



Requirements for Reference Materials in European Genetic Testing Labs

Results from three European surveys

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Results & Feedback from surveys;

1. [NGRL Manchester](http://www.ngrl.org.uk/Manchester/Pages/NGRLMhome.htm) - opinions on RMs within the UK genetic testing community.
www.ngrl.org.uk/Manchester/Pages/NGRLMhome.htm
2. [CRMGEN](http://www.crmgen.org) - Survey of Demand for CRMs
www.crmgen.org
3. [EuroGentest](http://www.eurogentest.org) - survey of positive controls
www.eurogentest.org

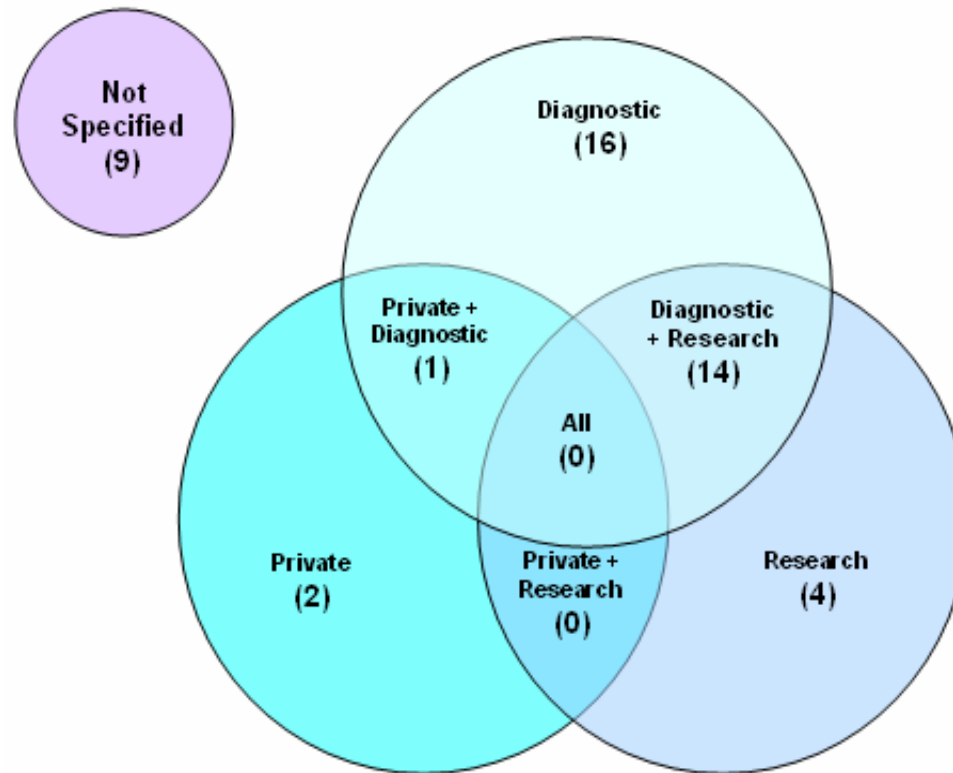


1. NGRL Manchester Survey

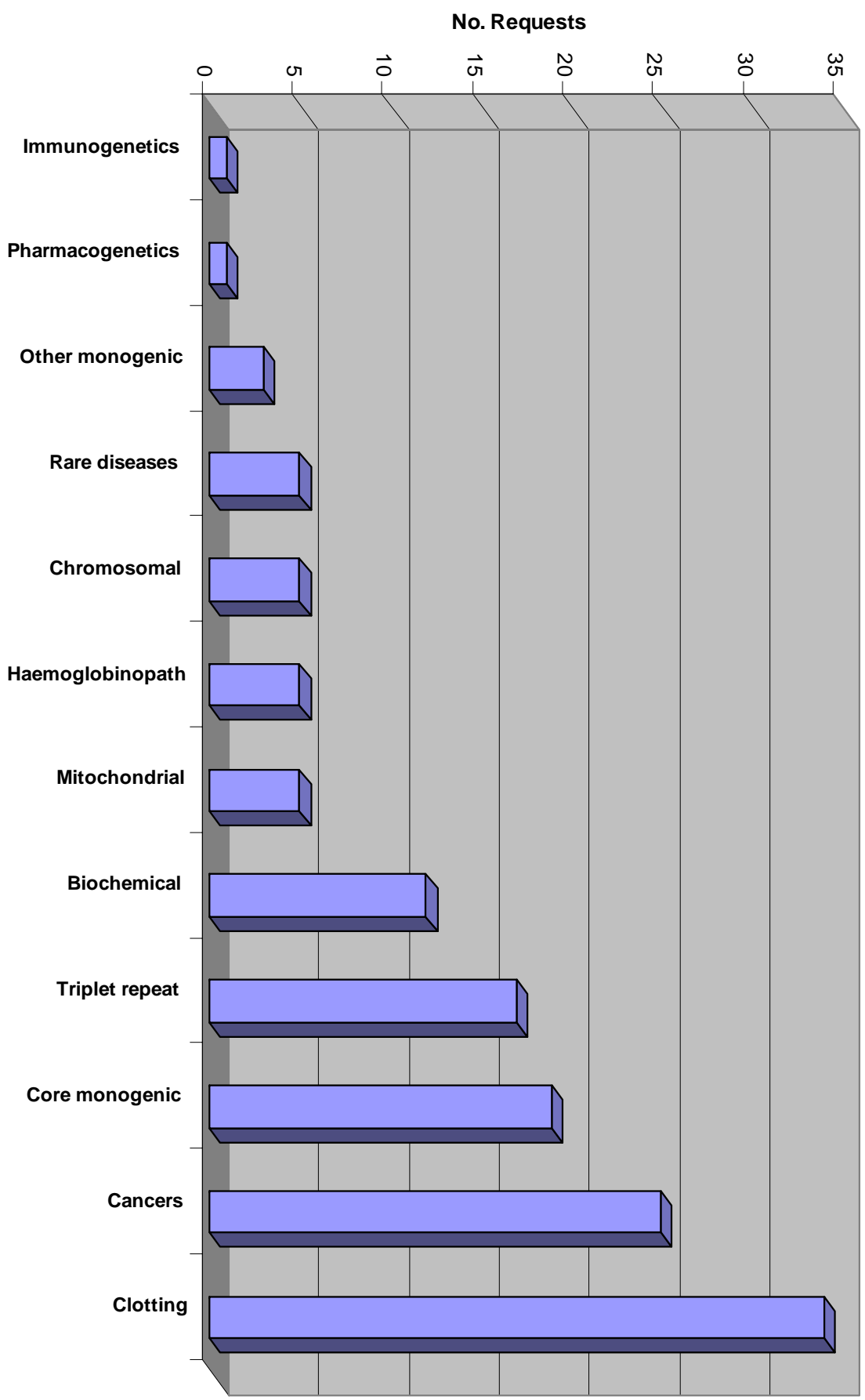
- Survey designed to seek opinions on RMs within the UK genetic testing community.
- Aim was to guide future RM development by the NGRLs and NIBSC.
- Available to download from the NGRL website www.ngrl.org.uk/Manchester

Distributed to 111 UK genetic testing labs

- 46 (41.5%) completed survey
- 17 (15.3%) not interested
- 48 (43.2%) no response

