The experience of Andalusia in eHealth and big data

Big data in health - IMI's HARMONY project, European Parliament, Brussels, 19 June 2018

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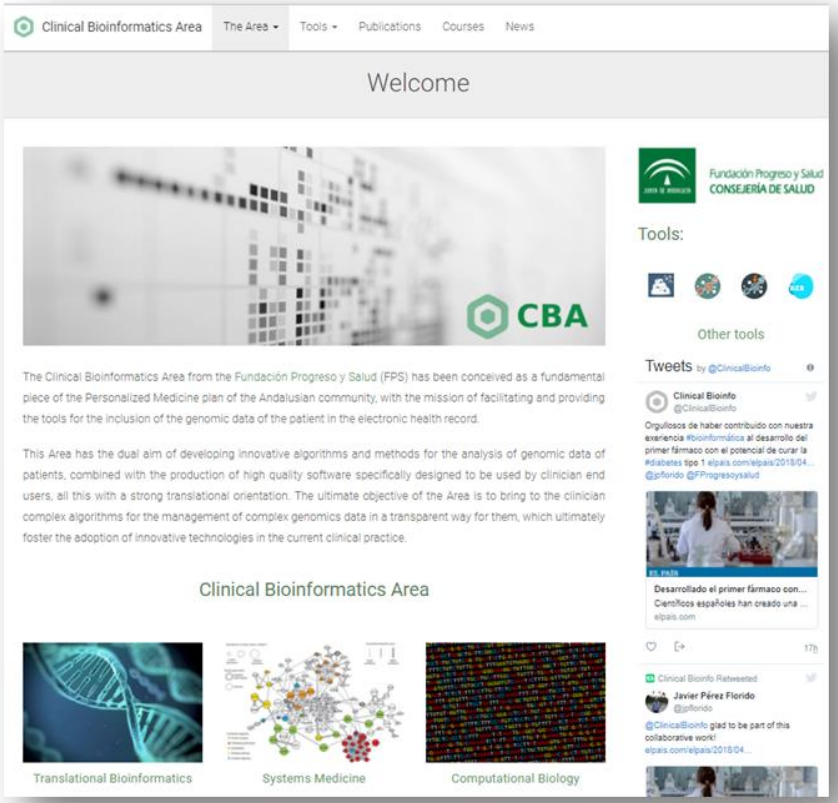
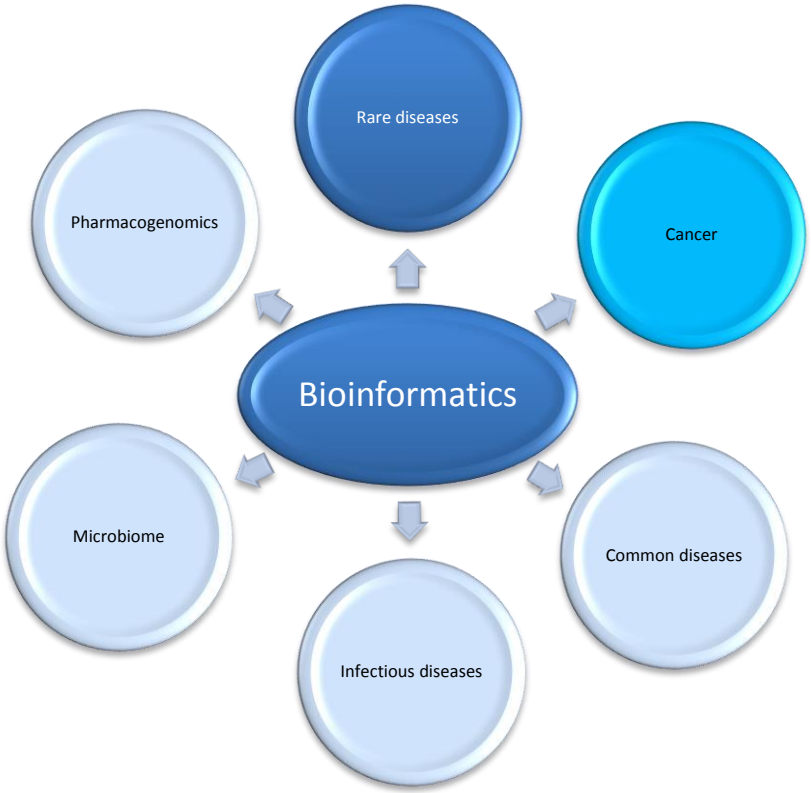
Summary

- **Genomic big data for personalized medicine**
- **Bioinformatics**
- Sustainability
- Scalability
- Generation of knowledge: prospective healthcare
- Data integration
- GDPR compliance

The clinical bioinformatics area





http://www.clinbioinfosspa.es/

The Bioinformatics Area, created in June 2016 in the Fundación Progreso y Salud, has as main goal supporting the Program of Personalized Medicine of the Andalusian Community by facilitating the use of genomic data for precision diagnostic and treatment recommendation, implementing a prospective health care functionality in the public health system.

