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Bioinformatics architecture for integrating genomics data into Electronic Health Records

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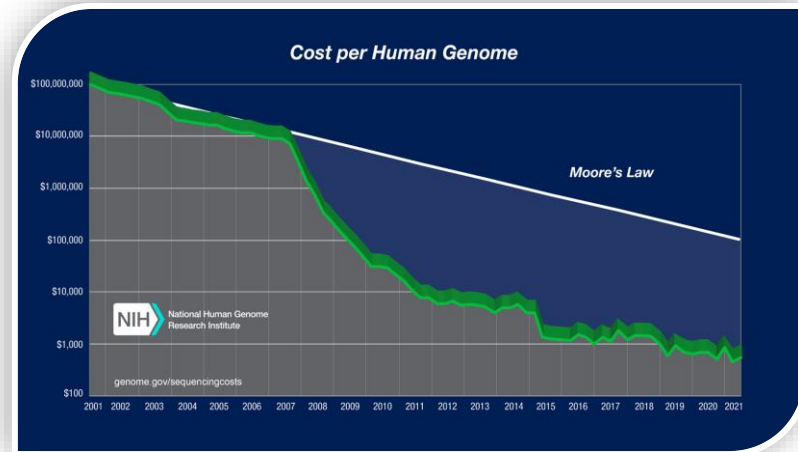
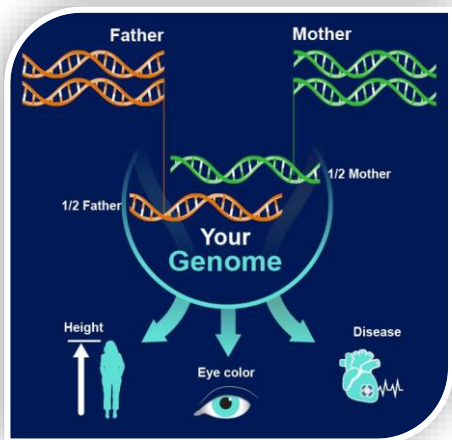


Agenda

1. Introduction
2. Methods
3. Results
4. Discussion & Conclusions



Introduction





Introduction

Context

- Most genetic tests that are run by NGS result in large volumes of genomic data, and interpretation can sometimes be challenging.
- EHR & CDSS are useful for supporting professionals in real-time decision making for complex situations, such as genomics

Objective

We present the design and implementation of a bioinformatic architecture aimed to organize the genomic information of the patients of the Hospital Italiano de Buenos Aires, and to make it available in the EHR through the CDSS.



Hospital Italiano de Buenos Aires



- Academic hospital
- EHR & HIS for more than 20 years
- Certified & accredited by several organizations





Methods

Work team



Reports and Genomic raw files



Bioinformatics software for annotation and filtering of raw files



Development and database technologies



Interoperability

Syntactic interoperability

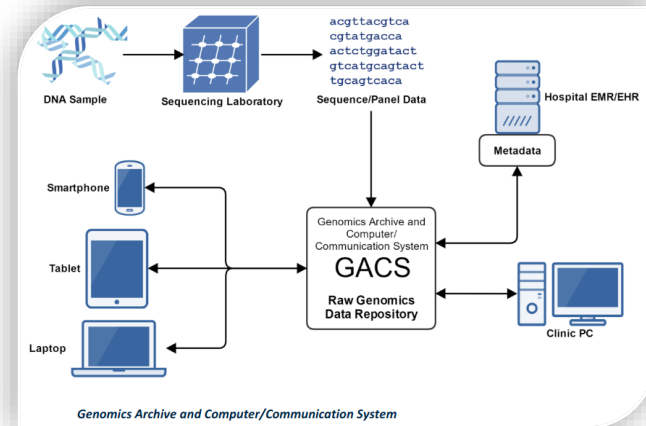
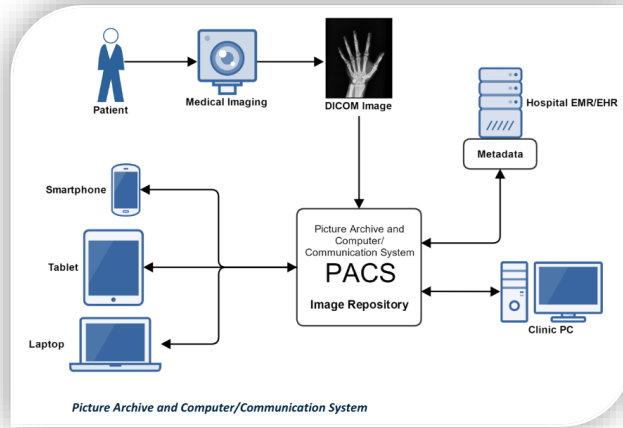


