Hypercholesterolemia and FH
Public Policy, Success Stories

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Iberoamerican FH Network

www.colesterolfamiliar.org

Spanish FH Foundation

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- Non profit organization devoted to FH patients and family members
- Created in 1997 by patients and healthcare professionals
- Raising awareness to place FH on the social and political Agenda to save lives
- Influencing health care providers to implement detection and treatment
- Reimbursement for chronic statin therapy in 2003 & ezetimibe in 2008/Parliament
- Reimbursement for PCSK9-I and LDL-Apheresis
- National FH Registry (SAFEHEART) started in 2004. Cascade screening program
- Guidelines for FH and FCH
- National Detection Program for FH (genetic testing)
- Economic Study of a National Detection Program for FH
- FH Risk Equation for predicting incident CVD events

CDC Evidence-based Classification of Genomic Test:

A Growing Number os Applications Ready for Prime Time

Tier 1	Supported by a base of synthesized evidence for implementation in practice	e.g; Newborn Screening, HBOC, Lynch syndrome, **Familial Hypercholesterolemia**
Tier 2	Synthesized evidence is insufficient to support routine implementation in practice; may provide information for informed decision making	e.g; many pharmacogenomic tests
Tier 3	Evidence-based recommendations against use, or no revelant synthesized evidence identified; not ready for routine implementation in practice	e.g; direct-to-consumer personal genomic tests

Dotson WD, Douglas MP, Kolor K, et al. *Clin Pharmacol Ther. 2014 Apr;95(4):394-402.*
list of applications by level of evidence on CDC public health genomics knowledge Base website:
https://phgkb.cdc.gov/GAPPKB/topicStartPage.do

Cost-Effectiveness of a FHDP in Spain

- Horizon 10 years: 2017-2026. **The design simulates a prospective cohort study**
- Genetic detection of 9.000 FH cases/year (2.250 IC and 6.750 relatives), ratio1:3
- Two perspectives were considered: **the payer and the social perspective**
- Participation of GP's is crucial

IF FHDP was applied in Spain, and 9.000 FH cases were detected in a year, whitin the next ten years, we could avoid:

- 847 Coronary Events (Mis+CPs)
- 203 Coronary Deaths
- > 200.000 days of work productivity lost
- Produce 767 QALYs
- **For each 6 FH (>18 years) detected and treated we avoid 1 fatal or no fatal Coronary Event**

Lázaro P.…Mata P. J Clin Lipidol 2017;11:260-71

FH in Latin America
Health and Social impact

- Population: 625 Million
- Prevalence: ±1/250: **2.500.000 million with FH**
- Premature CAD represent an important economic burden to the Healthcare systems

AVOID EVENTS/Year:

- Spain Model (FH Detection Program): **105 coronary events/year for 9.000 patients**
- Within the next 10 years, we could avoid:
- **≈ 300.000 coronary events (20-25% will die)**
- Coronary events occur in working age or in young people
- A Detection Strategy:
  - Decreases CV events and improves quality of life
  - Reduce costs and loss of labor productivity
  - **Important Health gain and money savings for the society and also save lives**

Adapted from Lázaro P….Mata P. J Clin Lipidol 2017;11:260-71

Iberoamerican FH Network

- Established in 2013 with 8 countries (Spain, Portugal, Argentina, Brazil, Chile, Colombia, Mexico and Uruguay)
- > 2 million with FH
- FH is underdiagnosed and undertreated
- IBA countries share centuries of common history

- **AIMS:**
- To increase awareness of FH
- To promote family FH cascade screening
- To develop a globlal collaboration and care for FH
- To establish a quality registry
- To share data and management of this disease
- Call to Action to put FH on the Public Health Agenda

IBA FH Network: Achievements

- > 10.000 FH patients with genetic diagnosis
- National genetic screening programs: Spain and Uruguay
- Regional detection program: Brazil (Sao Paulo) and Argentina
- Genetic testing started in most of the countries
- Patient organizations: Spain, Uruguay, Brazil, Mexico, Portugal…
- Share common registry: Spain, Brazil, Uruguay, Colombia, Chile..
- Homozygous FH Registry
- Coronary CT Imaging Evaluation of Subclinical Atheroescl: Brazil and Spain

Santos R et al. Journal Clin Lipidol 2017;11:160-66

Genetic Diagnosis

- Country identification rates: Spain 9%, Portugal 3%, Uruguay 2%
- LDLR mutations represent > 90% of FH patients
- APOB mut. represent 3.5% (Brazil & Spain); 5% in Portugal
- PCSK9 mutations represent < 1%. Only in Portugal and Spain
- In Spain, Portugal and Brazil 10-15 mutations represent 30-47% of all country FH cases
- Spain and Portugal share 5 of the 10 most common mutations