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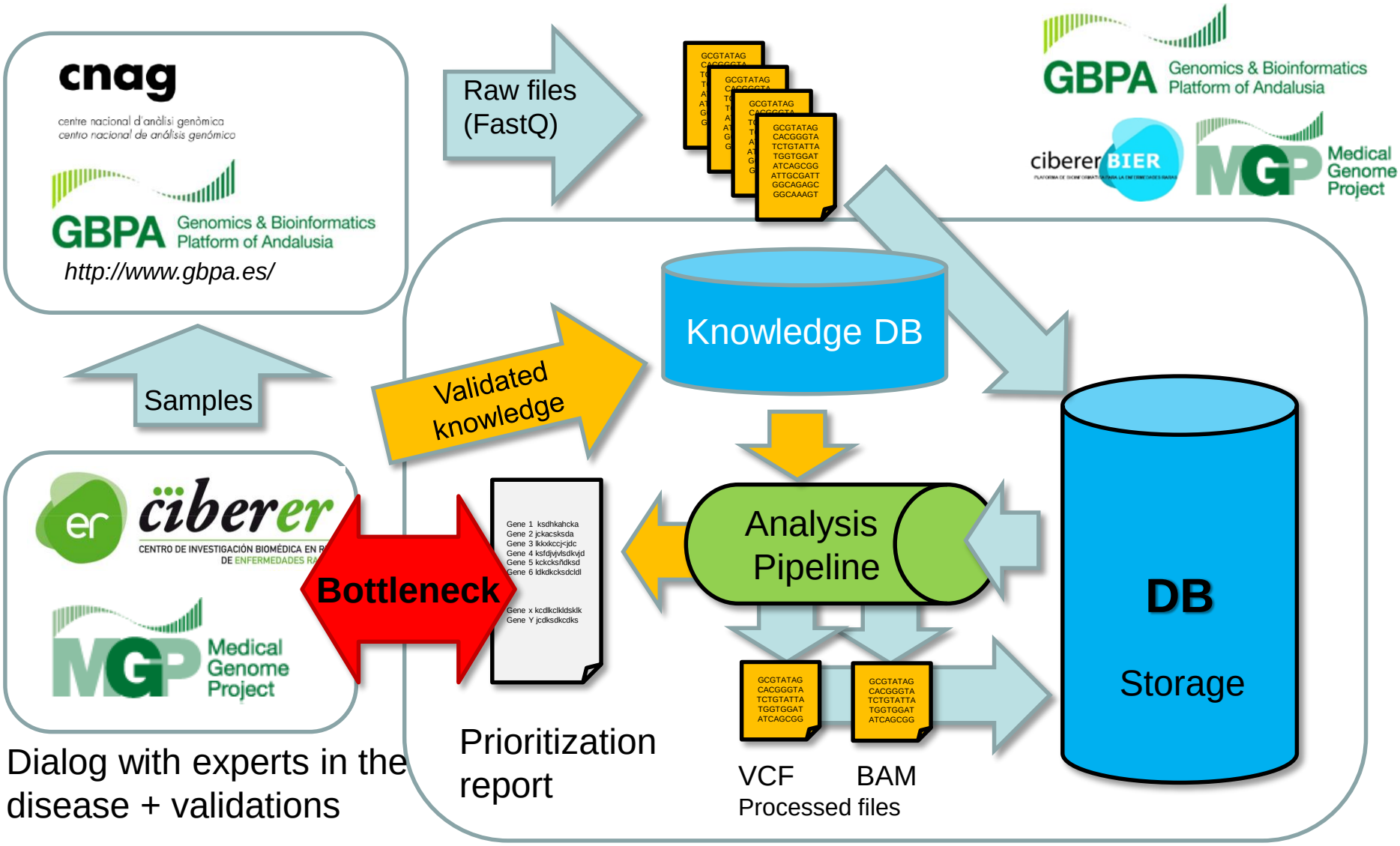