Semantic interoperability for Genetic variants and naming approved human genes



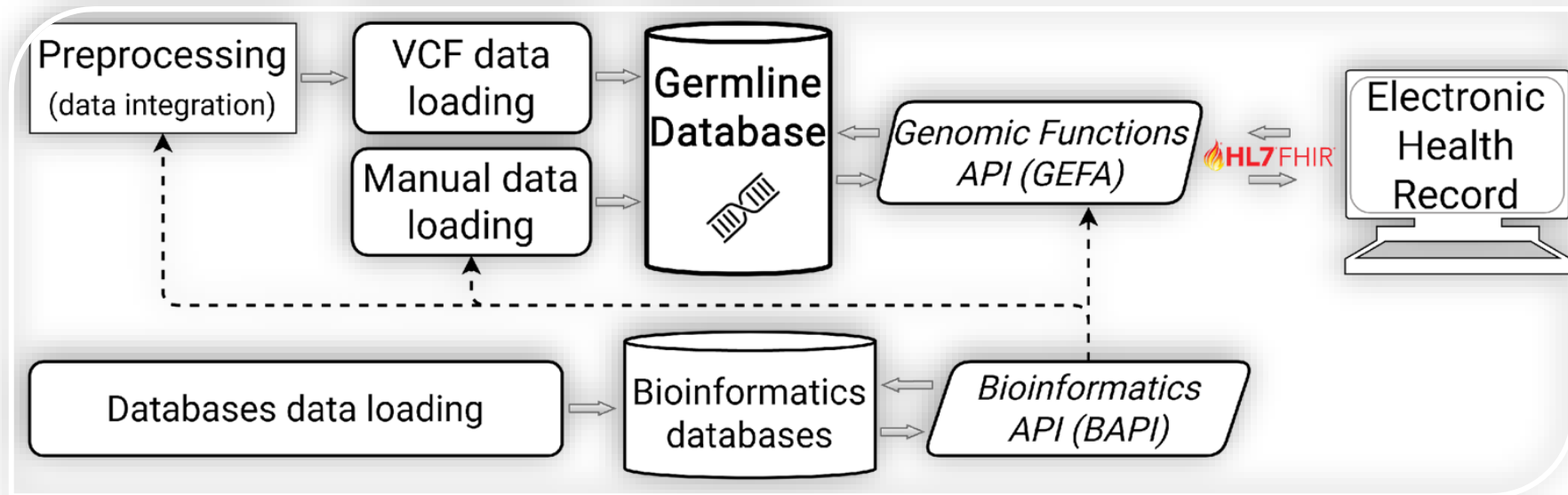


GHIS: Genomics in Health Information Systems



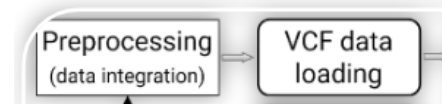


Results: Bioinformatics architecture



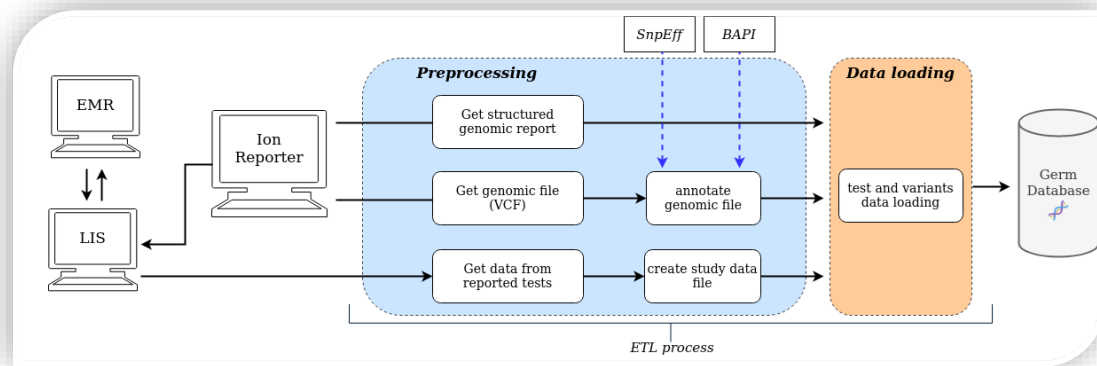


Automatic upload to the Genetic DB



"Incorporates the genetic data from the HIBA laboratory into the genetic DB"

- Integration with the LIS
- Integration with the systems of the DNA sequencing laboratory
- Processing and annotation of genomic files





