

Cross border testing in Europe:

Several problems to be solved!

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Starting point

- There were several unknown issues related to cross border genetic testing in RD
 - The reasons to sell/buy tests cross borders?
 - The magnitude of the business?
 - Did the laboratories face some problems?
 - Or the clinicians?
 - Is there any equality in access to genetic testing in EU?

Methods

- We performed (January-March 2014) a Survey targeted to molecular genetic testing laboratories and genetic counselling clinics in the EU Member States (MS)
- They were approached
 - via Orphanet (only those registered to Orphanet)
 - via EuGT, National Human Genetic Societies /personal contacts
- Questionnaires (Webropol) and phone interviews for both groups
- Piloting/evaluation of the questions in collaboration with Eurogentest

Results

170 (11%)
responses from
laboratories

105 (17%)
responses from
clinics

Table 1. Number of respondents by country.

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

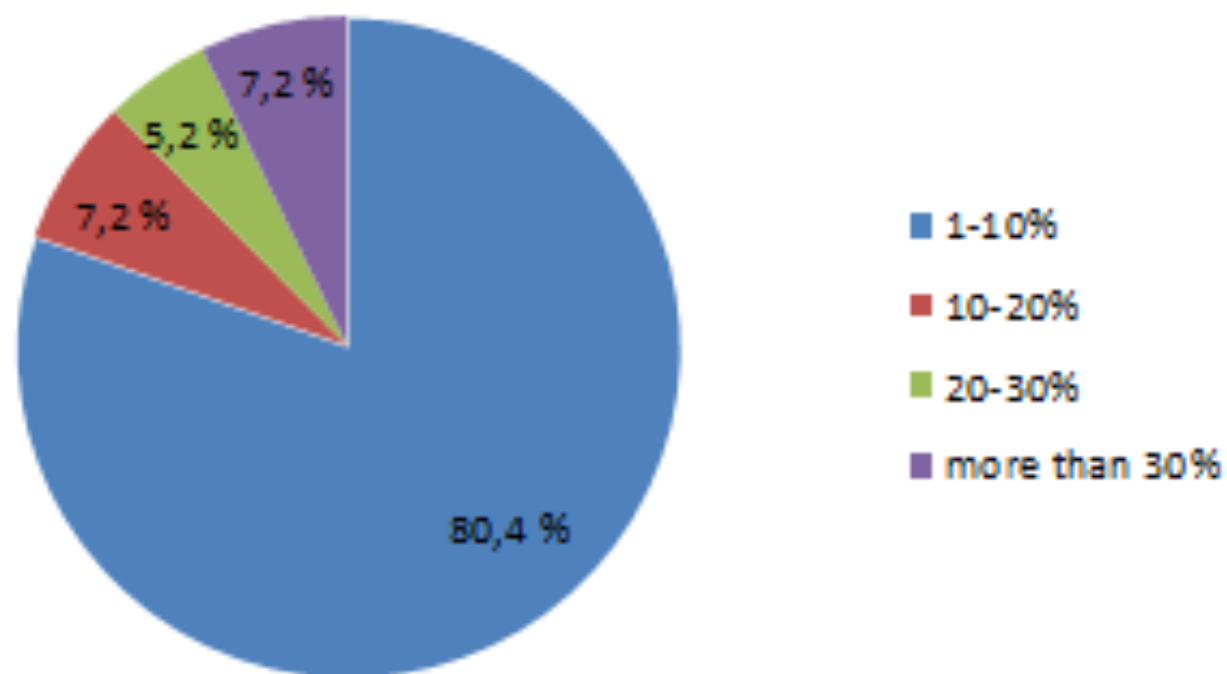
- Estimated number of RD cross border samples in the EU 2013: ~90 000 samples (based on sent samples by laboratories)