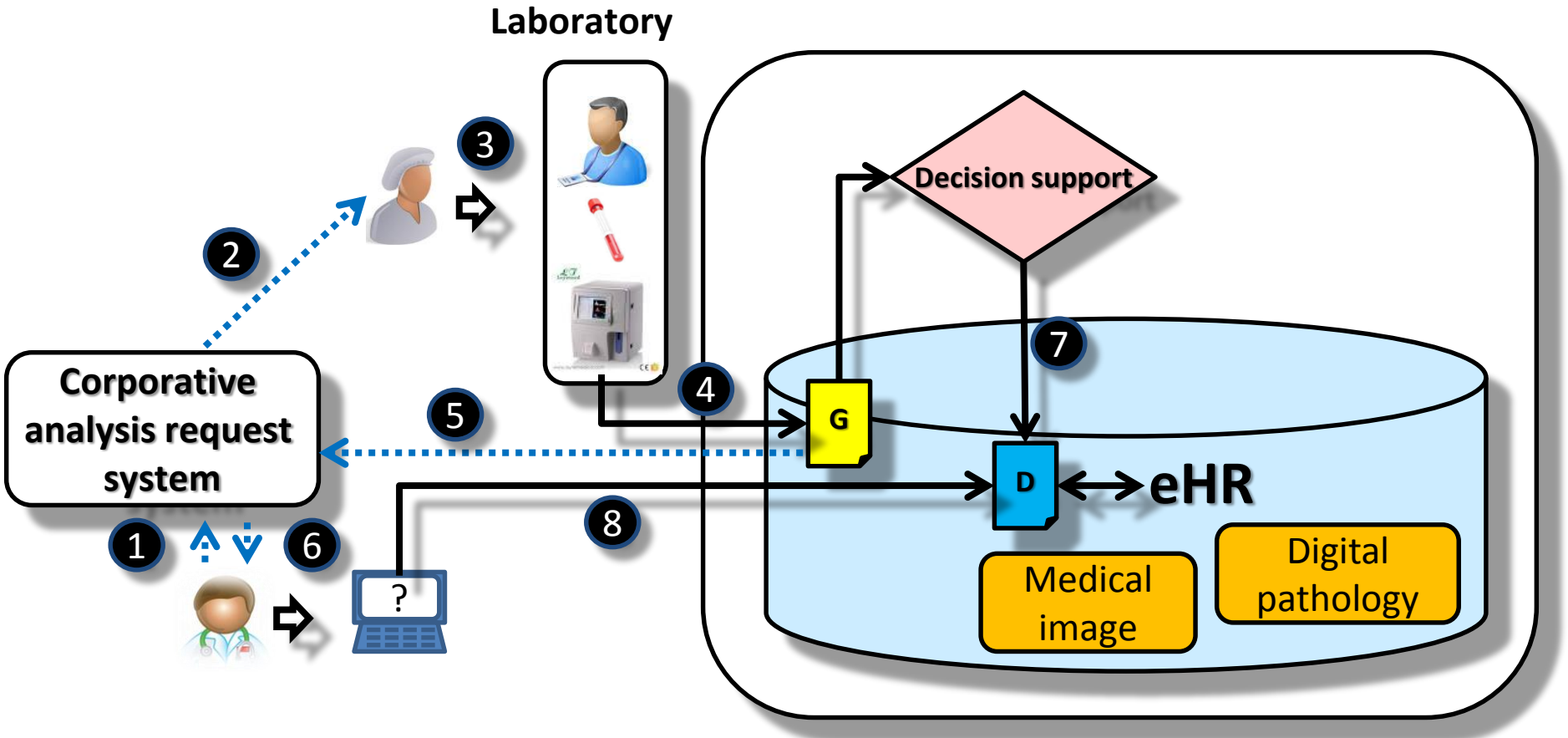
Data analysis and the sustainability of the cycle of knowledge generation