Manual upload to the Genetic DB

Manual data loading

"Incorporates genetic data from external laboratories to the genetic database"

- Patient validation
- Inference and validation of genetic data:
 - Fast, complete and error-free data upload
 - more than 100 GB of information from public genomic databases

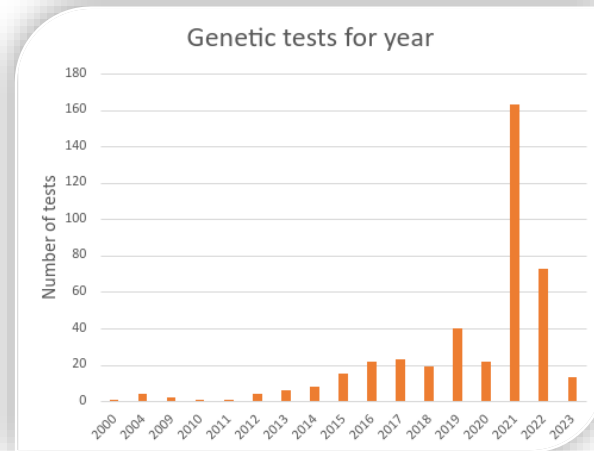
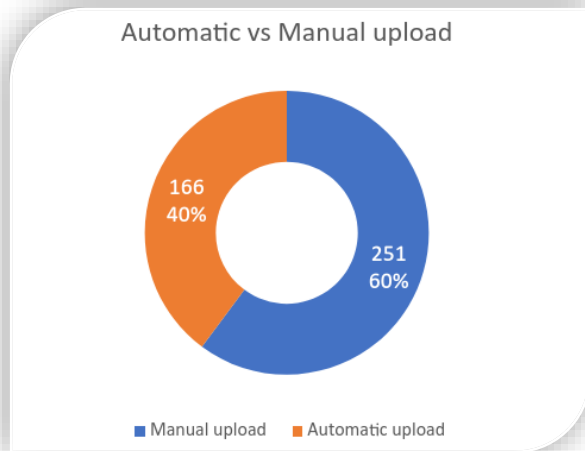
The screenshot shows the 'Nueva Variante' (New Variant) form in the Hivall system. The form is titled 'Nueva Variante' and includes a header with the Hivall logo and a user profile icon. Below the header, there is a table with columns for 'Fecha', 'Estado', 'Laboratorio', 'Origen', and 'Paciente'. The 'Fecha' field is filled with '01/01/2022'. Below the table, there are several sections for entering variant information:

- Variantes:** A section with a 'Tipo de Variante' dropdown menu, currently set to 'Sustitucion'.
- Variante Sustitucion:** A section with a 'Version del Genoma' dropdown menu, currently set to 'GRCh37 (hg19)'. Below this, there is a 'Gen' field with a dropdown menu set to 'TP53'.
- Transcrito:** A field with a dropdown menu set to 'NM_001126117' and a 'Version' dropdown menu set to 'NM_001126117.2'.
- Variante c:** A field with the value 'c.733G>T'.
- Variante p:** A field with the value 'p.Cys245Cys'.
- Significancia clinica en Clinvar:** A dropdown menu set to 'Pathogenic'.
- Significancia Clinica:** A section with radio buttons for 'Benigna' and 'Probablemente benigna'.



