EuroGentest workshops 2014

- **May 20th 2014**: E-course on the new version of ISO 15189: An interactive course on the similarities and differences in the new version of ISO 15189.
- **June 23-24 2014, Leuven (Belgium)**: ADVANCED WORKSHOP - Are we on the right track towards accreditation and what is new in the revised ISO 15189 standard? Specific quality requirements that are often forgotten or challenging to implement will be discussed in detail. Active input from the participants is expected.
- **July 8th 2014**: E-course on Internal Audit: An interactive course on the different steps in the audit process.
- **September 2014 (exact date and place to be confirmed)**: BASIC WORKSHOP - Validation of diagnostic tests in clinical molecular genetics: Meet experienced people in this field, learn about regulatory requirements (ISO 15189) or how to design a proper validation plan for new molecular genetic tests.

EuroGentest at ESHG in Milan (Italy), May 30-June 2014

Register now for the ESHG-EuroGentest-3GbTest satellite meeting, May 30th 2014, Milan (Italy): the meeting, entitled: "Clinical and quality issues when introducing new technologies in genomics" precedes the annual ESHG meeting and will focus on practical issues for lab people and for clinicians. The deadline for registration is Monday May 12th. Click here for more information.

We will also be there with a joint booth with ESHG! Look for us at booth n° 566 for useful information on the integration of EuroGentest with ESHG and for information on patient leaflets, CUGC's and other activities!

Proposed amendments to EU Regulation on Medical Devices are counter to patients' interests and unworkable, says ESHG

Recent amendments to the proposed Regulation on In Vitro Diagnostic Medical Devices (IVDs) currently before the European Parliament will restrict the rights of patients and doctors to carry out essential genetic testing, says the European Society of Human Genetics (ESHG) today (Monday 7 April 2014). Furthermore, an independent legal opinion now shows that the European Union (EU) has no competence to enact the Regulation as amended by the Parliament.

Please click here for the complete ESHG statement on the recent amendments to the proposed Regulation on In Vitro Diagnostic Medical Devices (IVDs).

European Commission adopts the two Decisions on the establishment of European Reference Networks (ERNs)

The Delegated Decision provides the criteria that European Reference Networks must fulfill and criteria that healthcare providers wishing to join such networks must fulfill. The Implementing Decision lays down the procedure on how to establish and evaluate the ERNs.

Both Decisions are expected to enter into force by the end of May, at the expiry of the two-month period for possible objection to the Delegated Decision by the EP and the Council.

The documents are available in the Register of Commission documents online.

Delegated Decision (access to PDF in English)

Implementing Decision (document can be requested)
Take part in the first ever Rare Chromosome Disorder Awareness Week 2nd - 8th June 2014

On behalf of Unique, CDO and the Bond family who suggested the initiative, we’d like to invite you to participate in the first ever ‘Rare Chromosome Disorder Awareness Week’ planned for June 2nd to 8th this year. During this week, we would like to see people with rare chromosome disorders, families, friends, support groups, professional colleagues, non-profit organisations and others to join together around the world to raise awareness of rare chromosome disorders.

We are hopeful that this is the beginning of a movement that will continue to grow year on year. We imagine that initial involvement through social media by you and your members, families and individuals will expand into awareness-raising events of your and their choosing as opportunities present themselves.

Click here for more information.

! New Clinical Utility Gene Cards

- Dent disease (Dent-1 and Dent-2) – Mar 2014
- Maturity-onset diabetes of the young – Feb 2013
- Oculocutaneous albinism – Feb 2013
- Phosphomannomutase 2 deficiency – Jan 2013
- Phosphomannose isomerase deficiency – Feb 2013
- Prader-Willi Syndrome – Apr 2013
- Transient Neonatal Diabetes Mellitus, 6q24-related – Feb 2013
- Williams-Beuren Syndrome [7q11.23] – Feb 2013

Click here for the full list of CUGC’s.

See also this publication: The future of Clinical Utility Gene Cards in the context of next-generation sequencing diagnostic panels.

Publications

By EuroGentest members:
- Developing noninvasive diagnosis for single-gene disorders: the role of digital PCR by Lyn Chitty and others.
- Offering prenatal diagnostic tests: European guidelines for clinical practice by Heather Skirton, Lyn Chitty and others.
- Higher Quality of Molecular Testing, an Unfulfilled Priority: Results from External Quality Assessment for KRAS Mutation Testing in Colorectal Cancer by Els Dequeker and others.
- Whole-genome sequencing in newborn screening programs by Pascal Borry and others.
- Raw data: research and health care goals differ by Martina Cornel and others.

For your interest:
- A massive parallel sequencing workflow for diagnostic genetic testing of mismatch repair genes
- Genetic Counselors’ Views and Experiences with the Clinical Integration of Genome Sequencing
- Noninvasive prenatal screening for fetal trisomies 21, 18, 13 and the common sex chromosome aneuploidies from maternal blood using massively parallel genomic sequencing of DNA

Other

- Connect to EuroGentest on LinkedIn
- Read more about EuroGentest