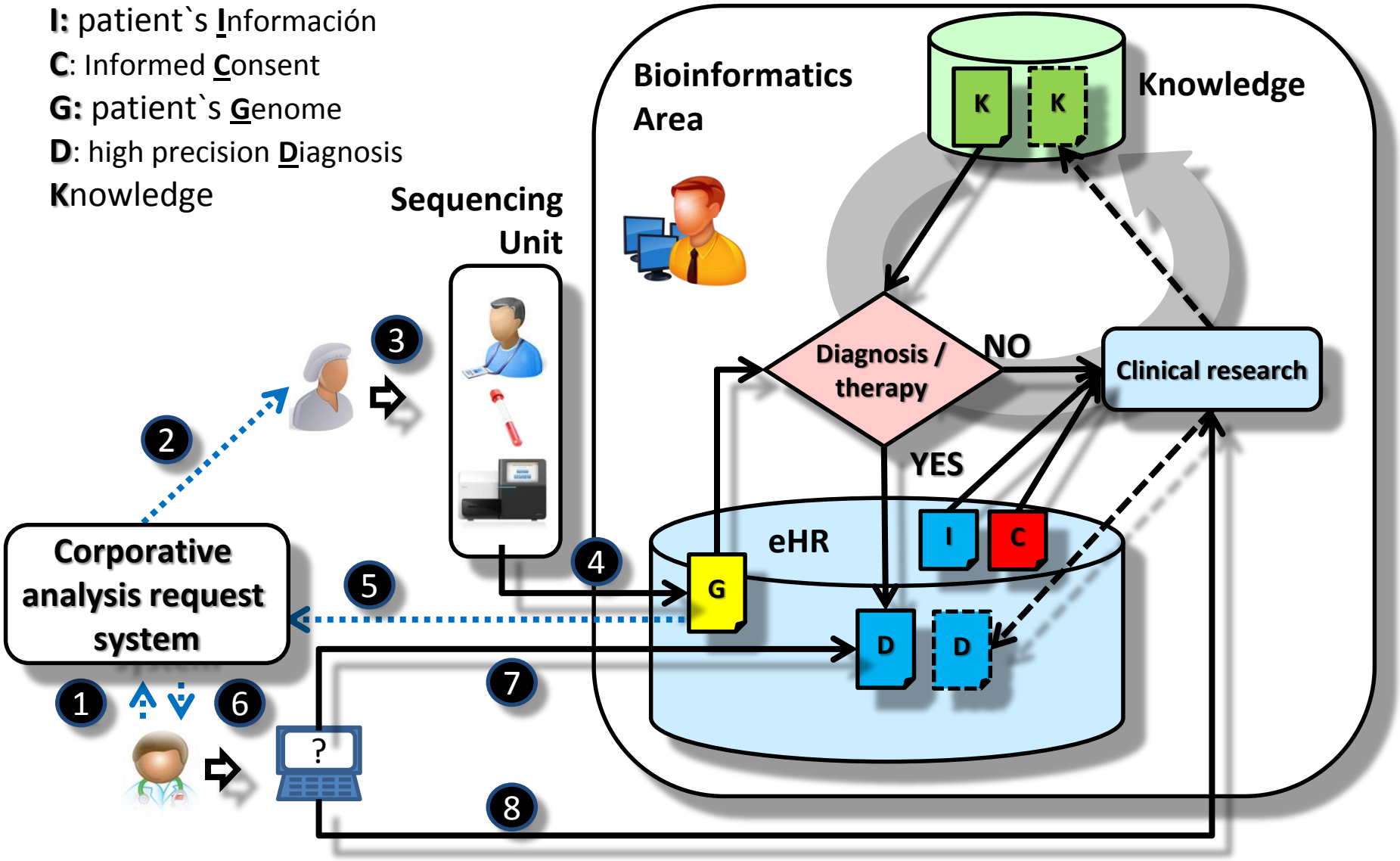
Sustainability requires tools for end users, which involves hiding the complexity of the analysis

