HeritaGen: Genetic and Genealogical Heritage Unification for Clinical Decision Making Support in Inherited Diseases

Databases with genealogical relationships linked to genetic information are very useful for epidemiological studies of genetic disorders. Integral analysis of this data helps clinicians in medical decision making, give genetic advice to families, identify individuals at risk of disease or discover new potential genetic causes for Mendelian diseases. A strategy was designed and implemented to collect the genealogical and genetic heritage of a delimited region to gain knowledge about primary immunodeficiencies (PIDDs) in the area. Next Generation Sequencing (NGS) technologies were used for whole exome sequencing (WES) of potentially immunodeficient participants. High Performance Computing (HPC) technologies were useful to manage the data. We have also developed the HeritaGen web platform to make it easier for clinicians to access and interpret information and help them in decision making.

Fuente de la publicación:

- José-Luis González-Sánchez, Bernabé Diéguez-Roda, José Antonio García Trujillo, Jonathan Gómez-Raja, Felipe Lemus-Prieto, Ana María Núñez-Cansado, María Peguero-Ramos, Álvaro Rodríguez-San Pedro and Silvia Romero-Chala. *HeritaGen: Genetic and Genealogical Heritage Unification for Clinical Decision Making Support in Inherited Diseases*. IWBBIO 2020, Congreso Internacional de Trabajo sobre Bioinformática e Ingeniería Biomédica. Octubre, 2020.