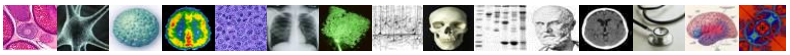

Los Desafíos de las Enfermedades Raras

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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, Elli Lilly, Genzyme, Grünenthal, GSK, Janssen, Lafrancol, Linde, Merck, MSD, Mundipharma, Novartis, Novo Nordisk, Pfizer, Roche, Sanofi-Aventis, Sanofi-Pasteur, Shire, Stendhal, Takeda, Zambon

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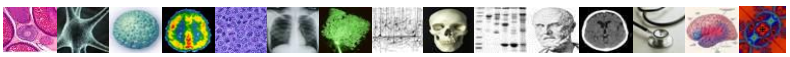

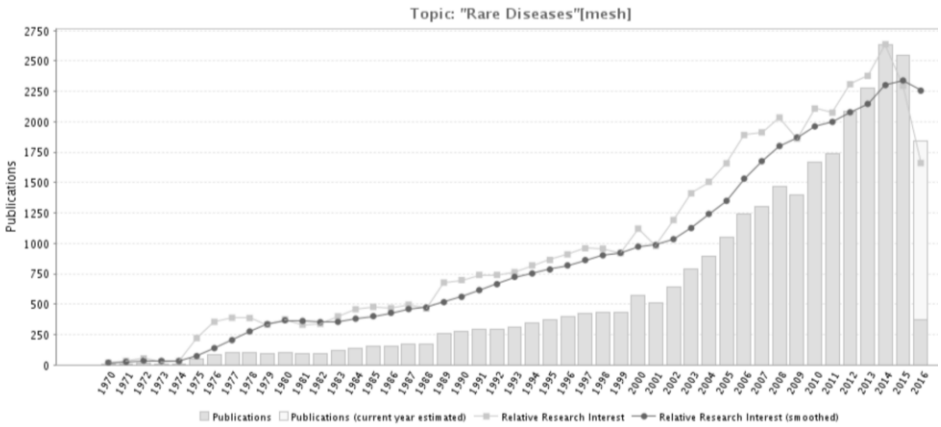

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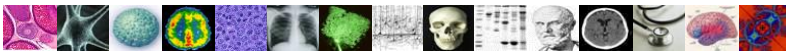

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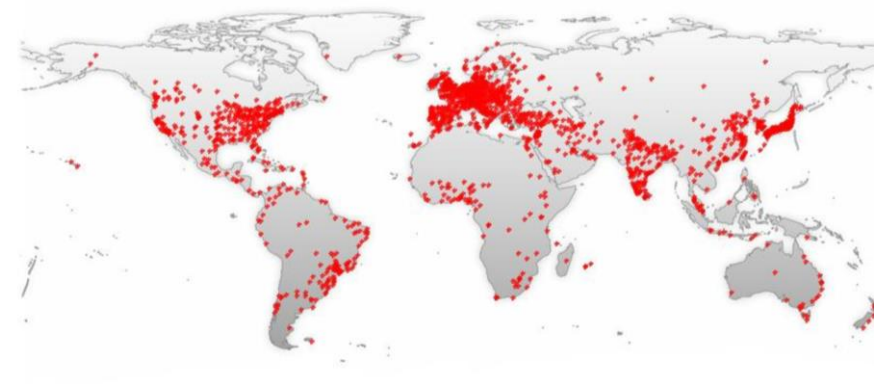

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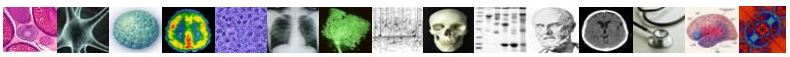

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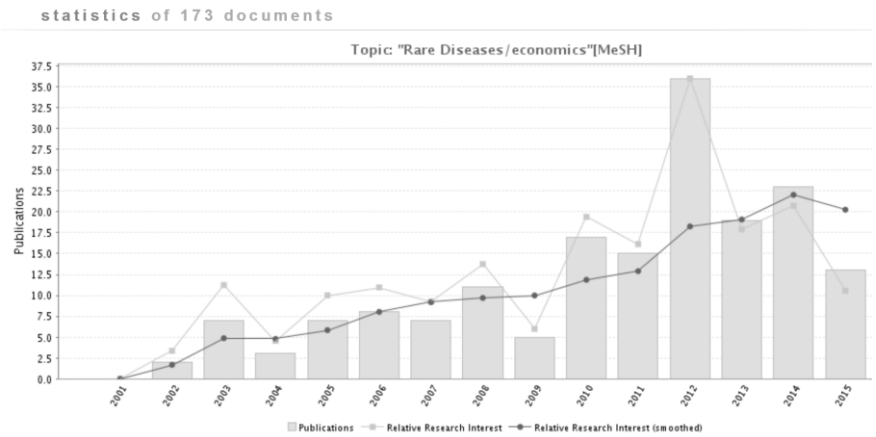

