EuroGentest: Reference Materials for Genetic Testing

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Abstract

The lack of currently available reference materials (RMs) for most genetic tests causes difficulties in validating and developing new assays, and results in tests being run without proper quality controls. On the other hand, the lack of accreditation of laboratories performing genetic testing does not create a large market for RM producers and this situation creates a vicious circle. EuroGentest is addressing this issue by defining the present and future needs for RMs, setting priorities for and supporting the development of new RMs and building an enduring network involving all stakeholders in RM development.

General Objective:

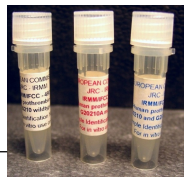
To promote the development and use of Reference Materials in Genetic Testing.

Aims:

- ✓ Promote the networking of developers and users of Reference Materials (RMs) to identify needs
- ✓ Develop a priority ranking of RMs required
- ✓ Support the development and the use of (new) RMs for Genetic Testing
- ✓ Inform stakeholders of the implications of the IVD Directive for Genetic Tests

Ongoing Work:

- ✓ Two International Symposia on RMs in Genetic Testing organised (Geel 2005 and Dublin 2007).
- ✓ Prioritization score for developed and publicized (see table below)
- ✓ Database of available RMs assembled and made available on EuroGentest web site
- ✓ Financial and logistical support to NIBSC UK for the development of a panel of RMs for Prader-Willi and Angelman syndromes
- ✓ Participation in the development and characterization of WHO panels for Fragile X syndrome and hereditary haemochromatosis
- ✓ Field trials organised with EU CF network for 2 different commercial multiplex RMs for cystic fibrosis testing
- ✓ Development of a guidance document for the use of RMs in Genetic Testing
- ✓ Proficiency testing study for Factor II mutations organised and evaluated
- ✓ Three IRMM/IFCC Certified Reference Materials for prothrombin G20210A mutation produced
- ✓ Workshop on the implications of the IVD Directive organised (Leuven, April 2007)



Plans:

- ✓ Publish reports on recently-held meetings
- ✓ Organize a third International Symposium on RMs for GT
- ✓ Publish guidance document for the use of RMs in GT
- ✓ Organize further trials of CF multiplex RMs
- ✓ Identify the needs of RMs for Cytogenetics
- ✓ Collaborate in the development of new RMs for SCAs and FRDA
- ✓ Develop a sustainable network to promote the aims listed above

Priority Ranking of RMs to be developed

Rank	Disease	Being addressed by*
1	FRAX	NIBSC/CRMGEN/NGRL, CDC
2	CF	CDC/Coriell, MMQCI, others
3	FVL/FII	NIBSC, CDC/ IRMM
4	BRCA	NGRL Wessex
5	HH	NIBSC, CDC/Coriell
6	HNPCC	NIBSC/CRMGEN/NGRL
7	PWS/AS	NIBSC/EuroGentest
8	DM	NGRL Wessex
9	HD	CDC/Coriell, NIST
10	DMD/BMD	
11	SMA	
12	Haemoglobinopathies	CDC/Coriell (HbS)

Conclusions:

These activities have raised awareness of the importance of using (certified) Reference Materials in genetic testing, and will promote to the availability of additional RMs in this field.

Several RMs are becoming available for the 12 most tested inherited diseases. Future needs have been identified in the fields of cytogenetics, molecular oncology, infectious disease, pharmacogenetics and prenatal diagnostics.

For more details, visit our website at www.eurogentest.org/unit1/referencematerials

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