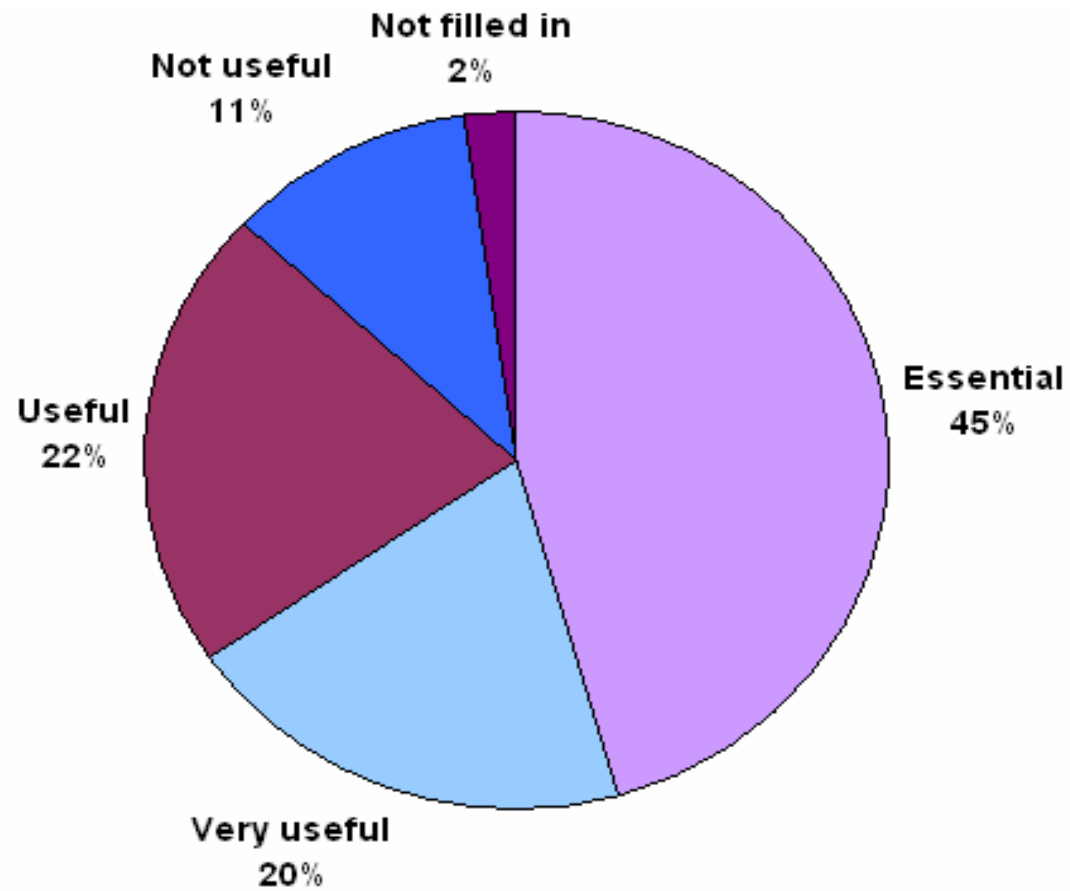


Requests to Develop RMs - Diseases



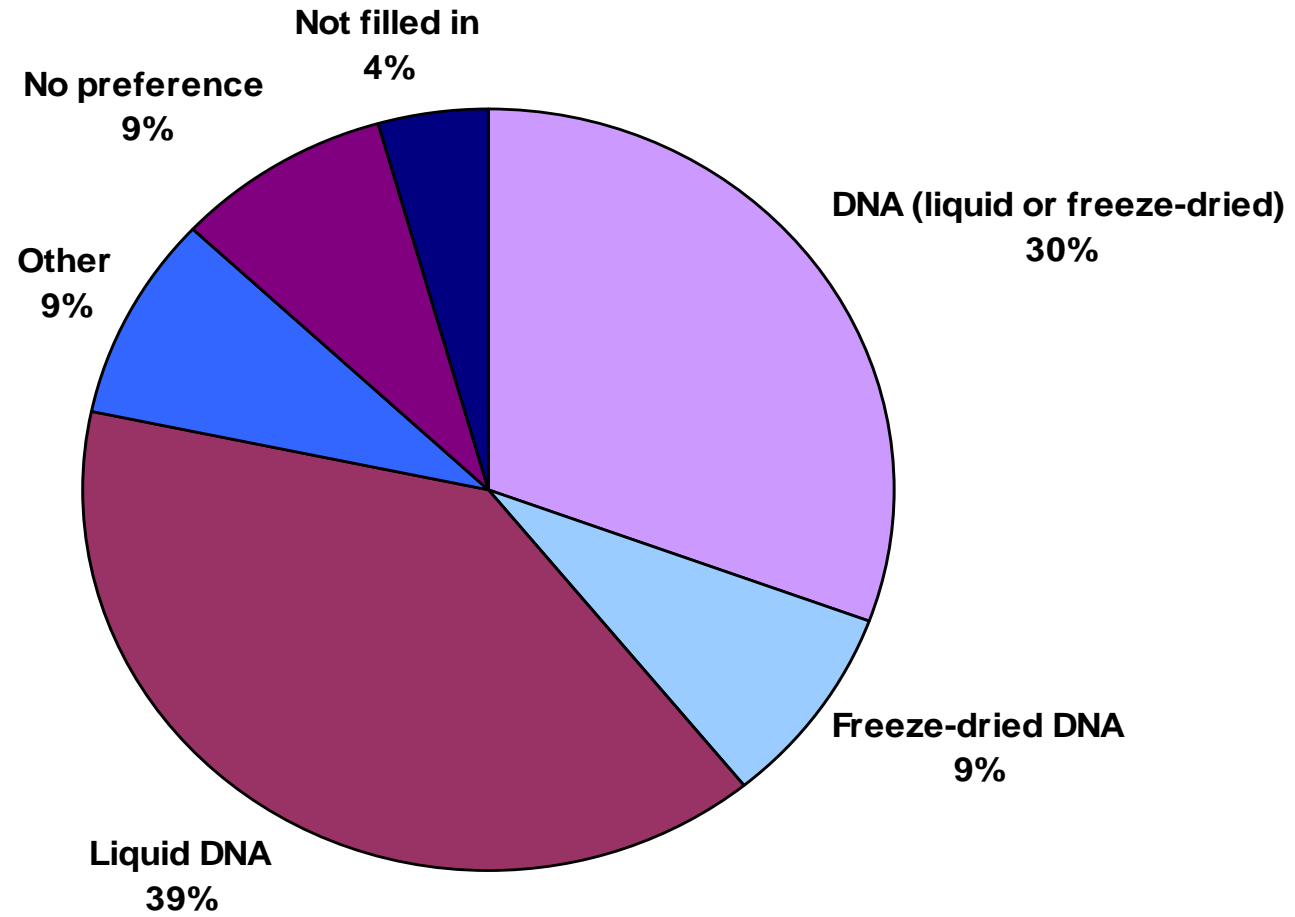


RM Usefulness

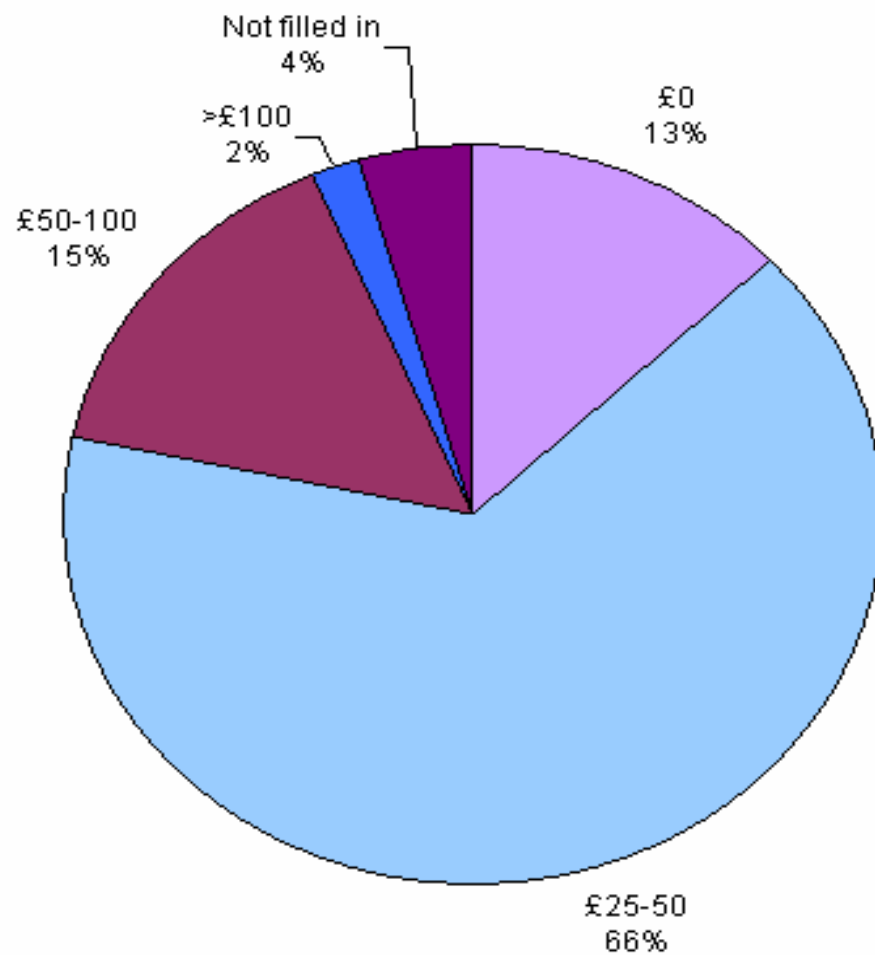




Preferred RM Format



How much would you be willing to pay for a Certified Reference Material (sufficient to run 100 control reactions)





Summary of NGRL Results

- Survey response rate was fairly good (42%)
- Most respondents were diagnostic labs and performed at least some molecular genetic testing.
- The most common requests for RM development were for:
 1. Clotting Disorders (Factor V def. Prothrombin mutations, Haemophilias)
 2. Cancers
 3. Core Diseases (CF, PWS)
- Vast majority thought RMs useful, very useful or essential for genetic testing.

