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From single gene sequencing to exome sequencing: Genetic testing of rare diseases

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Detecting gene mutations related to rare disease pathogenesis is a major need in patient healthcare. More than 3,000 rare diseases are known to be caused by mutations in the corresponding genes. This kind knowledge can be used for medical genetics and medical genomics, i.e., thousands of different gene tests may be developed internally in reference laboratories. Despite the obvious medical interest of this kind of genetic tests, there are only a few global labs offering more than 1,000 different gene tests in their portfolios. In this talk the professional pathway leading to the creation of the Institute of Genomic Medicine as well as the fast evolution of this reference laboratory will be described. After finding this genetics 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 six years after establishment, the portfolio of available molecular tests from the Institute includes more than 1,300 genes corresponding to more than 1,200 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 in our labs. Now the company includes a service for exome sequencing by Next Generation Sequencing. The key points for our development that can be summarized in (1) the creation of interdisciplinary teams between biologists, bio-informaticians and IT experts; (2) international collaboration; (3) active collaboration with the academia, small biotech companies worldwide and business people; (4) a strong commitment of the founders of the company on the idea that research has to benefit the Society; and (5) strong commitment within our team with business ethics will be described.

Biography

Manuel Perez Alonso has a degree in Biology and PhD in Molecular Genetics. He is currently working as a Professor in Genetics at the University of Valencia. He participated in five international genome sequencing consortia (as Principal Investigator) and in a number of basic research projects. He is Promoter and Founding partner in six biomedical companies: Valentia Bio-Pharma, The Institute of Genomic Medicine, Gen A Gen, Genera Biotech, Medi-Gene Press, all of them located at the University of Valencia Science Park. His research is now focused on the development of genomic 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 Bio-Region (BIOVAL) and President of the Spanish Association of Entrepreneurs in Science.

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