Table 3. Samples tested by laboratory type

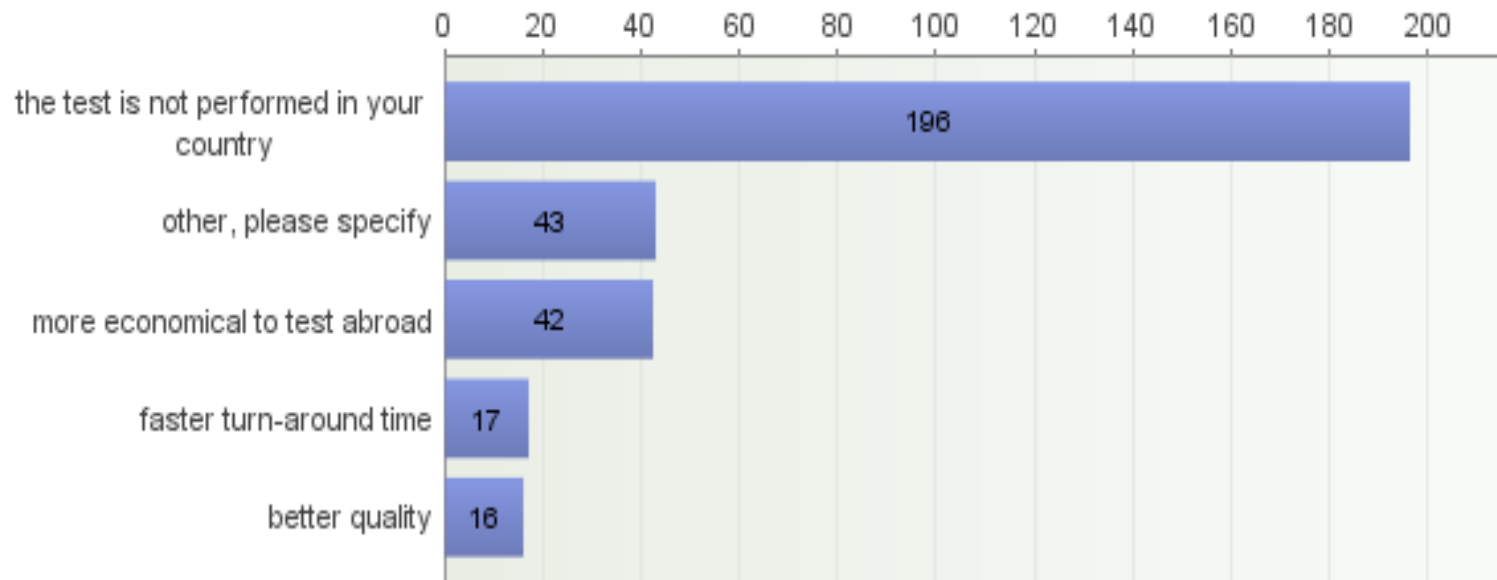
	Public (n=77)	Academic (n=43)	Research (n=25)	Private (n=19)	Other# (n=6)
All samples; mean	3286	1504	374	3230	822
All samples; median	500	239	50	180	463
Sent; mean	109	44	14	20	13
Sent; median	3	5	0*	10	5
Received; mean	24	84	42	70	7
Received; median	6	10	10	5	1

*18/25 sent 0 samples, #both public and academic or academic and research laboratories

Figure 2. The estimated percentage of cross border tests of all tests in the counselling clinics



- Cross border testing is most often offered on scientific interest and on courtesy basis
- The most common reason for sending samples abroad because the test was not performed in home country