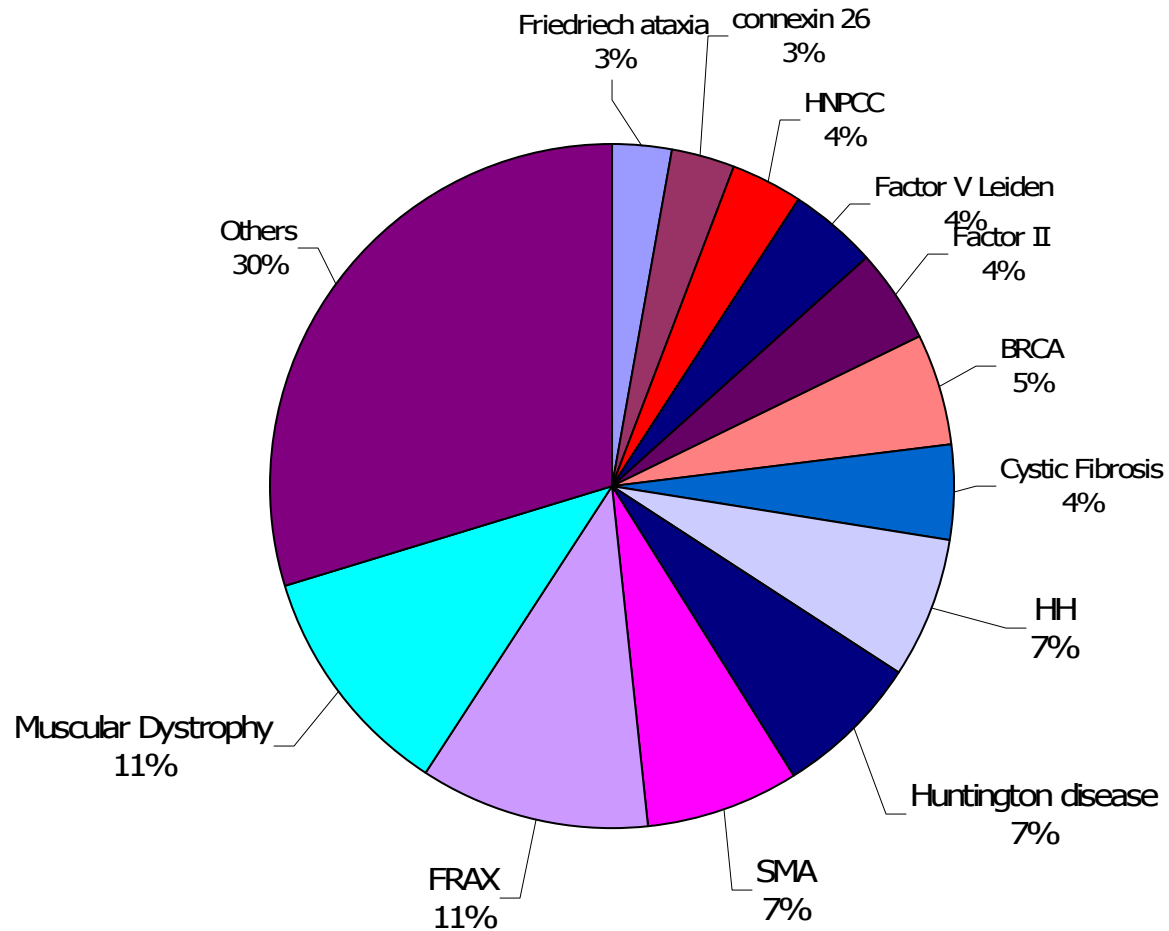


2. CRMGEN Dublin Survey

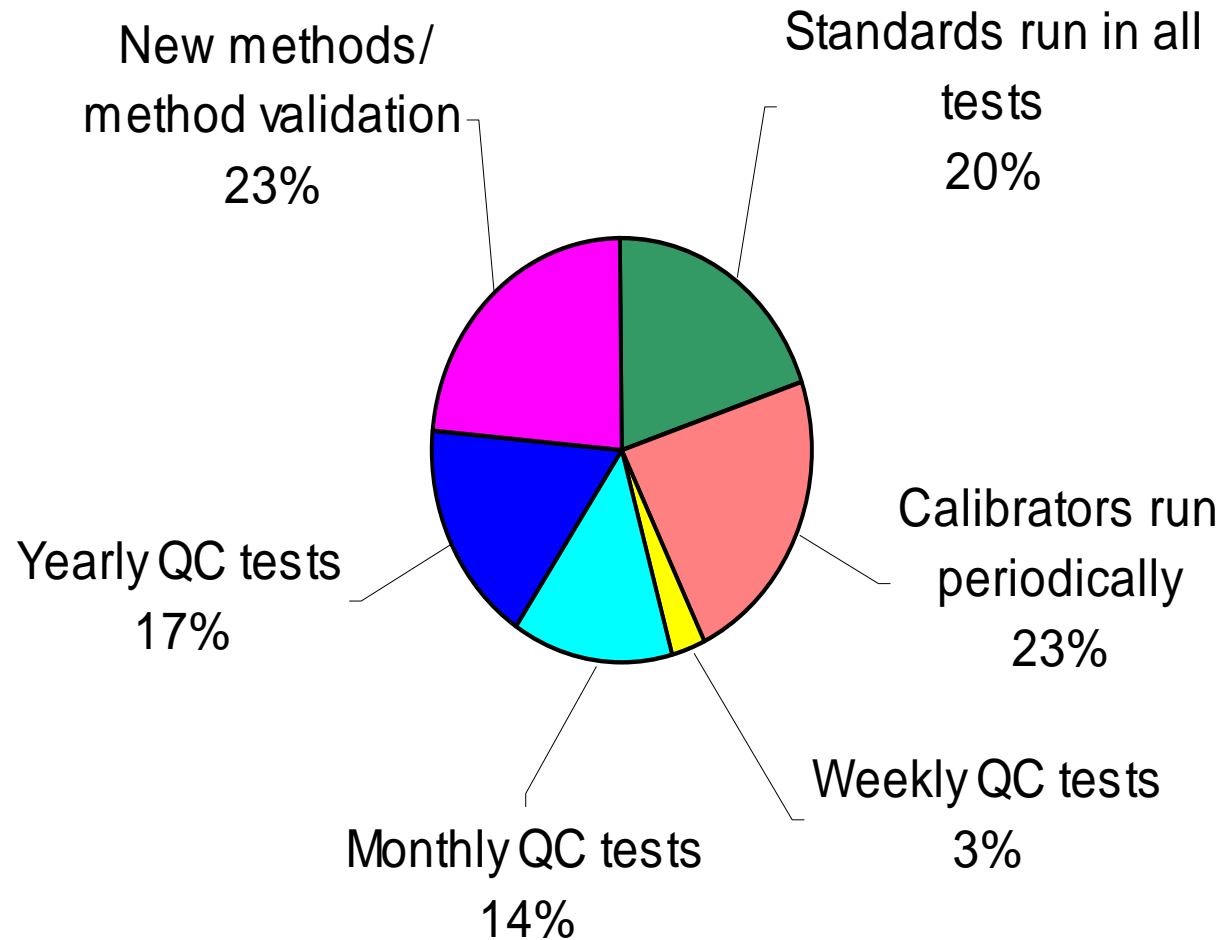
Survey of Demand for CRMs

- Survey distributed through EMQN network & on CRMGEN website www.crmgen.org
- 71 responses
- 24 countries represented
- Most respondents very supportive
- Responses are still coming through.