- A solution for the management of genomic data must be integrated the same way that other analyses of the health system.
- Genomic data are stored in the system, linked to clinical data the same way that other data are (medical image, digital pathology –under implementation-, etc.) for further potential clinical studies

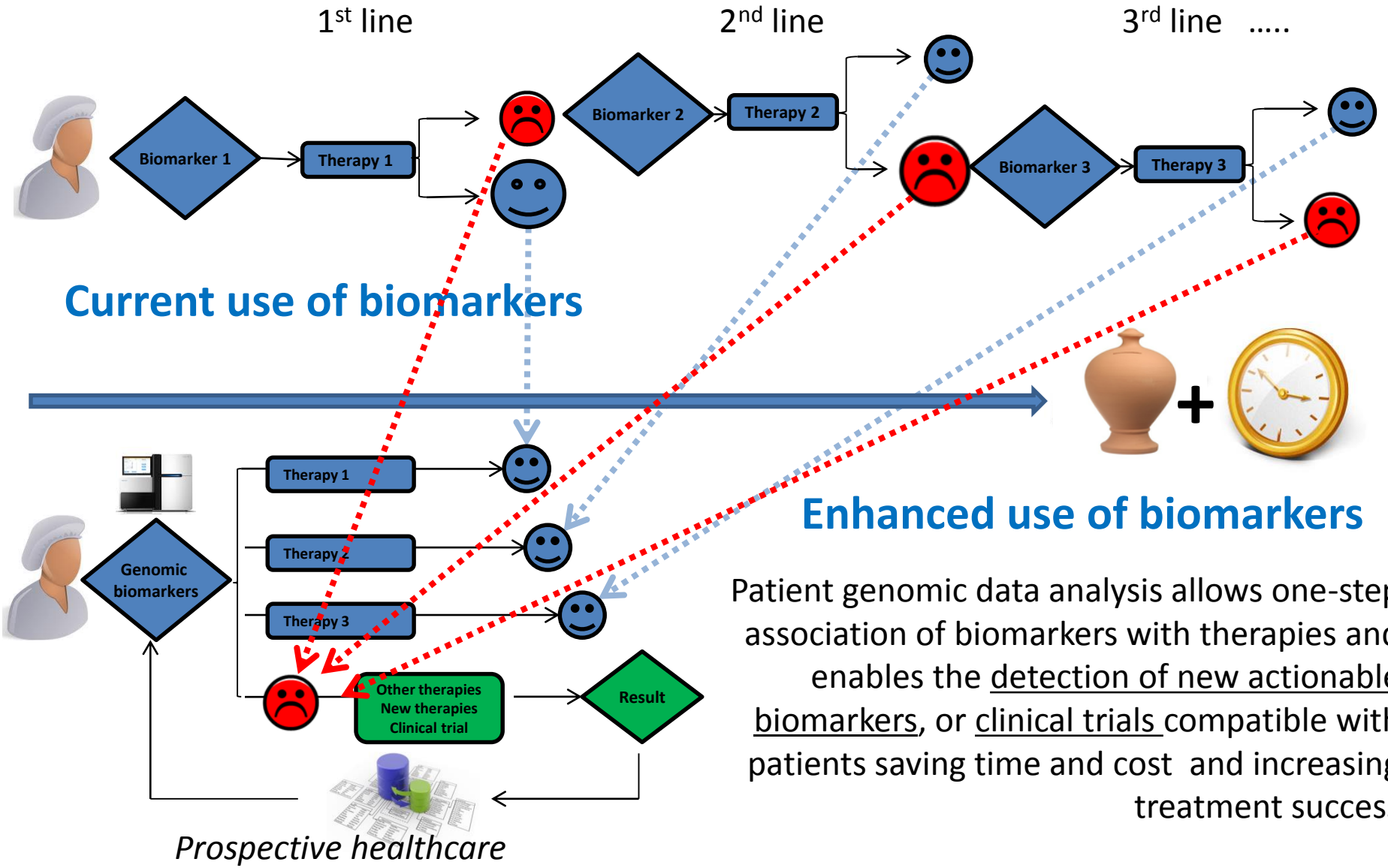


Our approach: hiding the complexity

I: patient`s **I**nformación
C: Informed **C**onsent
G: patient`s **G**enome
D: high precision **D**iagnosis
Knowledge



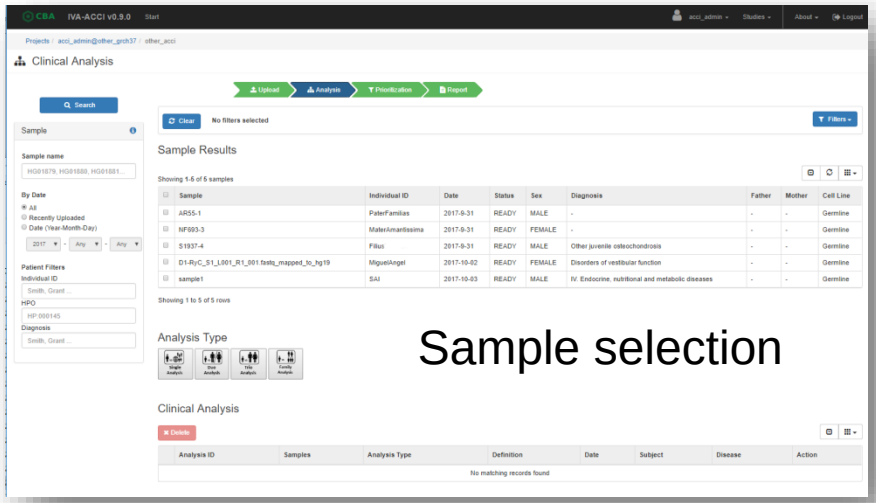
Personalized Medicine in cancer



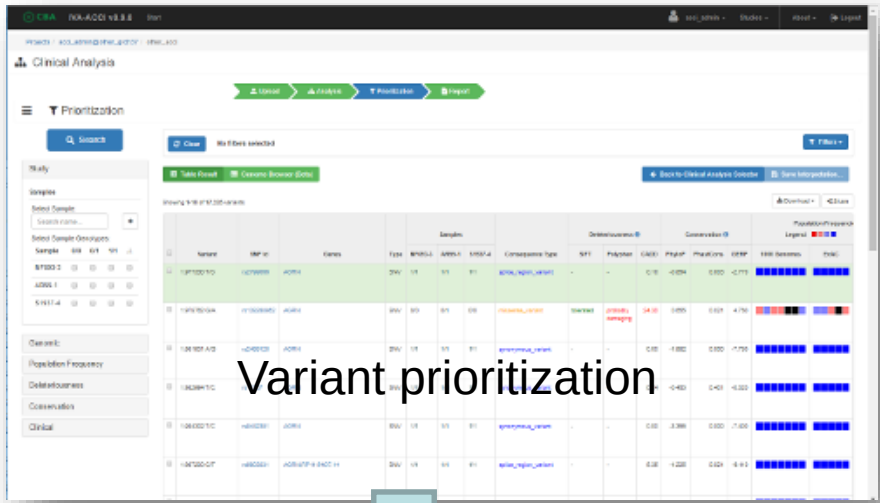
Enhanced use of biomarkers

Patient genomic data analysis allows one-step association of biomarkers with therapies and enables the detection of new actionable biomarkers, or clinical trials compatible with patients saving time and cost and increasing treatment success