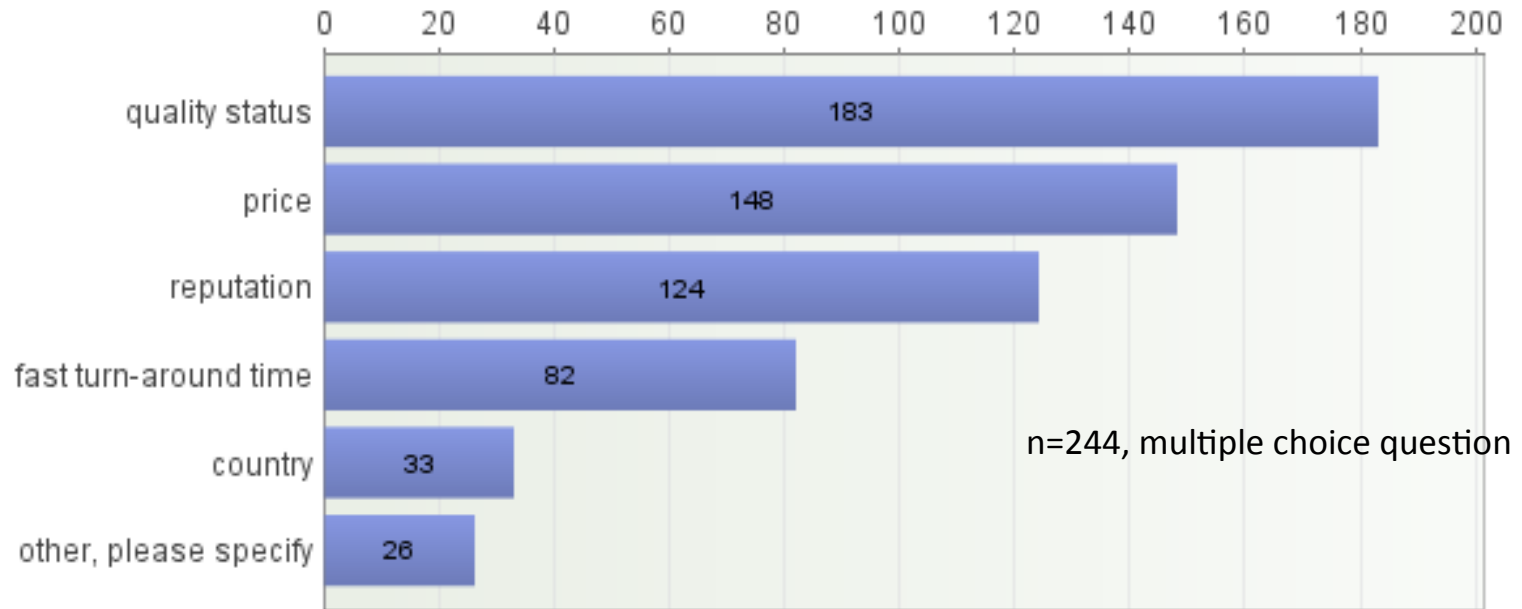


(other: for example subcontracting due to instrument breakage, collaborative projects, more comprehensive test available and the access to high throughput platform testing)

Results: quality

- Laboratories:
 - 22% reported that the customers often misunderstood the test coverage
 - 30% of labs experienced difficulties to decide if the requested test was predictive or diagnostic
 - 11% had problems in knowing whether pre-test counselling had been given prior to testing
- Clinics
 - Varying experiences about the informed consents
 - 19% experienced challenges in interpreting the results of cross order tests
 - 7% relied the results never or rarely

When selecting the test lab, quality was the first criteria



- 10% never used quality as a criterion
- Use of the brokers not common (2%)
- In half of the cases where results were returned with e-mail, unprotected mail was used

→ 13% experienced the test ordering easier from abroad than from home country

Major problems and worries

- Laboratories:
 - financial issues: costs of the tests, reimbursement and payment (mentioned in ~30% of the responses)
 - sending of biological samples cross borders
 - quality of genetic counselling
 - related bureaucracy
- Clinics:
 - high costs of cross border testing
 - different problems related to funding
 - quality of testing laboratories
 - difficulties in finding the testing laboratory

Respondent not well aware of National plans for RD

- 71% of the respondents from 23 different countries reported their country had a plan, 16% were uncertain if a plan existed and a further 13% replied their country did not have a plan (n=268)
- Respondents from most MS had discordant responses as one respondent stated, for instance, that there was a national plan while another reported that there was no such plan
- In addition, of those responding their country had a plan (n=186), it was unclear for 56% if the cross border testing was a specific issue in the plan and 54% were uncertain if the plan included a specific reimbursement plan for cross border samples



Picture: Thanks to Charlotte Rodwell
Eucerd Joint Action

Is there equality in genetic testing in EU/Europe?

- The answer is: NO
- In some countries cross border testing is not at all reimbursed (Romania) or very rarely (Poland) or only for children (Croatia) or the quality may be poor (Ireland)
- Some countries have practically all testing in their own supply (UK, France etc)
- Some countries easily allow cross border testing (Finland)

How to solve the problems?

- Eucerd Joint Action, ESHG and EuGT have partly shared goals in cross border testing.
- Joint Action focuses on RD and especially EU countries, EuGT focuses on quality issues, ESHG has a wider European focus and is also interested in non-RD genetics (like cancer etc).
- Maybe: a workshop aiming at a White Paper??
- Role of NHGSs?