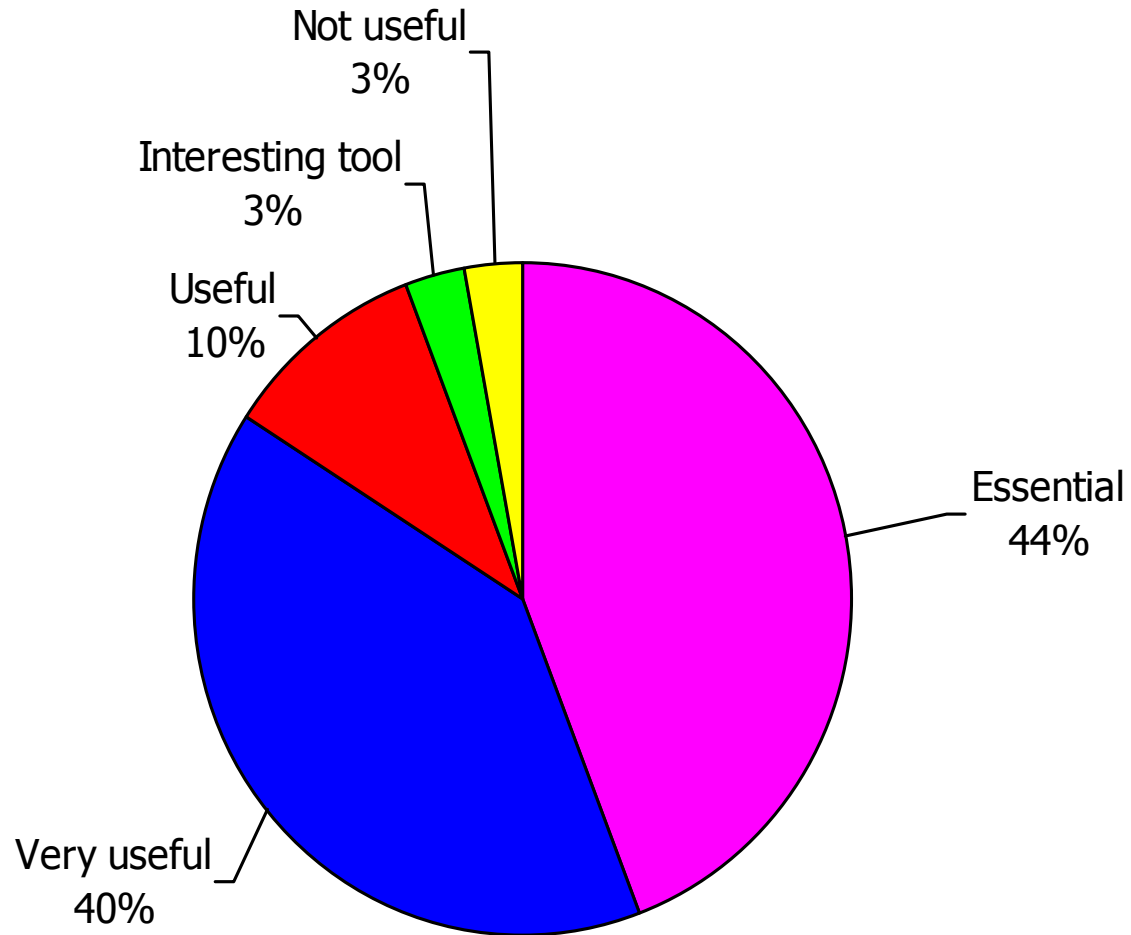
Nominate a Disease you would like to see CRM developed for:

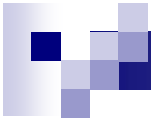


Once Certified Reference Materials become available, how frequently would you use them?

