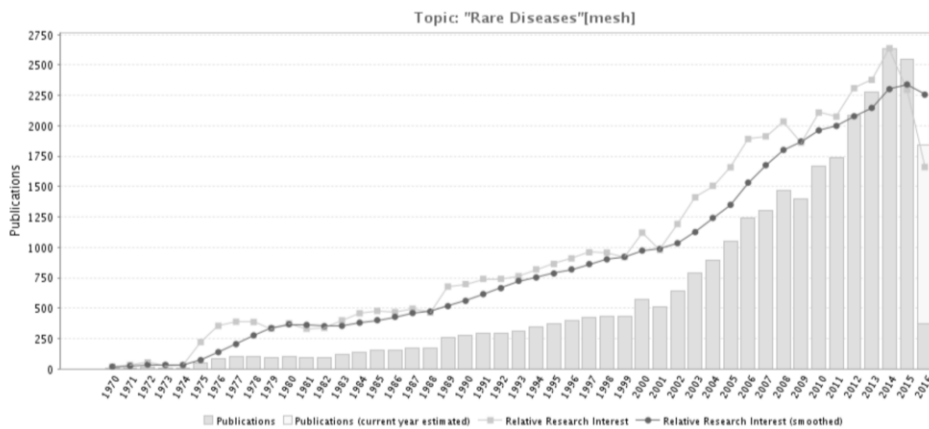
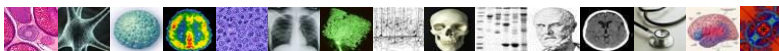


Los Desafíos de las Enfermedades Raras

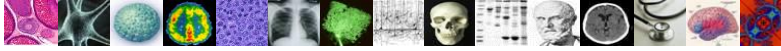
Diego Rosselli M.D., Ed.M., M.Sc.
 Departamento de Epidemiología Clínica y Bioestadística
 Facultad de Medicina, Pontificia Universidad Javeriana
 Sao Paulo, Septiembre de 2017

Conflictos de interés: He sido conferencista para Abbott, Abbvie, Acelity, Alexion, Amgen, Astrazeneca, Baxter, Bayer-Schering, Biomarin, Biotoscana, BristolMyersSquibb, Boehringer-Ingelheim, Eli Lilly, Genzyme, Grünenthal, GSK, Janssen, Lafrancol, Linde, Merck, MSD, Mundipharma, Novartis, Novo Nordisk, Pfizer, Roche, Sanofi-Aventis, Sanofi-Pasteur, Shire, Stendhal, Takeda, Zambon

