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CLINICAL UTILITY GENE CARDS

The EuroGentest Clinical Utility Gene Cards

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The ACCE[1] (analytical validity, clinical validity, clinical utility and ethical, legal and social implications) evaluation process for genetic testing is well established. Although the analytical validity of most tests does not seem to present major problems, little if any information is available in the literature on the other aspects of the majority of the molecular tests performed routinely in most labs.

This issue of the *European Journal of Human Genetics* contains abstracts of the first set of clinical utility gene cards available to all those concerned with defining reasonable indications for genetic testing for hereditary conditions in real settings of clinical genetic services. The full-text versions are available in the online editions of this journal.

These clinical utility gene cards focus in the first place mainly on Mendelian diseases. They contain two main sections: a section on the clinical validity (informative value) of available genetic tests and a section on their clinical usefulness in diagnostic, predictive, and prenatal settings.

Earlier, EuroGentest (http://www.eurogentest.org) had developed 'points to consider' regarding clinical indications for genetic testing.[2] The German Society of Human Genetics had endorsed these recommendations and started a process of developing disease-specific 'indication criteria' for genetic testing (http://www.gfhev.de). In this simple format and content, they are intended to provide quick guidance to referrers, service providers, and payers.

EuroGentest decided to relocate this activity to the European level. In a first step, the German guidelines, in an English translation, were temporarily placed on the EuroGentest (http://www.eurogentest.org/professionals/documents/info/public/unit3/geneCards.xhtml), ESHG (http://www.eshg.org), and Orphanet (http://www.orpha.net) websites, inviting for general and specific commentaries from the scientific community. In a second step, presented in this journal, the guidelines have now been reedited and referenced by a number of European experts.

Although the clinical utility gene cards claim to represent the state of the art at the time of publication, the rapid medico-scientific progress in this field will require regular updates, a service that EuroGentest hopes to be able to provide in the years to come.

We hope that this activity will be found useful and incite expansion. This editorial is thus also intended to serve as a call to the genetics community to collaborate on further submissions of clinical utility gene cards for diseases not yet covered (http://www.orpha.net).

CONFLICT OF INTEREST

The authors declare no conflict of interest.

1 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.
2 Javaher P, Kääriäinen H, Kristoffersson U *et al*: *Comm Genetics* 2008; **11**: 75–120, see page 118.

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