Genetic tests in the database

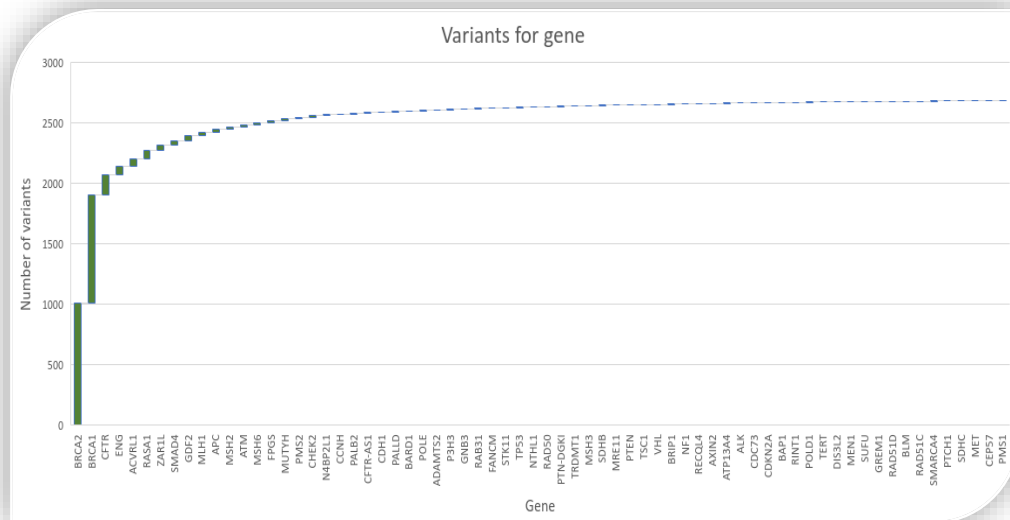
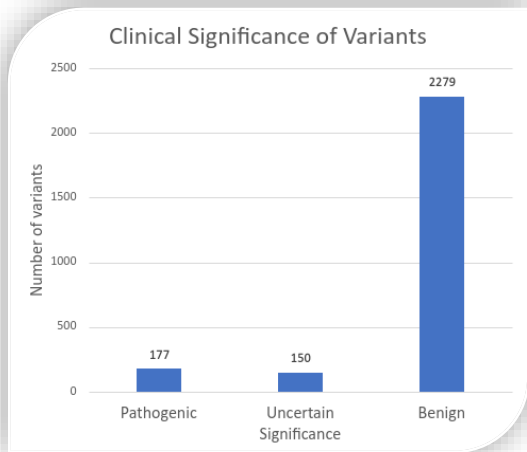
Germline genetic tests: 417





Genetic variants and genes in the database

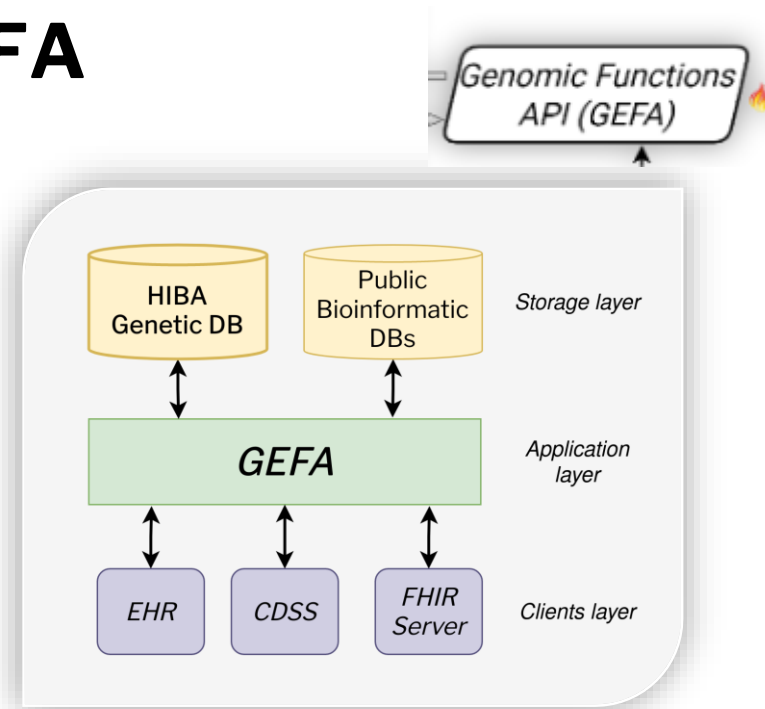
Germline variants: 2606





Genomic functions API - GEFA

- API to consult the genetic information of the patient
- Simplifies the search through high-level functions
- DB encapsulation
- Simplifies technical aspects of access
- Enrich responses with access to bioinformatics databases
- Returns the results using FHIR or Json format





Genetic rules in the CDSS

- Based on international guidelines
- Follow-up rule of the patient who has pathogenic variants in the BRCA1 or BRCA2 genes
- Rules for referral to genetic counseling for patients at higher risk of having a pathogenic variant due to diagnosis of:
 - breast cancer in women under 46
 - breast cancer in men
 - ovarian cancer
 - pancreatic cancer



Recomendaciones ✕

Prácticas preventivas / de seguimiento

Usted está creando el problema

- [CANCER DE MAMA](#)

Su paciente presenta un riesgo aumentado de poseer una variante patogénica en los genes BRCA1/BRCA2.

Le sugerimos solicitar:
Entrevista de Asesoramiento genético

[Solicitar](#) [Descartar](#)

Para más información sobre BRCA1/BRCA2 [presione aquí](#)



Conclusion

The implementation of an architecture such as the one presented in this work makes it possible to incorporate germline genomics into EHR, allowing for health professionals who are not specialists in genetics to incorporate genomic data into clinical decisions and therefore improve patient care.



TEAM

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Thank you!
Any questions?

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