

# Cross Border Genetic Testing for Rare Diseases

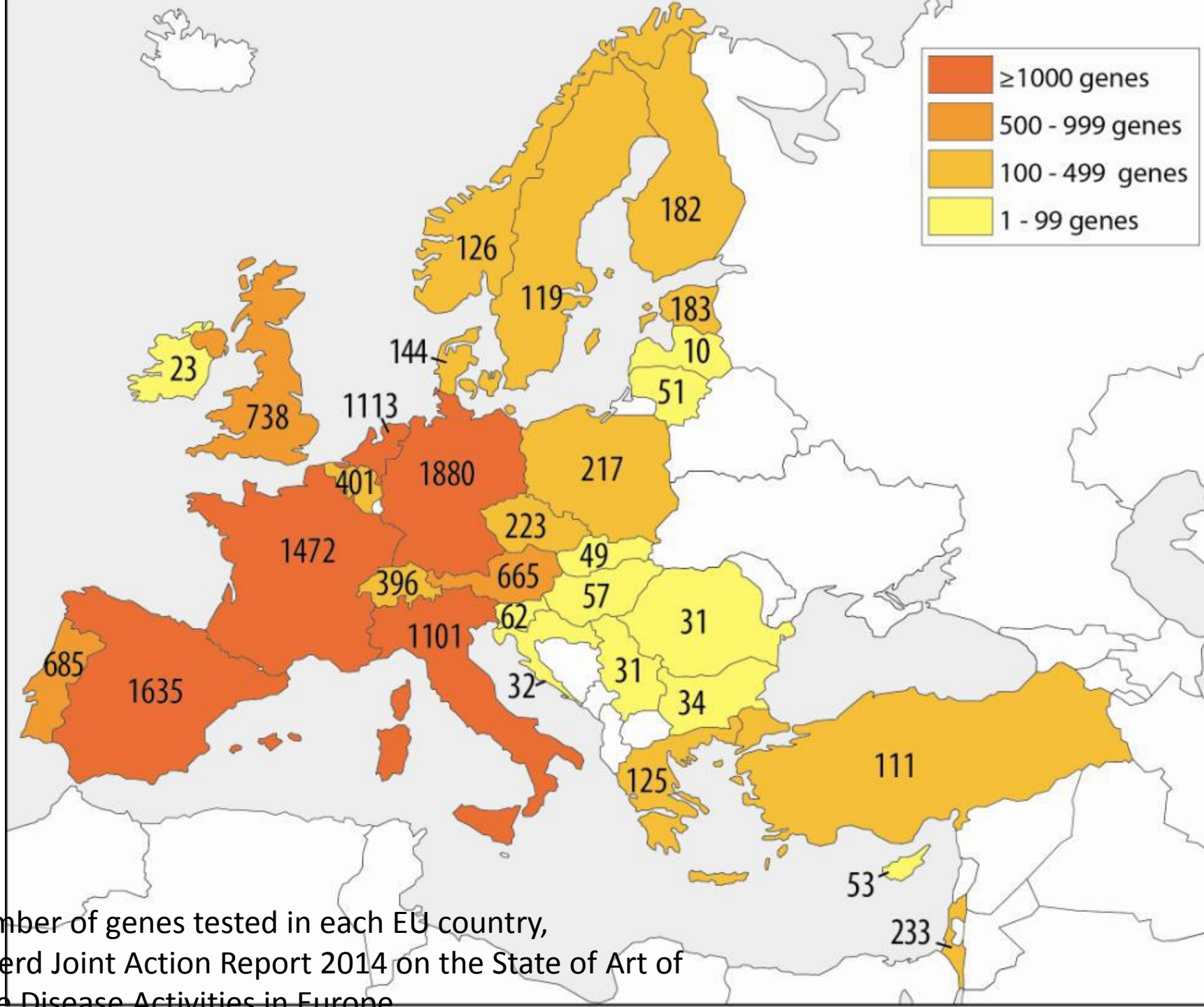
EUCERD Joint Action WP8

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# Starting point

- Possibilities and demand for genetic testing have increased rapidly.
- Most EU-countries do not perform all testing that would be needed.
- There were rumours but no data about different problems, inequalities and challenges related to cross border testing.



