Abstract

Since the sequencing of the human genome has been completed, the demand for genetic analysis in the human healthcare system is drastically increasing, and the extension of molecular genetic diagnostics is urgently needed. However, the majority of genetic diseases are molecularly and clinically highly heterogeneous, and until recently the available techniques lacked the required capacity to analyze several genes in parallel. The recently introduced high-throughput whole genome sequencing (WGS) technology now offers the unique opportunity to extend molecular genetic analysis by introducing these techniques and to develop tailor-made medical resequencing approaches for molecular genetic diagnosis of heterogeneous disorders. The “High throughput molecular diagnostics in individual patients for genetic diseases with heterogeneous clinical presentation” (TECHGENE) project aims to deliver crucial innovations leading to these approaches and to deliver a proof-of-principle for its implementation in selected model disorders.

Mission
TECHGENE aims to deliver crucial innovations leading to these approaches and to deliver a proof-of-principle for its implementation in selected model disorders. The model disorders have been selected with increasing genetic complexity and represent the majority of non-multifactorial genetic disorders. The current momentum to perform these innovations by a European consortium of clinical genetic diagnostic laboratories and research laboratories and industrial stakeholders will lead to a front-running position of European laboratories and small to medium-size enterprises (SMEs) in this field. The consortium consists of leading scientists and established laboratories providing cutting-edge knowledge with respect to quality management aspects, ethical and societal issues, and cost-effectiveness issues. This is the only approach that will warrant the development of diagnostic tools designed to restrict genetic testing to relevant medical factors. For European SMEs this project offers the opportunity to identify niches in the steadily increasing molecular genetic market. A specially designed training program will take care of rapid dissemination of the acquired knowledge and tools across Europe.

The European Union project TECHGENE focuses on the use of massive parallel sequencing techniques for the development, optimization, and implementation of diagnostic tools for genetic disorders.

It aims to extend the genetic diagnostics from the relatively simple monogenic disorders to the more complex genetically heterogeneous disorders.

To do so, a number of model disorders have been selected with increasing genetic complexity and which represent the majority of non-multifactorial genetic disorders. The following model disorders have been selected:

- Disorders caused by a wide variety of mutations in one or two genes (e.g., hemoglobinopathies and hereditary breast cancer)
- Disorders caused by mutations in one or two major genes and several minor genes (e.g., sensory disorders, including blindness, deafness, and Usher syndrome)
- Disorders caused by mutations in several equally important causative genes (e.g., paraplegias and ataxias)
- Disorders mainly caused by rare mutations in rare genes (e.g., mental retardation)

In the TECHGENE project, new massive parallel sequencing diagnostics will be developed for these model disorders, and a proof-of-principle will be delivered. The model disorders can be considered as prototypes for a wider group of diseases, and thus, in the near future, the tools developed will be
applicable for other diseases.

**Consortium History**

2009: Project launched

**Financing**

The project is funded through the European Union Seventh Framework Programme.

**Links/Social Media Feed**

Homepage [http://www.techgene.eu/](http://www.techgene.eu/)

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