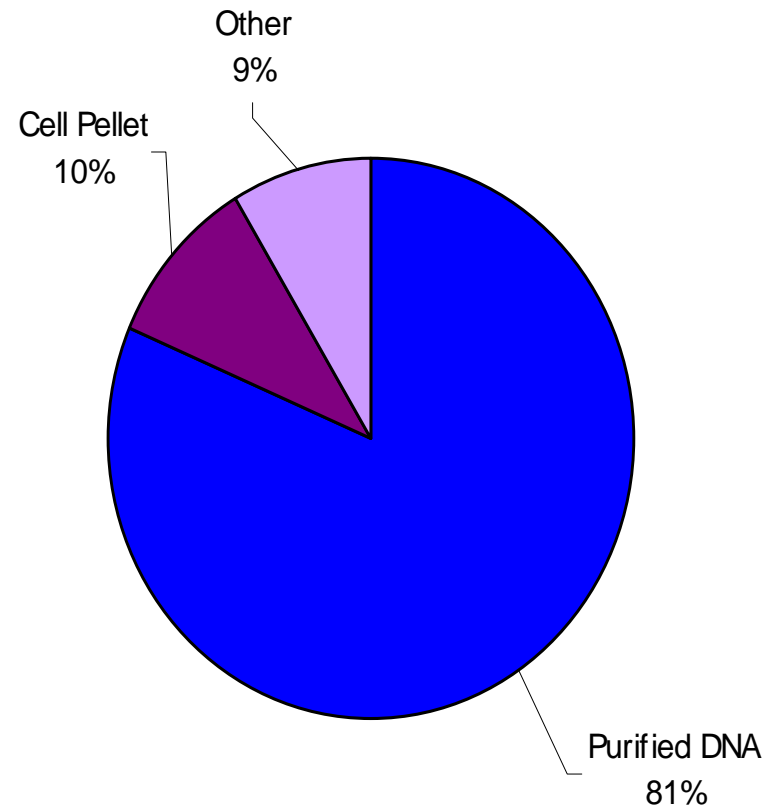


How useful do you consider CRMs?

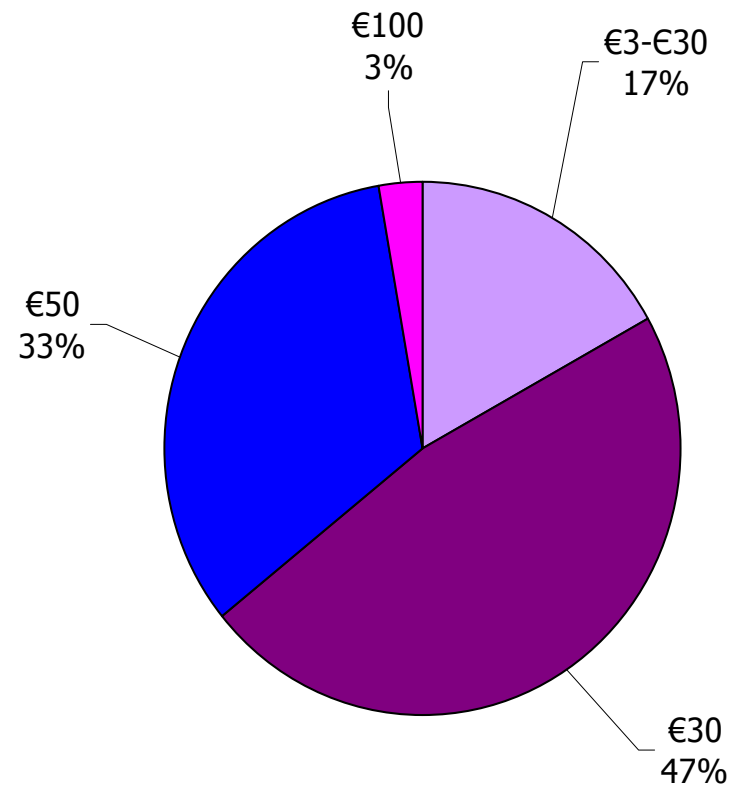




Preferred RM Format



How much would you be willing to pay for a Certified Reference Material (sufficient to run 3 assays)





Summary of CRMGEN Results

- Survey response rate was good - 71 laboratories in 24 countries.
- The most-requested RMs were for Fragile X syndrome, muscular disorders and Huntington disease.
- 82% were willing to pay between €30-€50 for 3 assays
- 83% of labs considered that CRMs are essential or very useful for their work.



3. WP1.6 - Survey of Positive Controls

Participants:

Assessors of EQA schemes run by the European Molecular Genetics Quality Network (EMQN)

Questions:

What types of controls do you use in your laboratory?

What controls would you like to have available?

- 19 Respondents
- 25 Diseases covered

“Open” nature of survey makes analysis difficult, but some themes emerge



Themes from Survey

What types of controls do you use in your laboratory?

- Most using “in-house” controls
- Not many rotating controls routinely
- Variation in practice for mutation scanning

What controls would you like to have available?

- “no need for other controls than the current ones”
- “as many as possible”
- Rare, hard-to-find controls



From these 3 Surveys; Conclusions

- Wide variation in current practices
- Wide variety of perceived needs for RMs
- Diverse opinions on which RMs are most needed
- Difficult to assess demand
- Divergence: Rare, hard-to-find mutations Vs frequently-used tests

Prioritization will not be easy!



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