Number of genes tested in each EU country,  
Eucerd Joint Action Report 2014 on the State of Art of  
Rare Disease Activities in Europe

# Aims for the EJA Survey

- To investigate the experiences of the laboratories and genetics clinics on cross border genetic testing.
- Areas on interest: the volume, the reasons for purchasing tests abroad, funding, the possible problems etc.
- To collect suggestions on how the identified problems of cross-border testing could be overcome, i.e. problems leading to unequal access of genetic testing.

# Final aim

- To improve access to (genetic) diagnostics for all rare diseases in all EU countries

# Methods

- Survey targeted to molecular genetic testing laboratories and genetic clinics in the EU countries, with the help of Orphanet
- Questionnaire data collected between January 7<sup>th</sup> and March 4<sup>th</sup> 2014
- Phone interviews between February 3<sup>rd</sup> and March 13<sup>th</sup>
  - Selected respondents from different MS (five laboratories/four clinics)

# Respondents

- 170 responses from laboratories (11%), 105 responses from clinics (17%)
- Laboratories: Public 77, Academic 43, Research 25, Private 19 and Others 6; no responses from Slovenia, Luxembourg and Lithuania
- Clinics: no responses from Luxembourg, Hungary, Greece and Estonia

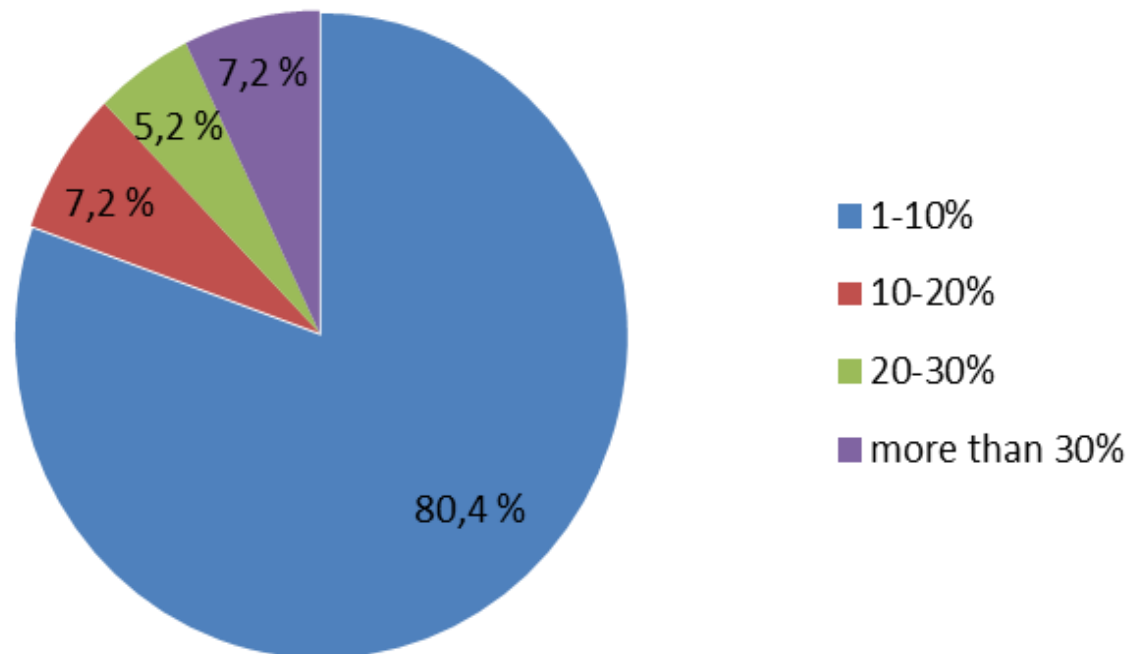
Laboratories		Counselling clinics	
Italy	34	United Kingdom	36
France	25	Italy	11
Spain	23	Spain	10
Germany	19	Portugal	5
United Kingdom	15	Germany	4
Greece	5	France	4
Austria	5	Belgium	4
Portugal	4	Lithuania	3
Netherlands	4	Ireland	3
Hungary	4	Finland	3
Finland	4	Sweden	2
Belgium	4	Romania	2
Poland	3	Poland	2
Czech Republic	3	Netherlands	2
Sweden	2	Malta	2
Slovakia	2	Croatia	2
Romania	2	Bulgaria	2
Estonia	2	Austria	2
Denmark	2	Slovenia	1
Cyprus	2	Slovakia	1
Bulgaria	2	Latvia	1
Malta	1	Denmark	1
Latvia	1	Czech Republic	1
Ireland	1	Cyprus	1
Croatia	1	Luxembourg	0
Slovenia	0	Hungary	0
Luxembourg	0	Greece	0
Lithuania	0	Estonia	0