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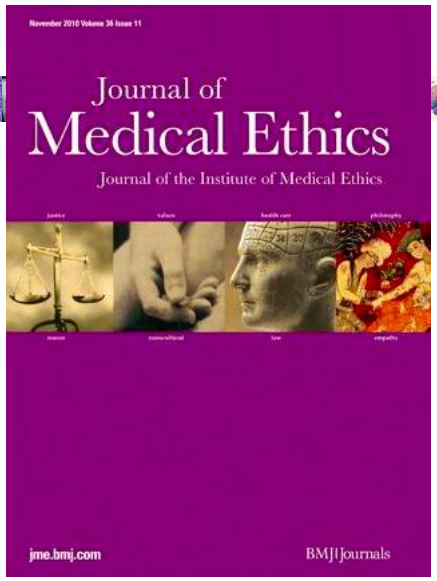

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Brief report

OPEN ACCESS

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

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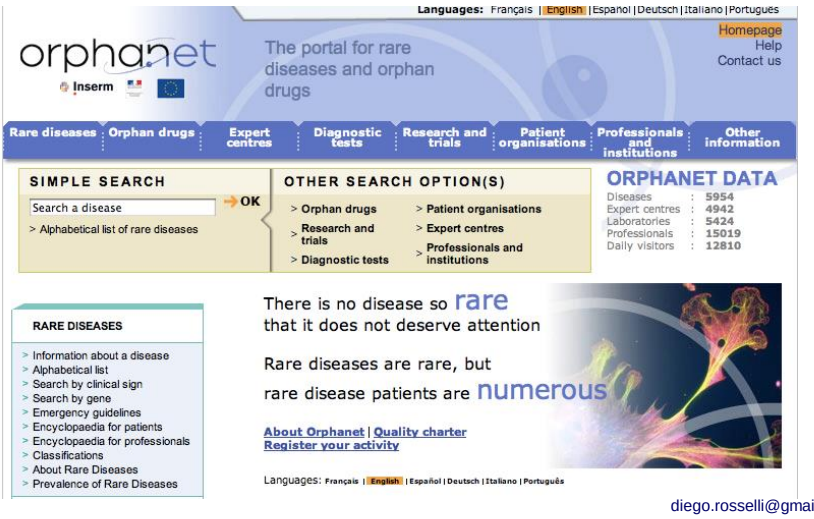
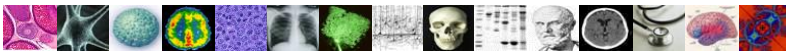
ABSTRACT
Mucopolysaccharidosis VI is an autosomal recessive lysosomal storage disorder associated with severe disability and premature death. The presence of a mucopolysaccharidosis-like disease in indigenous ethnic groups in Colombia can be inferred from archaeological findings. There are several indigenous 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-





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	Organizaciòn Mundial de la Salud
Suecia	10	Swedish National Board of Health and Welfare
Turquía	1	Ministry of Health
Unión Europea	50	Regulation ECn141/2000

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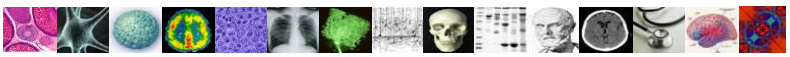

La investigación en enfermedades raras:

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

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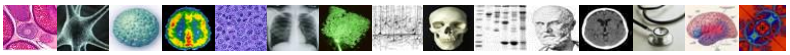

Tabla 3. Ejemplos de tamaños de muestra de estudios clínicos en enfermedades de depósito lisosomal.

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-glucoronidasa)	10	45
GM1 gangliosidosis	20	127
lipofuscinosis ceroide neuronal infantil tardía (LINCL)	10	30
leucodistrofía 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.

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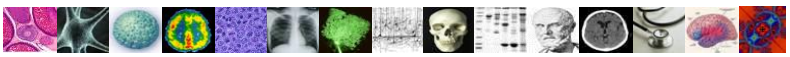

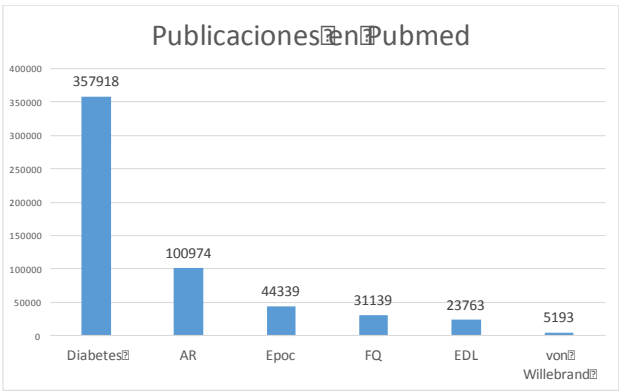

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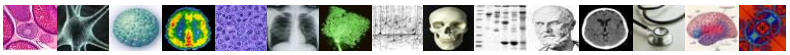