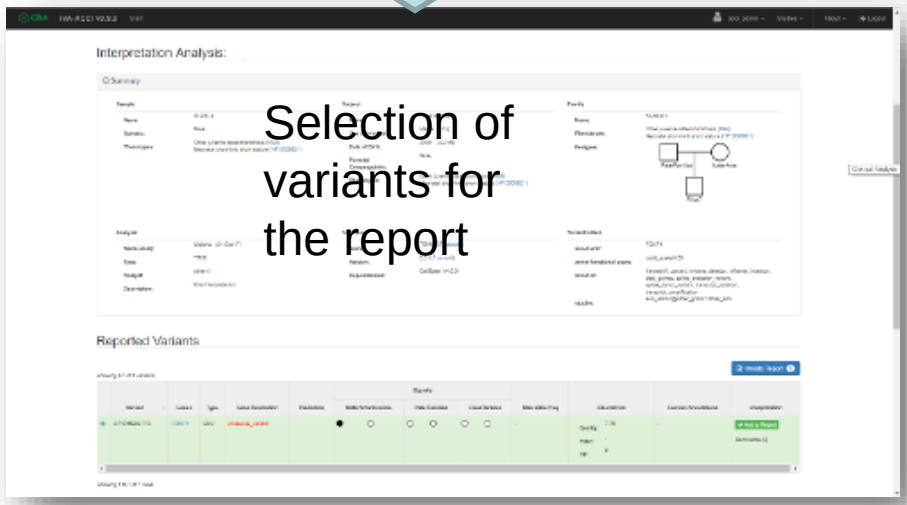
Front end: Personalized Medicine Module (MMP)



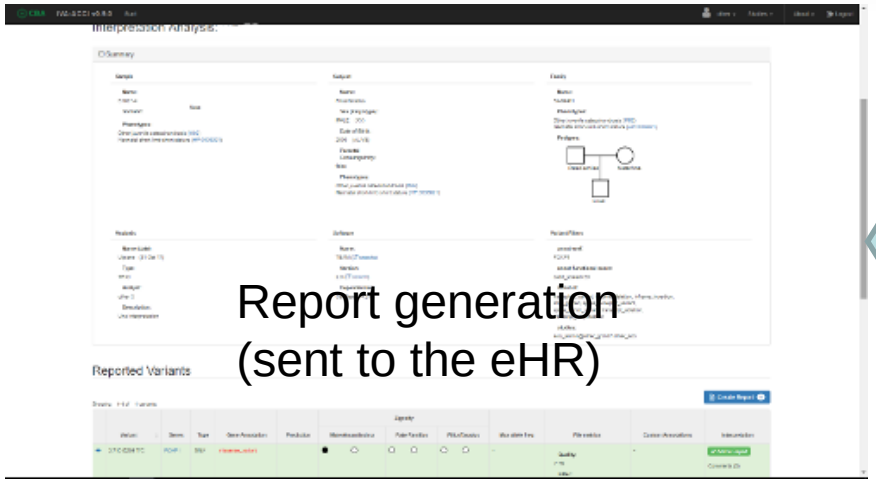
Sample selection



Variant prioritization



Selection of variants for the report



Report generation (sent to the eHR)

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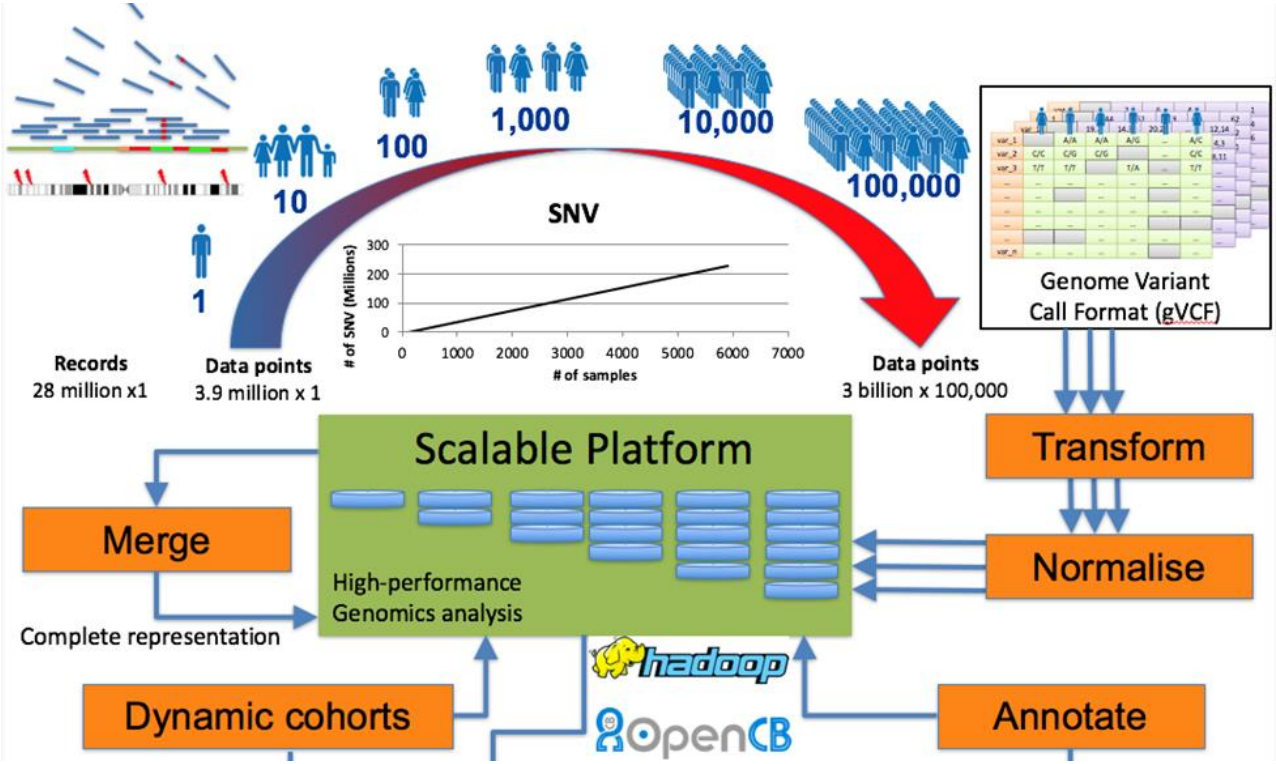
Backend: OpenCGA, a scalable storage and genomic data management platform