# Impressions from the Survey

- Estimated number of all rare disease samples tested in the EU 2013: 2,5 million (excluding screening tests)
- Estimated number of cross border samples: >90 000 samples (based on the number samples sent by laboratories)

## The percentage of cross border tests of all tests in the counselling clinics



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Every year, nearly 500,000 patients are treated at HUS hospitals. Patients requiring advanced specialist medical care are admitted across Finland. Every patient is entitled to high-quality care delivered in time.

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Suomeksi | På svenska



genetics

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**In Genetics Clinic,  
about 50% of testing  
is bought from abroad,  
often UK, the Netherlands  
or Germany**

Providing the best  
possible care

Every year, over 100,000 patients are treated at HUS hospitals. Patients requiring specialized medical care are treated across Finland. Every patient is entitled to high-quality care delivered in time.



> Hospital  
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## Pricing

### Example Reports

[Pan Cardiomyopathy Panel](#)

[Arrhythmia Panel](#)

### Family Member Testing

### Product Catalogue 2014 – PDF

### Cardiovascular Disorders

[Hyperlipidemia Panel – 10 genes](#)

[Pan Cardiomyopathy Panel – 103 genes](#)

[Core Cardiomyopathy Panel – 72 genes](#)

[Heart Panel – 133 genes](#)

[Noonan Sdr Panel – 12 genes](#)

## Genetic Tests

Blueprint Genetics provides rapid, comprehensive and up-to-date genetic testing for clinical medicine. By selecting your area of specialty from the provided categories, find detailed information on the provided tests.

Our diagnostics is based on the novel targeted next generation sequencing technology developed at Stanford University, USA (Myllykangas et al. 2011) and interpretation tools. BpG has implemented a rigorous interpretation process. Our genetic testing service is marked by a special ethical responsibility. We adhere to the highest diagnostic standards for our customers. All NGS panels are tailored to the specific clinical boundaries and known mutations outside the exon or exon-intron regions. Our tests are built on high quality sequencing data as we do not de

About 75%  
of the samples  
come from abroad

[Pricing](#)[Example Reports](#)[Pan Cardiomyopathy Panel](#)[Arrhythmia Panel](#)[Family Member Testing](#)[Product Catalogue 2014 – PDF](#)[Cardiovascular Disorders](#)[Hyperlipidemia Panel – 10 genes](#)[Pan Cardiomyopathy Panel – 103 genes](#)[Core Cardiomyopathy Panel – 72 genes](#)[Heart Panel – 133 genes](#)[Noonan Sdr Panel – 12 genes](#)[Genetic Tests](#)

Blueprint Genetics provides rapid, comprehensive and up-to-date genetic testing services in the field of clinical medicine. By selecting your area of specialty from the provided list, you can access a range of diagnostic tests, find detailed information on the provided tests and their clinical utility.

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# Results: selected examples

- The most common reason for **sending samples** abroad was because the test was not performed in home country
- **Orphanet**, Genetest, recommendation and reputation are the most important means of finding a testing laboratory
- **Quality, reputation and price** are the most important selection criteria
- Laboratories have **difficulties in collecting payments** from abroad
- Different MS have **variable funding** sources for cross border tests
- Permission for cross border testing requires **long processes** in some MS
- In general: **differences between the MS are huge** when it comes to allowing/financing cross border testing





## Clinician

To find a high quality laboratory performing the test needed

To get permission to send the sample

Consent process

To be able to understand the report and trust the quality

Sending the sample, logistics

To get a high quality sample and referral

To take population related issues into account

To write an understandable report

To manage to collect the payment



## Laboratory



# Next steps?

- EJA Workshop on Cross Border testing 15-16.12.2014
- How to improve access for testing?
  - To make cross border testing easier by
    - find the price/scope of the testing more easily?
    - having similar consent requirements in EU?
    - simplifying bureaucracy?
    - simplifying logistics?
    - having lab webpages in English?
  - To have high quality testing in every/most countries

Thank you!