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## How to create one of the main reference laboratories for the genetic testing of rare diseases

Manuel Pérez Alonso

The Institute of Genomic Medicine, Spain

Detecting gene mutations leading to rare diseases is a major need in healthcare. More than 3,000 rare diseases are known to be caused by mutations in the corresponding genes. This kind of knowledge can be used in Medical Genetics and Genomic Medicine, i.e., thousands of different gene tests may be developed in reference laboratories. Despite the obvious medical interest of this kind of tests, there are only a few global labs offering more than 1,000 different gene tests in their portfolio.

In this talk I will describe the creation and quick development of the The Institute of Genomic Medicine ([www.imegen.es](http://www.imegen.es)). After founding this reference laboratory, in 2009, the Institute has developed into one of the main Reference Laboratories for the Genetic Testing of Rare Genetic Diseases, providing more than 6,000 gene tests per year. Only five years after establishment, the portfolio of available molecular tests from the Institute includes more than 1,200 genes corresponding to more than 1,000 rare genetic diseases. This list grows every week on demand, i.e., we accept requests from clinics or hospitals in Europe, America or Asia for new tests to be developed from our labs. I will discuss the key points for our development that can be summarized in (1) the creation of interdisciplinary teams between biologists, bioinformaticians and IT experts; (2) active collaboration with the academia, small biotech companies and business people; (3) a strong commitment of the founders of the company on the idea that research has to benefit the Society; and (4) strong commitment within our team with business ethics.

### Biography

Manuel Pérez Alonso has a Degree in Biology and PhD in Molecular Genetics. He is Genetics Professor at the University of Valencia. He participated in five international genome sequencing consortia and (as PI) in a number of basic research projects. He is promoter and founding partner in four biomedical companies: ValentiaBioPharma ([www.valentiabiopharma.com](http://www.valentiabiopharma.com)), The Institute of Genomic Medicine ([www.imegen.es](http://www.imegen.es)), GenAGen ([www.genagen.es](http://www.genagen.es)) and Genera Biotech ([www.generabiotech.com](http://www.generabiotech.com)), all of them located at the University of Valencia Science Park. His research is now focused in the development of genomics tools for genetic testing. He also contributes to biopharmaceutical research through the study of the pathways leading to rare genetic disease. He is President of the Valencia BioRegion (BIOVAL) and President of the Spanish Association of Entrepreneurs in Science.

[alonsom@uv.es](mailto:alonsom@uv.es)