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 Societes /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.
• 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

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

*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 of 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?