In collaboration with Genomics England (GEL)



Currently, the fastest and more powerful genomic database engine in the world.
Used in the GEL for genomic data management

Extensive capabilities to query across genotype and phenotype relationships

https://github.com/opencb/opencga

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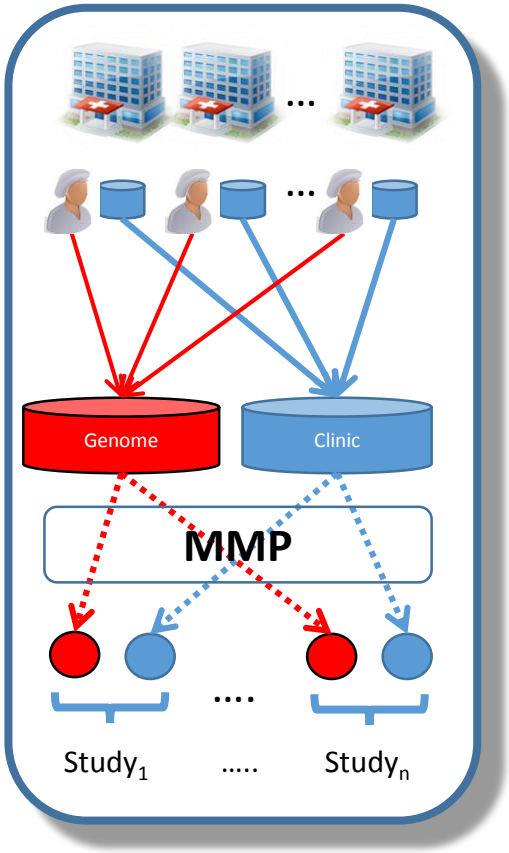
Prospective healthcare is facilitated by a model that integrates genomic data and universal EHR



- Revolutionary concept: the whole health system becomes an enormous potential prospective clinical study
- Clinical data dynamically associated to genomic data
- Possibility of many clinical studies by reanalyzing genomic data under diverse perspectives (with no extra investment)
- Growing genomic DB with increasing study possibilities