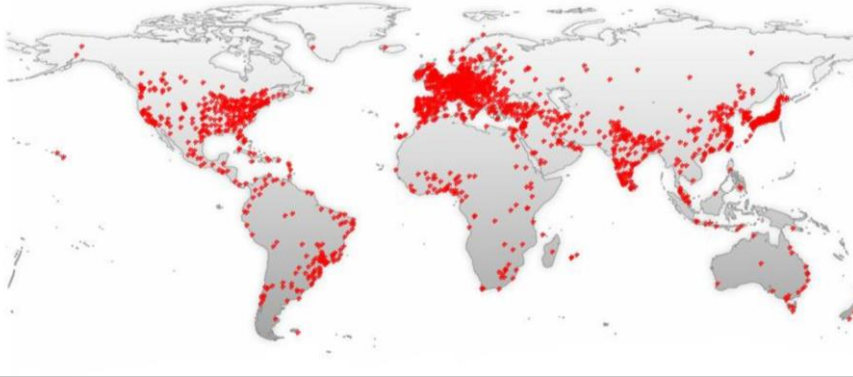
diego.rosselli@gmail.com



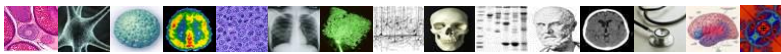
diego.rosselli@gmail.com



3/21

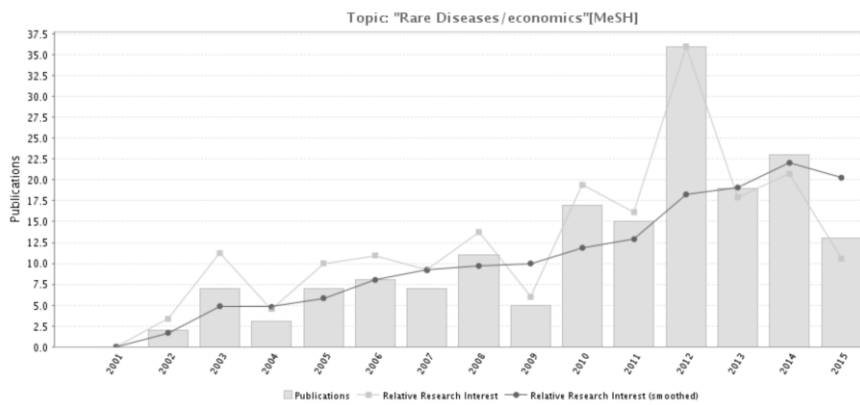


diego.rosselli@gmail.com

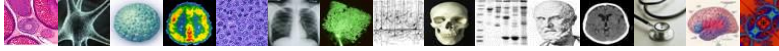


4/21

statistics of 173 documents



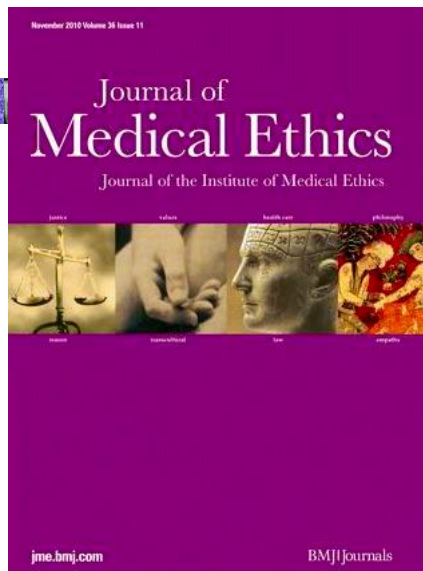
diego.rosselli@gmail.com



5/21

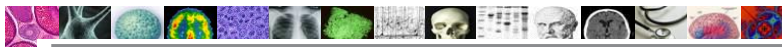


diego.rosselli@gmail.com



6/21

diego.rosselli@gmail.com



Ethical and economic considerations of rare diseases in ethnic minorities: the case of mucopolysaccharidosis VI in Colombia

Diego Rosselli,¹ Juan-David Rueda,¹ Martha Solano²

¹Clinical Epidemiology and Biostatistics, Pontificia Universidad Javeriana, Bogota, Colombia
²Department of Neuropediatrics, Fundacion Cardioc infantil, Bogota, Colombia

Correspondence to
Dr Diego Rosselli, Clinical Epidemiology and Biostatistics, Pontificia Universidad Javeriana, Carrera 7 No 40-62, Bogota 110311, Colombia;
diego.rosselli@gmail.com

Received 25 August 2011
Revised 8 February 2012
Accepted 5 April 2012
Published Online First
1 May 2012

ABSTRACT
Mucopolysaccharidosis VI is an autosomal recessive lysosomal storage disorder associated with severe disability and premature death. The presence of a mucopolysaccharidosis-like disease in indigenous ethnic groups in Colombia can be inferred from archaeological findings. There are several indigenous patients with mucopolysaccharidosis VI currently receiving enzyme replacement therapy. We discuss the ethical and economic considerations, regarding both direct and indirect costs, of a high-cost orphan disease in a marginalised minority population in a developing country.

