Executive Summary of the 7th Meeting of the EUCERD now online

The seventh meeting of the European Union Committee of Experts on Rare Diseases (EUCERD) took place on 31 January – 1 February 2013 in Luxembourg; the plenary session was preceded by breakout sessions by stakeholder group to discuss the draft EUCERD Recommendations on Common Principles and Consensus on patient registries and data collection for rare diseases.

The public executive summary is available on the website. Click here for more information.

Leaflet on Genetic Tests for Health Purposes translated in 17 languages

The Leaflet on Genetic Tests for Health Purposes was produced by the Council of Europe, with the support of Eurogentest and the European Society of Human Genetics. It aims at providing general objective information on genetic tests, including their nature and the potential implications of their results. It presents the different types of tests available, their applications in the medical field and the extent and limit of the significance of the information resulting from these tests.

The leaflet has been translated now in 17 languages, for which we would like to thank the many voluntary translators! More languages are in preparation.

Click here for more information and to download the leaflet.

Belgian Medical Genomics Initiative Kickoff Meeting: ‘Bringing genomics to the clinic: potential impact and challenges to overcome’

This meeting will be organized by the Belgian Medical Genomics Initiative (BeMGI), a new BELSPO-Interuniversity Attraction Poles project. It is specially aimed at reaching medical specialists, who may very soon be users of NGS for diagnostic and research purposes.

It will take place Tuesday 18 June 2013 from 17h30 – 20h30 in Centraal Auditorium, Campus Gasthuisberg Leuven, Belgium.

Click here for more information.

EuroGentest workshops

E-course: 31 May 2013 (more information will follow)
Workshop on Interpretation of diagnostic genetic results (biochemical, cytogenetics and molecular)

! New Clinical Utility Gene Cards

Catecholaminergic polymorphic ventricular tachycardia (CPVT) – Apr 2013

Click here for the full list of CUGC's.
Publications

By EuroGentest members:
- Psychosocial consequences of predictive genetic testing for Lynch syndrome and associations to surveillance behaviour in a 7-year follow-up study. By Helena Kääriäinen and others.
- The clinical implementation of non-invasive prenatal diagnosis for single gene disorders: Challenges and progress made. By Lyn Chitty and others.
- Acceptability of a minimally invasive perinatal/paediatric autopsy: healthcare professionals’ views and implications for practice. By Lyn Chitty and others.
- Advancing social research relationships in postnatal support settings. By Heather Skirton and others.

For your interest:
- We screen newborns, don't we?: realizing the promise of public health genomics.
- A Streamlined Protocol for Molecular Testing of the DMD Gene within a Diagnostic Laboratory: A Combination of Array Comparative Genomic Hybridization and Bidirectional Sequence Analysis.
- The policy of public health genomics in Italy.
- The Italian National External Quality Assessment Program in Molecular Genetic Testing: Results of the VII Round (2010-2011).
- Quality Control Methods for Optimal BCR-ABL1 Clinical Testing in Human Whole Blood Samples.
- Incorporating DNA Sequencing into Current Prenatal Screening Practice for Down's Syndrome.

Other

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