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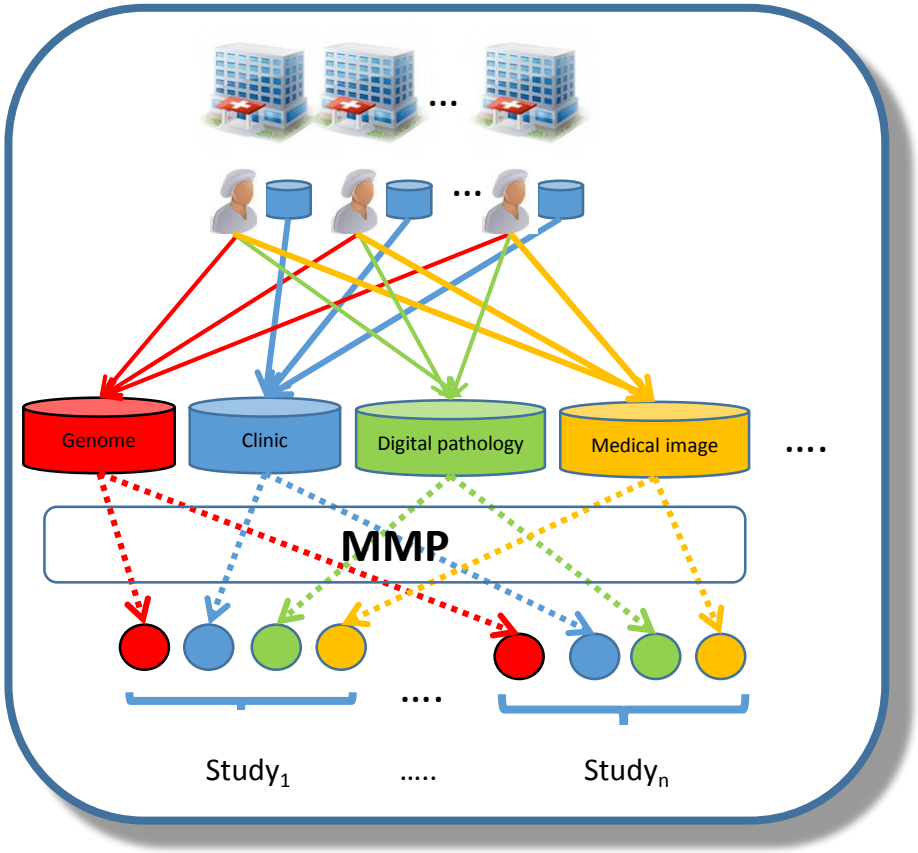
Future vision involves *big data* integration:

Genomic data are especially relevant but not the only useful *big data*



- Other *big data* are being collected (medical image, digital pathology, wearable devices, etc.)
- Clinical data dynamically associated to different *big data*
- The whole health system becomes a enormous potential prospective clinical study
- Immense possibility for data reusability
- Growing genomic DB with increasing study possibilities

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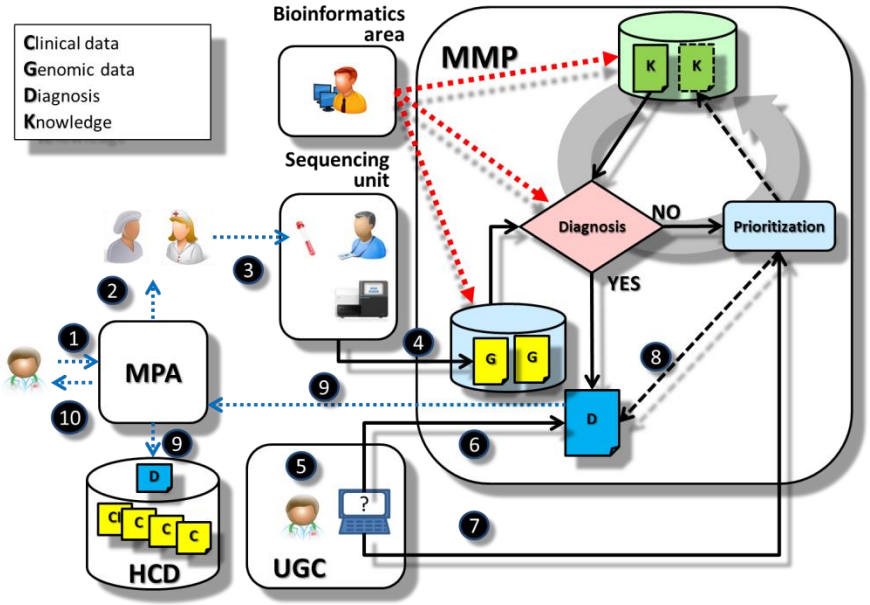
GDPR compliance

The system has been designed in a way that is compliant with EU and Spanish General Data Protection Regulation

- Clinicians requesting for a genomic diagnostic have access to eHR and get the result of the test.
- Geneticists have access to eHR and can query the genomic data (but never extract them)
- IT have access to de-identified genomic data and no to eHR.



Genomic and clinical data within the health system enable Personalized Medicine

- **Database of patients with prospective clinical information.** Patients sequenced:
  - Will have different responses to treatments in the future
  - Can have other diseases in the future
  - Dynamic diagnostic of undiagnosed patients as knowledge databases update
  - Dynamic assignment of treatments for patients without therapeutic options as knowledge databases update
- **Preventive medicine**:
  - Dynamic discovery of pharmacogenomics relevant variants in sequenced individuals
  - Dynamic discovery of new risk variants in sequenced individuals
  - Dynamic discovery of reproductive risk variants
- **Health system as a prospective genomic study for clinical knowledge generation**:
  - Prospective discovery of new biomarkers of response to drugs, therapies, prognostic, etc.
  - The pool of disease or risk variants is limited and could be surveyed soon

Clinical Bioinformatics Area
Fundación Progreso y Salud, Sevilla, Spain, and…

…the INB-ELIXIR-ES, National Institute of Bioinformatics and the BiER (CIBERER Network of Centers for Research in Rare Diseases)

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