

DEVELOPMENT OF A GENOMIC DIAGNOSTICS UNIT (UDIGEN)

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Reference of the project: RTC□2017□6471□1

Budget: 904,828.99 €

Duration: September 2018 – December 2021

Partners:

- Instituto Tecnológico y de Energías Renovables (ITER) – Project Coordinator
- Universidad de La Laguna (ULL)
- Fundación Canaria de Investigación Sanitaria (FUNCANIS)

Summary of the project:

The main purpose of the UDIGEN project is the development of an integrated genomic analysis platform, from DNA sequencing to data interpretation, to facilitate genetic counseling in order to reduce the time between a genetic disease is suspected and a definitive diagnosis. UDIGEN is constructed with the synergy between supercomputing and high-throughput DNA sequencing infrastructures of ITER, the recognized experience of ULL in Big Data analytics, and of FUNCANIS in the analysis and interpretation of human genetic variation.

The activities of UDIGEN will be based on the experience acquired on three sub-studies:

- a) A retrospective orthogonal validation study.
- b) A population study in the reference samples.
- c) A study of patients with suspected genetic disease.

As a result of the experience, it is expected that the routine use of exome sequencing will be introduced as a first-line test to sustain the diagnosis of genetic-based disorders, and the consequent reduction of time and costs until a conclusive diagnosis is reached. Additionally, the project will develop bioinformatics tools for the management, analysis and interpretation of genetic variation, and their connection with diseases using efficient Big Data analysis procedures.



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