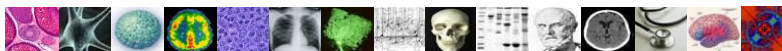
Rare diseases warrant different ethical and economic considerations, particularly when present in indigenous populations or other minorities.

development.⁹ In Europe, the cost per year per patient has been estimated to range between €150 000 and €450 000.¹⁰

Evidence of the presence of an MPS-like disease dates back to prehistoric times in southwestern Colombia. Several clay figurines from the Tumaco culture (300 BC to 500 AD) depict patients with craniofacial and spinal malformation, interpreted by experts as cases of MPS.^{11–13} The Totoró is one of 82 indigenous groups that survive in Colombia; they live in the southwestern Andean highlands, and their population is estimated to be 4130.¹⁴ Several cases of MPS VI have been confirmed in this and other indigenous groups in Colombia.

These patients normally have a late diagnosis and encounter various barriers to access to therapy, which are common findings in rare diseases. An ethical dilemma, often seen when treating indige-

diego.rosselli@gmail.com



orphanet The portal for rare diseases and orphan drugs

Languages: Français | English | Español | Deutsch | Italiano | Português

Homepage Help Contact us

Rare diseases Orphan drugs Expert centres Diagnostic tests Research and trials Patient organisations Professionals and institutions Other information

SIMPLE SEARCH

Search a disease → OK

> Alphabetical list of rare diseases

OTHER SEARCH OPTION(S)

- > Orphan drugs
- > Research and trials
- > Diagnostic tests
- > Patient organisations
- > Expert centres
- > Professionals and institutions

ORPHANET DATA

| | |
|----------------|---------|
| Diseases | : 5954 |
| Expert centres | : 4942 |
| Laboratories | : 5424 |
| Professionals | : 15019 |
| Daily visitors | : 12810 |

RARE DISEASES

- > Information about a disease
- > Alphabetical list
- > Search by clinical sign
- > Search by gene
- > Emergency guidelines
- > Encyclopaedia for patients
- > Encyclopaedia for professionals
- > Classifications
- > About Rare Diseases
- > Prevalence of Rare Diseases

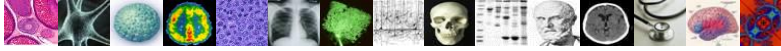
There is no disease so rare that it does not deserve attention

Rare diseases are rare, but rare disease patients are numerous

About Orphanet | Quality charter Register your activity

Languages: Français | English | Español | Deutsch | Italiano | Português

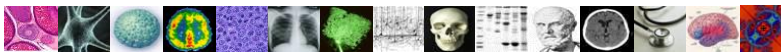
diego.rosselli@gmail.com



9/21

| País | Prevalencia (x 100.000) | Fuente |
|----------------|------------------------------------|--|
| Australia | 11 | Orphan Drug Program 1997 |
| Colombia | 20 | Ley 1392 de 2010 (originalmente 50, luego modificado por Ley 1438 de 2011) |
| Estados Unidos | 67 | Orphan Drug Act 1983 |
| Francia | 50 | Regulation EC n 141/2000 |
| Holanda | 50 | Regulation EC n 141/2000 |
| Inglaterra | 2 | National Commissioning Group, para enfermedad "ultra rara" |
| Japón | 40 | Orphan Drug Act 1993 |
| Noruega | 10 | Norwegian Directorate of Health |
| OMS | 65 | Organización Mundial de la Salud |
| Suecia | 10 | Swedish National Board of Health and Welfare |
| Turquía | 1 | Ministry of Health |
| Unión Europea | 50 | Regulation ECn141/2000 |

diego.rosselli@gmail.com

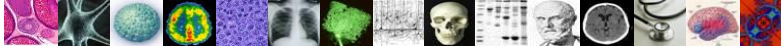


10/21

La investigación en enfermedades raras:

- Pocos incentivos
- Tamaños de muestra reducidos
- Desenlaces "blandos"
- Altos costos

diego.rosselli@gmail.com



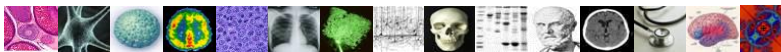
11/21

Tabla 3. Ejemplos de tamaños de muestra de estudios clínicos en enfermedades de depósito lisosomal.

