GENOMIC MEDICINE INITIATIVES (GMI) in South America

GABRIELA REPETTO, MD
Center for Genetics and Genomics
Facultad de Medicina
Clínica Alemana Universidad del Desarrollo
and Sociedad de Genética de Chile
grepetto@udd.cl
BACKGROUND: R&D SPENDING

%GDP (Billion U$)

<table>
<thead>
<tr>
<th>Country</th>
<th>R&amp;D Spending</th>
</tr>
</thead>
<tbody>
<tr>
<td>Brazil</td>
<td>1.24 (35.4)</td>
</tr>
<tr>
<td>Uruguay</td>
<td>0.32 (0.24)</td>
</tr>
<tr>
<td>Argentina</td>
<td>0.61 (5.4)</td>
</tr>
<tr>
<td>Chile</td>
<td>0.39 (1.6)</td>
</tr>
<tr>
<td>Peru</td>
<td>0.1 (0.15)</td>
</tr>
<tr>
<td>Colombia</td>
<td>0.26 (1.5)</td>
</tr>
</tbody>
</table>

http://data.worldbank.org/indicator/GB.XPD.RSDV.GD.ZS
SURVEY REGARDING GMI SENT TO GENETIC SOCIETIES

- Latin American Network of Human Genetics (RELAGH)
- Latin American Genetics Association (ALAG)

1. Current initiatives
2. If +: Goals/Funding/Status
WES in 100 patients with rare disorders
http://www.bitgenia.com/

Platform for development of genomic capabilities
Initial focus on local variation
http://urugenomes.org
Centro de Investigación Genética y Genómica - CIGG

Platform for development of genomic capabilities (cancer, rare disorders, pharmacogenetics)

BIPMed
Public genomic database
Population reference + patients with epilepsies

In partnership with GA4GH

www.bipmed.org
INITIATIVES

Population database
http://www.chilegenomico.cl

Prospective cohort
(n 10,000/10 year f/u)
Cancer and cardiovascular diseases
http://www.accdis.cl

Ancestry informative markers

CORFO + Pfizer
Lung and breast cancer
Targeted HTS in 6000 cancer samples with PGx focus
https://pfizercmp.com/
Current Project:
joining efforts to create a public
database of Chilean variation and
contribute to regional/ worldwide
information
With collaboration from BIPMed
CHILEAN GENETICS SOCIETY presented a proposal to the Ministry of Health for the development of Medical Genetics and Genomics

5 Areas
Clinical Services  
Research  
Education  
Regulation  
International collaboration

3 Aspects
Objectives  
Gaps  
Proposals
Several countries in South America are developing capabilities for GM

Current focus on:
- Local variation databases
- Emerging studies on specific conditions: rare diseases and cancer

Mostly at research level
MAIN CHALLENGES

- Funding
- Education and training
- Elevated costs of HTS, biobanks, etc
- Insertion in clinical care
- Regional and worldwide collaboration
GENOMIC MEDICINE INITIATIVES (GMI) in South America

GABRIELA REPETTO, MD
Center for Genetics and Genomics
Facultad de Medicina
Clínica Alemana Universidad del Desarrollo
Sociedad de Genética de Chile
grepetto@udd.cl