GALAXYDX – A WEB-SERVER DEDICATED TO DIAGNOSIS DATA ANALYSES

Vivien DESHAIES[1,2,3] , Alban LERMINE[1,2,3] , Séverine LAIR[1,2,3] , Nicolas SERVANT[1,2,3], Elodie GIRARD[1,2,3], Julien TARABEUX[4,5] , Philippe HUPE[1,2,3], Claude HOUDAYER[4,5] , Emmanuel BARILLOT[1,2,3]

[1]Institut Curie, [2] INSERM U900, Bioinformatics and Computational Systems Biology of Cancer, 75248 Paris, France, [3]Mines ParisTech, 77300 Fontainebleau, France, [4]INSERM U830, Génétique et biologie des cancers, 75248 Paris, [5]Biologie des Tumeurs, 75248 Paris

Vivien.Deshaies@curie.fr, Alban.Lermine@curie.fr, Severine.Lair@curie.fr, Nicolas.Servant@curie.fr, Elodie.Girard@curie.fr, Julien.Tarabeux@curie.fr, Philippe.Hupe@curie.fr, Claude.Houdayer@curie.net, Emmanuel.Barillot@curie.fr
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We built **GalaxyDX**, an implementation of Galaxy containing a suite of softwares used for **the analyses of diagnosis sequencing data** (PGM torrent suite, BWA, GATK, VarScan, Annovar, … etc). Galaxydx allows Clinicians as well as Biologists to be autonomous to perform a complete set of analyses such as: (1) mapping, (2) variant calling, (3) variant filtering, (4) variant annotation, (5) rearrangements calling and (6) visualization through diagnosis dedicated Genome browser (Alamut).

We also work on **data integrity and confidentiality** by modifying the Galaxy writing methodology. Analyses in Galaxydx are organized by project and user, output files are owned by the user who generates them. It allows us to systematically check system rights on data before any process.

Place of GalaxyDX in Diagnosis Process at institutCurie

Automated QC/Analysis Pipeline

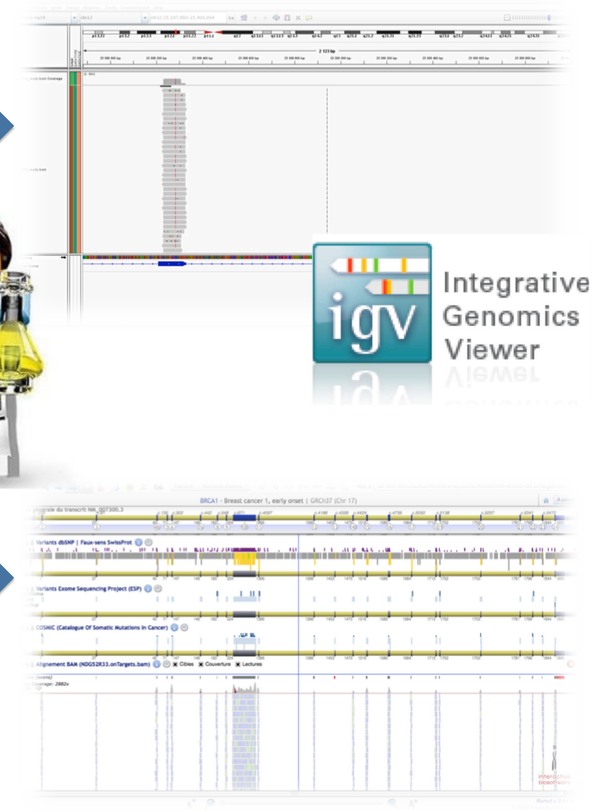
System Rights checking

GalaxyDX Tools Design New Pipelines

- ALIGNMENT
- COVERAGE CALCULATION
- VARIANT CALLING
- VARIANT ANNOTATION
- RGT CALLING
- VARIANT FILTRATION & REPORT

GalaxyDX Libraries

- RAW DATA (BAM, FASTQ, SFF)
- ALIGNED DATA (GENOME, ON TARGETS)



TOOLS

MAPPING

Tmap 3.6.2
Bowtie2
BWA
TopHat2

VARIANT CALLING

TorrentVariantCaller 3.6.2
GATK Unified Genotyper
GATK2 Hapotype Caller
VarScan
Freebayes

ANNOTATION

Annovar
SnpEff

OTHER ANALYSIS TOOLS

Rearrangement calling
Coverage for targeted genes
Variant decision
Variant table report maker

FORTHCOMING

Tmap 4.0.2
TorrentVariantCaller 4.0.2
GATK3

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institutCurie Inserm Galaxy MINES ParisTech