Welcome
Welcome to our autumn edition, which comes just as the EuroGentest team sets off to South America to give a series of presentations on the expertise gathered in the project to date. As you know, I have now handed over my role as official coordinator as planned to Gert Matthijs, but am still extremely active working on behalf of EuroGentest. In particular, this involves looking at possibilities for maintaining the project on course at the end of 2009.

Genetic Services Quality Committee

The ESHG recently approved a new Genetic Services Quality Committee whose aim is to improve the quality and provision of genetic services in Europe for the benefit of users. The first meeting was held in June at the ESHG conference in Barcelona. This initial meeting focussed on defining the scope and identifying perceived areas of need within the genetic community. The Quality Committee has supported the initiative by NISBC to have the Fragile X reference materials adopted by WHO. The next Quality Committee meeting will be held in December.

New web pages

Our web masters have redesigned several of our web pages and upgraded many of its functions to provide you with a better platform for genetic testing.

During the course of the EuroGentest project we discovered that there is so much content about the subject of genetic testing, that the EuroGentest website has had to be redesigned several times with this latest version as the result. The current content has been redistributed across 5 target groups: medical professionals, laboratories, students, patients & family and the industry. This is to make sure everyone

Quality issues

Next quality workshop
Validation of diagnostic tests: 8-9 January 2009, Prague, Czech Republic. Registration forms, contents and more detailed information is available on the EUGT website. More here

Are you using array CGH in your diagnostic work?
EuroGentest is looking at the different reference DNAs currently in use for array CGH, with a view to producing an ideal reference sample or pool of samples for diagnostic use. If you currently use a CGH (or other genome-wide copy number variation methodology), please complete the short survey at http://www.eurogentest.org/survey/aCGH/new.xhtml - we estimate it should take you less than 5 minutes to complete the 10 short questions!

Information services

Orphanet integration continues
The latest stats from Orphanet are fairly impressive. Its directory of 5,857 rare diseases is not only used by health professionals, but also increasingly by patients, families and researchers who all together consult over 20,000 pages every day. Through our partnership, EuroGentest has helped add new functions to the website, targeted particularly at biologists and researchers. By enhancing information on genes associated with rare diseases, Orphanet now offers the possibility to query by gene when searching for a clinical laboratory to perform a specific test and to find information on research activities linked to specific genes. By cross-referencing a gene with each rare disease to which it is associated, Orphanet provides a search option that allows users to easily capture all genes linked to a particular disease or all diseases linked to a particular gene. To access this service, click on the “rare diseases” tab on the front page, followed by the “gene” sub-tab that appears. Orphanet has also integrated the other leading scientific databases for genetics to facilitate smooth navigation between them. It is thus now possible to access additional information on a particular gene via OMIM, Genatlas, HGNC and Swissprot – all from Orphanet’s gene page.

New technologies

Alamut is a decision-support software application developed by Interactive Biosoftware for mutation diagnostics in medical molecular genetics is the subject of the latest technology evaluation by EuroGentest. Over 400 variants from 14 genes were tested and Alamut was found to provide accurate and high quality nomenclature, often better than manually generated nomenclature. The only problems found were in the protein-level nomenclature for a few complex variants, and with some cases of the interpretation of data from external data sources. Genomic and HGVS coordinates of genes were found to be in complete agreement with other data sources. More...

Diagnostic Molecular Genetics Best Practice Meeting - Open Call
EMQN in conjunction with EuroGentest are issuing an open call for topics for best practice meetings to be held in 2009. This call is subject to available funding. The aim of these meetings will be to draft new best practice guidelines or update existing ones for molecular genetics testing. Expressions of interests for a disease or technique specific best practice meetings
can find his or her content as fast as possible. Every target group has a homepage which groups together the most relevant and interesting documents, tools and web pages for that particular group.

All presentations, publications, reports and other documents have been put into a document repository. This is a database of all the documents which can be searched by using a new search engine tool, created on several of the new web pages.

EUGT General Assembly

General Assembly for all participants & collaborators on Thursday 20/11 and Friday 21/11.

More...

Upcoming news - more in next issue

- Gender and Genetics
- South America
- AGM
- Education workshop

EuroGentest

Harmonizing genetic testing across Europe

For more information visit www.eurogentest.org

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