European Journal of Human Genetics (2013) 21, 1

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EDITORIAL

The EuroGentest Clinical Utility Gene Cards continued

European Journal of Human Genetics (2013) **21,** 1; doi:10.1038/ejhg.2012.161

We are happy to announce that the EuroGentest Clinical Utility Gene Cards (CUGCs),[1] first launched in this Journal by the end of 2010, will receive continuous support through EuGT2 (www.eurogentest.org) and the European Society of Human Genetics, at least until 2013. By this time, 300 of these guidelines for the application of genetic tests in clinical practice will have been created, and the majority of them will have received at least one update so as to closely reflect the state of the art. The first such updates are published in this issue of this Journal. CUGCs are built on the ACCE[2] (analytical validity, clinical validity, clinical utility, and ethical, legal and social implications) evaluation process for genetic testing, and have an emphasis on the clinical utility aspects. The *European Journal of Human Genetics* continues to publish printed abstracts and offers free access to the full-text versions in its on-line editions. In addition, both the EuroGentest (www.eurogentest.org) and the Orphanet (www.orpha.net) websites provide links to all available gene cards. The EuroGentest website, in addition, gives monthly updated information on the status quo of the collection of CUGCs, whether completed, under preparation or still waiting to be initiated. The CUGCs seem to be well received by the scientific and clinical community: the projected rate of downloads from this Journal's website per gene card and year ranges between approximately 600 and 1500, with an average above 1000.

The Clinical Utility Gene Cards represent either alternatives or complements to other collections serving similar purposes, in particular the NCBI GeneReviews (http://www.ncbi.nlm.nih.gov/books/NBK1116/) and the UK-GTN Gene Dossiers (http://www.ukgtn.nhs.uk/gtn/Information/Services/Gene+Dossiers). Out of the 60 currently published CUGCs (http://www.eurogentest.org/web/info/public/unit3/geneCards.xhtml) 12 titles are exclusively present in the CUGC initiative, while only 7 titles are covered by all three initiatives. The overlap of the CUGC guidelines and the NCBI GeneReviews amounts to 39 diseases, CUGCs and UK-GTN Gene Dossiers to two diseases.

In order to satisfy the highest standards of quality CUGCs will continue to pass through a carefully designed editorial process. Potential first authors are identified by the section editor, often based on external suggestions. Authors are selected on the basis of objective evidence of expertise, including their publication record. First authors are requested to invite a group of co-authors encouraging international representation. All CUGCs undergo peer review. The same procedure is applied to updates.

We hope that this initiative will continue to be a useful service to the scientific and clinical community.

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1 Schmidtke J, Cassiman J-J: The EuroGentest1 Clinical Utility Gene Cards. *Eur J Hum Genet* 2010; **18**: 1068.
2 Burke W, Atkins D, Gwinn M *et al*: Genetic test evaluation: information needs of clinicians, policy makers, and the public. *Am J Epidemiol* 2002; **156**: 311–318.