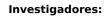


Estirpex-2



<u>CénitS</u> [1]

Idioma Inglés Descripción:



The Estirpex [2] project involved the deployment of free software in the infrastructure of LUSITANIA [3], to process the genetic sequence of certain exomes, store the information generated and filter and visualize the results obtained; but also, the opening of a series of lines of work aimed at optimizing the processes of obtaining high-level information, related to the massive sequencing studies carried out.

The experience that the <u>COMPUTAEX</u> [4] Foundation has demonstrated in the development of other projects in the field of healthcare, is reinforced by the rise that the Precision Medicine concept is experiencing, from which it is deduced that the moment in which development is framed of Estirpex-2 is unbeatable.

The Estirpex-2 project pursues, under the RIS3 Strategy (Strategy for Research and Innovation for the Specialization of Extremadura, Areas of Excellence in Health and Excellence of ICT), the continuity of the work developed in the Estirpex [2] project. Specifically, Estirpex-2 corresponds to the study, development and deployment of services for relevant economic sectors in the region that can benefit from the technology of Next-Generation Sequencing, supported by the use of supercomputing.

Estirpex-2 sought to expand the research and work carried out during the Estirpex project by deploying a catalog of ultrasequencing services that would allow the most important economic sectors in the region to access the advantages offered by mass sequencing.

The deployment of these services implied the assumption of responsibilities in terms of security, due to the sensitive nature of the information that could be treated. Therefore, it was necessary to carry out a security study that established the measures that should be implemented for an adequate legal compliance.

Objetivos:

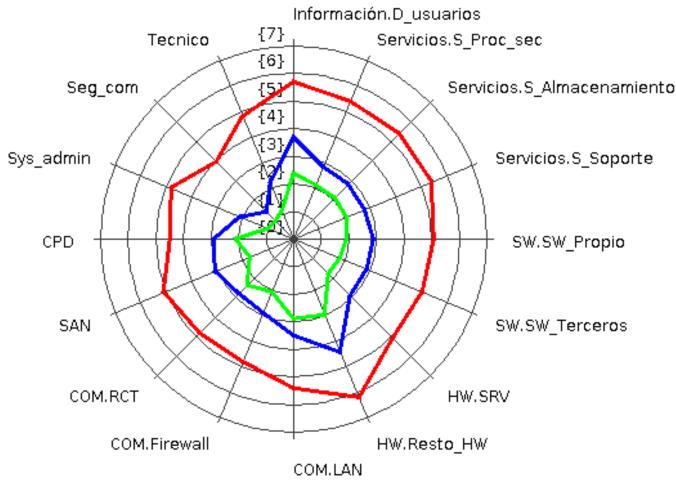
- Studying the technical implications associated with each project activity.
- Implementing software solutions to support the deployment of an NGS service of the catalog.
- Analysing the security of assets and information systems associated with the service.
- Bringing the services of the catalog closer to the economic sectors of the region that may be interested, emphasizing the added value that the genome sequencing of autochthonous, animal and plant species could have in the primary sector.

Metodología:

The first phase of the project consisted of a detailed analysis of the technical implications associated with each of the following project activities:

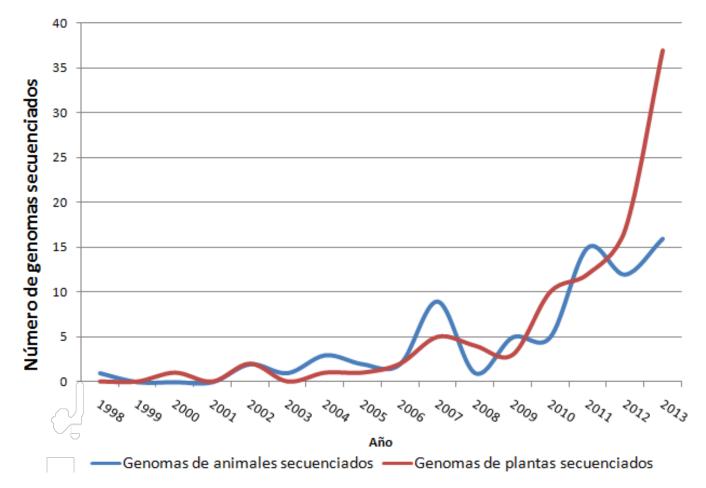
- Biochemical techniques and methods, as well as the necessary equipment, to carry out a process of Next-Generation Sequencing.
- Performance of the sequencers available in the market (cost, precision, time used, number of base pairs generated, etc.).
- More appropriate massive sequencing studies according to the platform used (resequencing, assembly of genomes, etc.).
- Software necessary to process the genetic sequences of the most typical studies that can be carried out with the sequencers that are currently on the market, namely: resequencing, assembly of genomes (by Novo sequencing) and RNA-Seq.
- Infrastructure that allows, not only the obtaining of high level genetic results in a time as low as possible, but the optimization of the use of computational resources (Cloud Computing).
- Strategic value of the services: given the importance that the primary sector has in the economy of the region, the collaboration between members of the <u>SECTI</u> [5] (Science, Technology and Innovation System of Extremadura) or other entities specialized in the plant and animal life, together with <u>COMPUTAEX</u> [4], could be very rewarding for the region.





During the development of Estirpex-2, software solutions have been implemented to support the deployment of an NGS service of the catalog:

- A prototype, in the OpenNebula platform, for the automatic processing of sequences generated by a resequencing service, including manual interpretation tools.
- Pedigree drawing software for specialists in genetic counseling.



Objetivos alcanzados:

IÉNITS

- Establishment of a catalog of services of processing and analysis of genetic sequences obtained by next-generation sequencing techniques at the CénitS center.
- Establishment of the necessary security measures to preserve the security of the information of the assets involved in the provision of next-generation sequencing services.
- State of the art of the use of ultra-sequencing techniques of animal and plant species and their possible applications in Extremadura, such as the characterization of certificate of origin or the cataloging of native species.
- Development of the **PedigreeX** tool, which facilitates pedigree drawing in an interactive way, facilitating this work for genetic counselors.

Fuentes de financiación:

Estirpex-2 is part of CENITAL 2, an innovation and research project developed under the Operational Program ERDF Extremadura 2007-2013, within priority 1: "Development of the Knowledge Economy".

Web:

http://www.cenits.es/enlaces/publicaciones/estirpex [6]

URL del envío:https://www.cenits.es/node/1605

Enlaces

[1] http://www.cenits.es/cenits [2] https://www.cenits.es/proyectos/estirpex [3] https://www.cenits.es/cenits/lusitania [4] https://www.cenits.es/fundacion [5] http://secti.gobex.es/ [6] http://www.cenits.es/enlaces/publicaciones/estirpex