HPC Services to Characterize Genetic Mutations Through Cloud Computing Federations

One child in every 200 births may be affected by one of the approximately 6,000 monogenic diseases discovered so far. Establishing the pathogenicity of the mutations detected with NGS (Next-Generation Sequencing) techniques, within the sequence of the associated genes, will allow Precision Medicine concept to be developed. However, sometimes the clinical significance of the mutations detected in a genome may be uncertain (VUS, Variant of Uncertain Significance) which prevents the development of health measures devoted to personalize individuals' treatments. A VUS pathogenicity can be inferred thanks to evidences obtained from specific types of NGS studies. Therefore the union of supercomputing through HPC (High-Performance Computing) tools and the cloud computing paradigm (HPCC), within a Data Center federation environment offers a solution to develop and provide services to infer the pathogenicity of a set of VUS detected in a genome, while guaranteeing both the security of the information generated during the whole workflow and its availability.

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Ligações
[1] http://www.hrpub.org/journals/article_info.php?aid=6000