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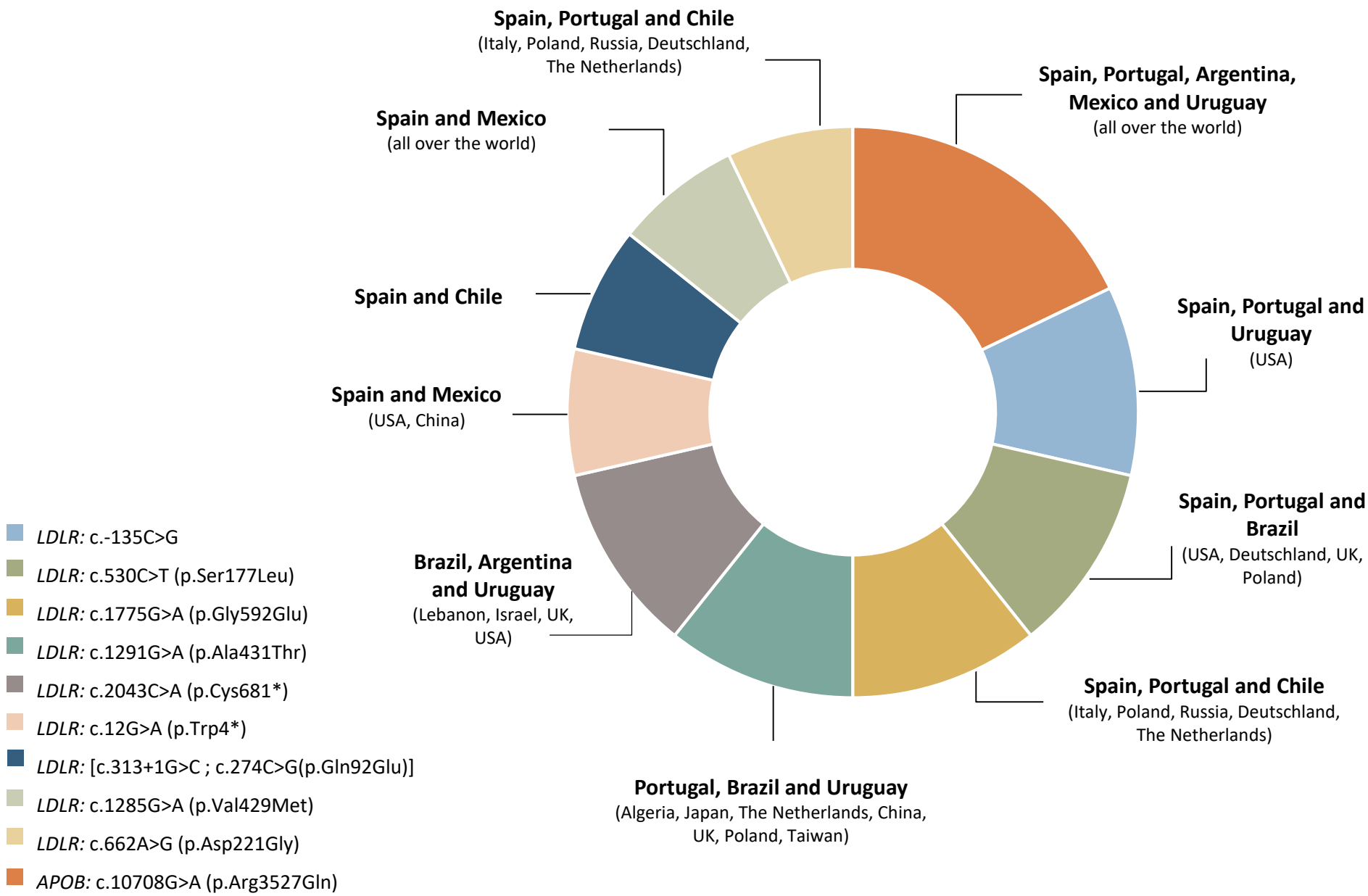

Figure 1. Most common mutations in IBA FH network (in the 7 countries that presently form the network), based on the 10 most common mutations in each country. Only mutations shared by at least by 2 countries are represented.

Santos R et al. Journal Clin Lipidol 2017;11:160-66

FH and the Future

- In this photo you can see two Spanish citizens diagnosed with FH
- Unfortunately many worlwide citizens are not aware that they have the disease, until it is too late to reverse
- These people, and their families are relying on us to find and diagnose them
- It is in our hands to support a plan for the early detection of FH
- We are ready to work with you to achieve this goal
- Every child deserve high quality care

Un largo camino por recorrer,
que ya hemos iniciado