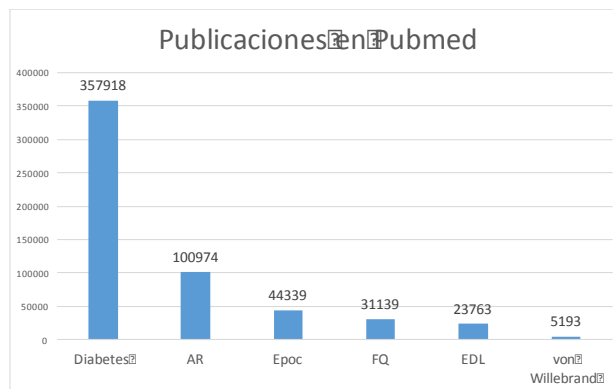
| Enfermedad | Tamaño de muestra | |
|---|--------------------|-------------|
| | Estudios Fase I-II | Fase III-IV |
| α-manosidosis | 12 | 52 |
| aspartil-glicosaminuria | 12 | 45 |
| galactosialidosis | 12 | 52 |
| mucopolisacaridosis IVA (MPS IVA) | 20 | 52 |
| MPS VII (déficit beta-D-glucuronidasa) | 10 | 45 |
| GM1 gangliosidosis | 20 | 127 |
| lipofuscinosis ceroides neuronal infantil tardía (LINCL) | 10 | 30 |
| leucodistrofia metacromática | 20 | 127 |
| MPS IIIA (déficit de hexosaminidasa) | 20 | 127 |
| Niemann-Pick B (déficit de esfingomielinasa) | 10 | 30 |
| deficiencia de lipasa ácida lisosomal | 15 | 30 |

Adaptado de Miyamoto & Kakkis, 2011.

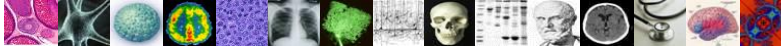
diego.rosselli@gmail.com



12/21

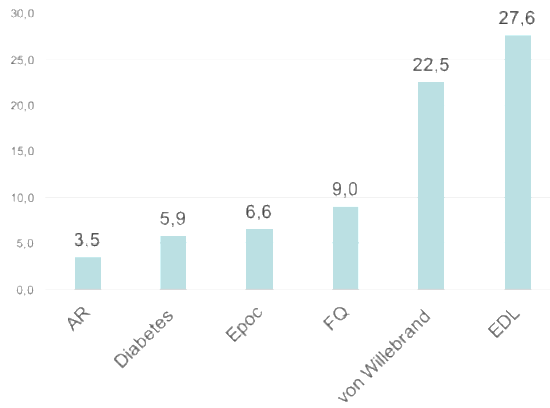


diego.rosselli@gmail.com

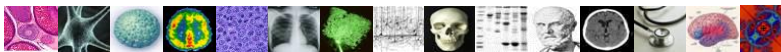


13/21

Porcentaje de casos clínicos



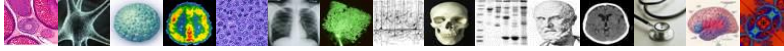
diego.rosselli@gmail.com



14/21



diego.rosselli@gmail.com



15/21



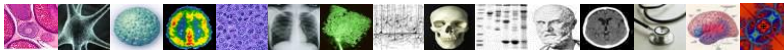
diego.rosselli@gmail.com



diego.rosselli@gmail.com



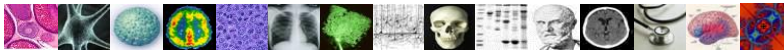
diego.rosselli@gmail.com



18/21



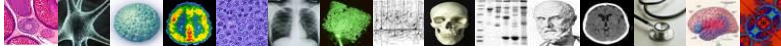
diego.rosselli@gmail.com



20/21



diego.rosselli@gmail.com



21/21

Es fácil tomar una decisión
desde un escritorio en la
capital, sin mirar a los
pacientes a los ojos.

diego.rosselli@gmail.com