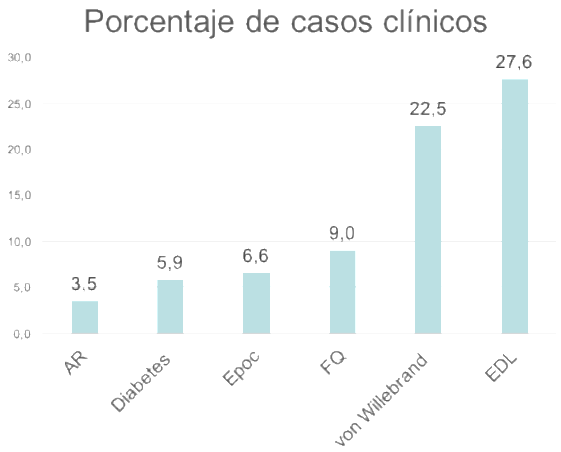

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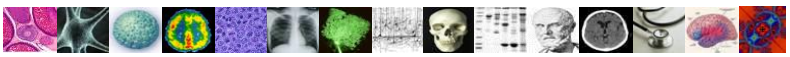

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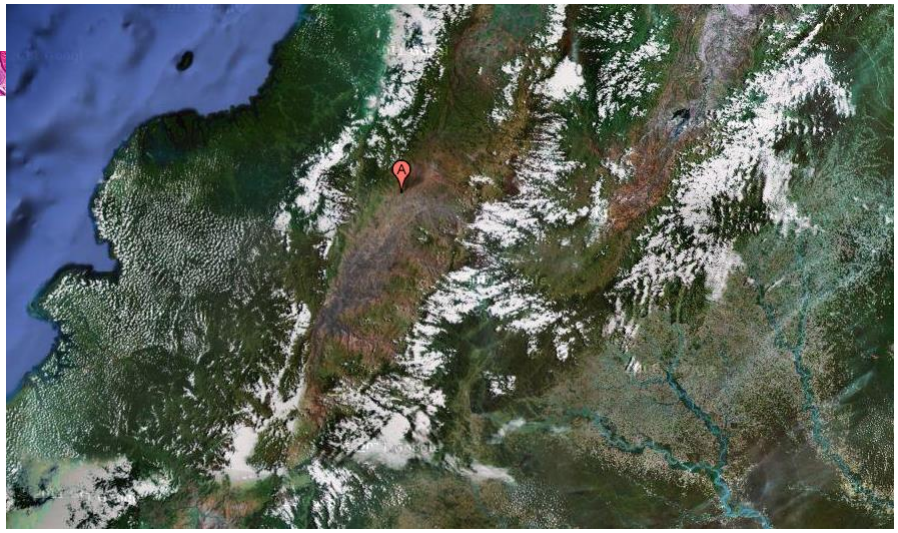

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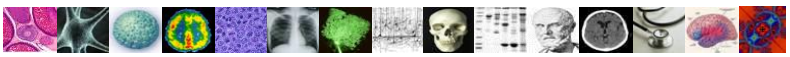

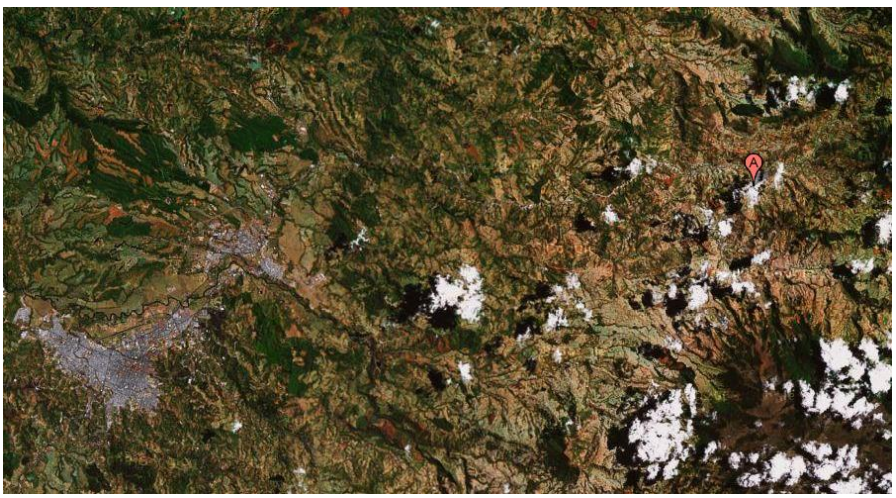

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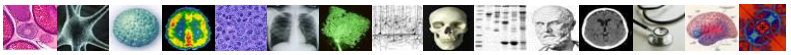

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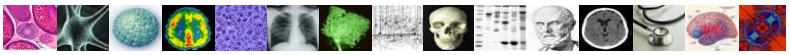

Es fácil tomar una decisión desde un escritorio en la capital, sin mirar a los pacientes a los ojos.

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