Clinical Utility Gene Cards (CUGCs) are disease-specific guidelines authored by multinational expert teams. They are dealing with the risks and benefits of the application of genetic tests in the clinical setting. Based on the ACCE framework (Analytical validity, Clinical validity, Clinical utility, Ethical, legal and social issues) the CUGB backbone consists of three parts: disease characteristics, test characteristics and clinical utility.

Clinical utility refers to the ability of a genetic test to significantly affect the clinical setting and patient outcome. In order to assess this information and make it available to clinicians, geneticists, referrers, service providers and payers, EuroGentest designed the Clinical Utility Gene Cards (CUGCs).

CUGCs represent concise and comprehensive documents for the clinically validated use of genetic testing. Subsequent to the completion the documents are peer-reviewed and published by the European Journal of Human Genetics (EJHG).

NGS panel data collection

With the long-term goal of adapting the CUGC format to next-generation sequencing (NGS) technologies, EuroGentest designed the NGS panel database. This data collection presents diagnostic NGS panels from different providers including panel name and tested genes. This information is further linked to the according disease(s) and genetic background, including OMIM genotype and Orpha number, see figure 2. In the current version the database does not include the association between gene symbol and OMIM phenotypes.

On the one hand this database gives users the opportunity to quickly identify diagnostic options according to the different search terms: disease, gene, OMIM number, panel name, provider and Orpha number. On the other hand it determines any overlap or gap of tested genes between different panels. Genes deemed essential by the providers can be easily identified. Based on these “core genes” the CUGC initiative can be expanded in order to cover NGS-based genetic test application in diagnostics.

Figure 1: Strategy of CUGC establishment.

Figure 2: Exemplary illustration of the NGS panel data collection, online available as an Excel 2007 file.

NGS panel providers were identified through a web search using the web search engine Google. In order to specify the test information the respective laboratory websites were reviewed. We initially focused on European NGS panel providers. As of March 6, 2014, we identified 28 laboratories having launched a total of 957 clinical NGS tests covering 2335 genes.

We encourage NGS providers from the commercial and academic sectors to contact us regarding their current services in order to include them in this database: eurogentest@mh-hannover.de.

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