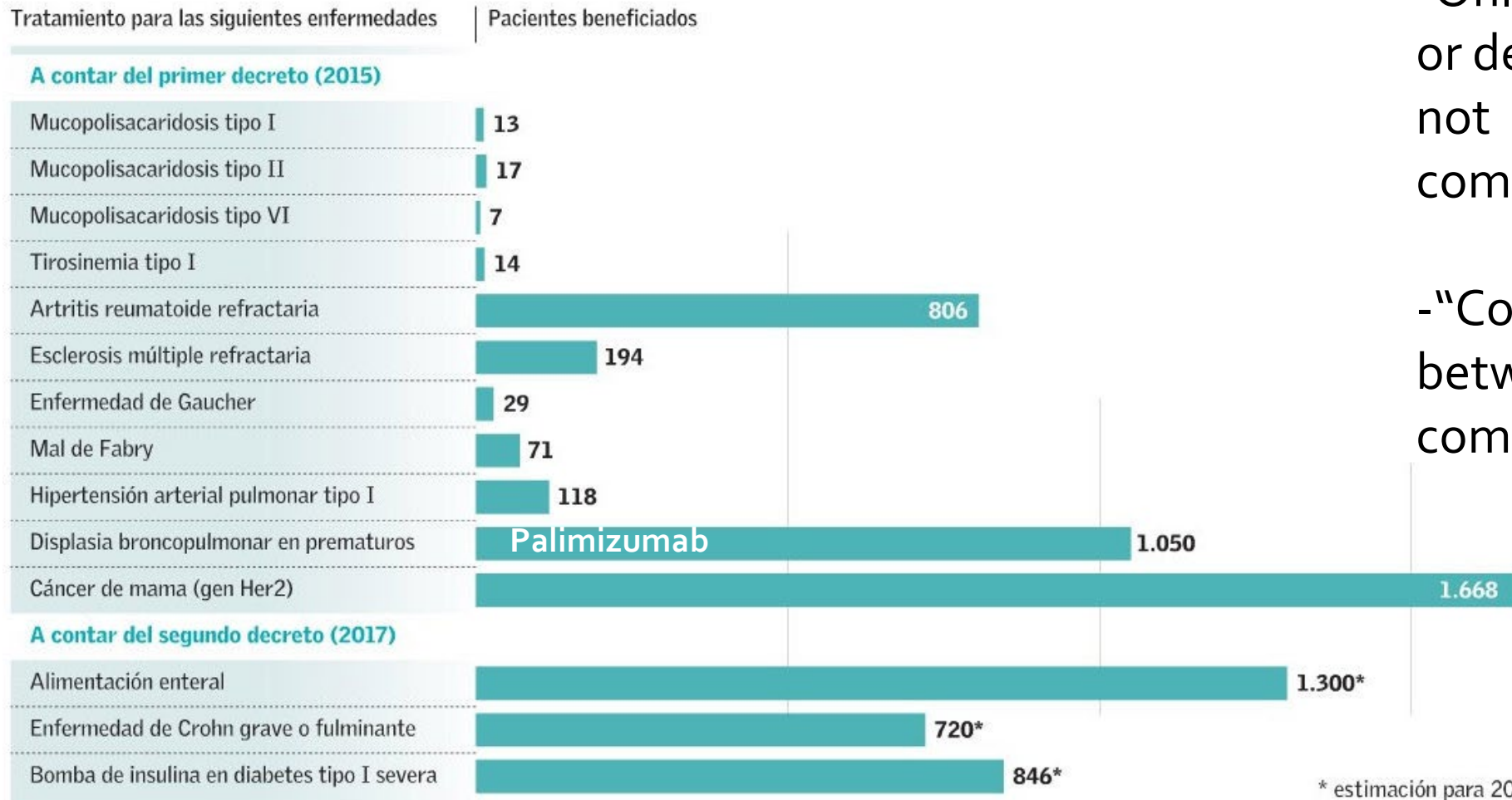
16 RDs: MPS I, II, VI, Gaucher, Fabry, tyrosinemia type 1 Epidermolysis Bullosa, Huntington, Multiple sclerosis, ALS

National RD awareness and education day (2020)

# ...But this is not a rare diseases law

-Only high cost medications or devices are covered, not diagnosis or comprehensive therapies

-“Competition” for resources between RDs and with common conditions



\* estimación para 2017

## 2. Current National Activities and Areas of Emphasis

### 1. Development of a (draft) National plan for RDs

Convened by Senator Carolina Goic and Minister of Health

Multi-stake holder (n=150) collaborative working groups:

- Education, and Awareness
- Registries, information and Research
- Governance and organization
- Clinical Services
- Financial protection

1st document to be delivered to the Minister of Health on Friday April 30, 2021

### 2. Announcement by MoH of commitment to include RDs in Health Goals and Plans for 2021-2030

# 3. Challenges, Barriers, or Obstacles

Lack of local definition of RDs

Lack of information on n patients, diagnosis (or lack thereof), needs, available services and gaps

Many immediate needs, but gaps will take a long time to fill  
how to establish priorities?

Need for consensus in several areas

Funding mechanism.... Specific fund vs integrated in the system

Operational mechanism for services: National institute vs integrated system

Quality standards: which? By whom? (vs "self-declared" centers of excellence)

# 4. Legislative or Policy Needs in Research, Regulatory, or Healthcare Services



2021: First explicit inclusion of RDs, biobanks and data sharing in Ministry of Science's RFA

Impact of future data privacy law in research and clinical care?

Role of telehealth in contributing to access

# **5. Opportunities for Future Collaborations & 6. Suggested Activities for Country, Region, or a Sustainable ERCAL**

Repository and analysis of programs and legislations in LAC countries, with implementation perspective (what works? what really doesn't?)

Consensus on best practices, in regional contexts for specific RDs

Joint educational activities and shared resources-

Repository of educational materials, for patients and clinicians in Spanish and Portuguese





# Announcement and Invitation



Global Genomic  
Medicine Collaborative

- Home
- About Us ▾
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- Events

**6th G2MC International Conference**  
 Genomic Medicine Implementation in Low-Resource Settings  
**September 29 – October 1, 2021 | Virtual Meeting**

<https://g2mc.org/santiago2021/>



**G2MC Young Investigators Forum**  
 6th G2MC International Conference

- Abstract Template
- Abstract Submission Form
- Ask a Question



April 30 for oral presentations  
May 31 for virtual posters

Trainees and recent graduates  
FOCUS on Implementation in Limited Resources Settings  
Networking with global leaders

<https://g2mc.org/2021YIF/>