

GDA CIBERER 2016

Genomic Data Analysis

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Outline

1. Computational Genomics

2. GDA CIBERER course



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Príncipe Felipe Research Center

Goal: biomedical research

- **Basic research** in genes, targets, molecular and cellular processes, Nanomedicine and Computational Medicine
- **Translation into clinical practice:** personalized medicine, cancer, rare diseases, metabolic and functional impairment

<http://www.cipf.es/>



Who are we?

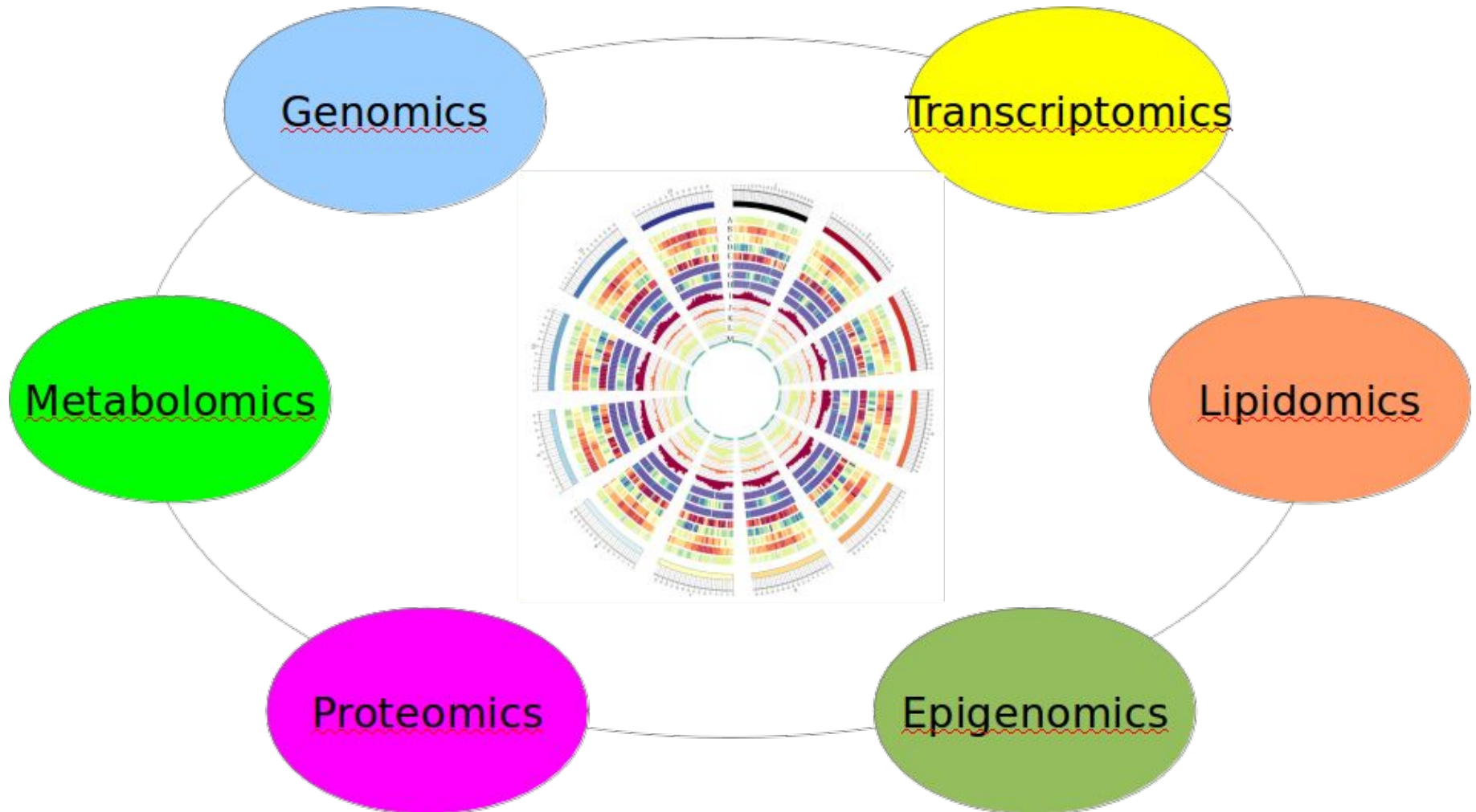
- The **Computational Genomics** Department, in Research Center Prince Felipe
- **Team:** multidisciplinary group of 14 researchers and technicians led by Joaquín Dopazo

<http://bioinfo.cipf.es/>

Who are we?

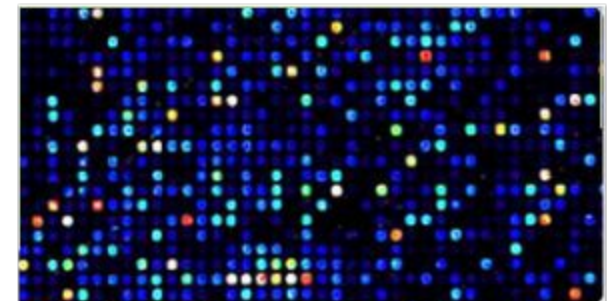
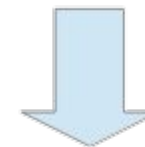
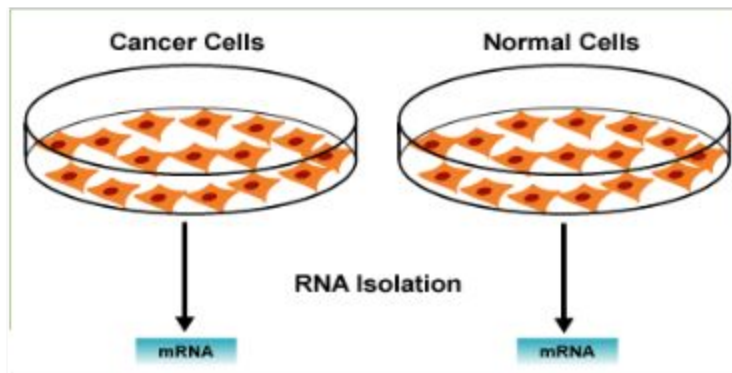


Computational Genomics



Computational Genomics

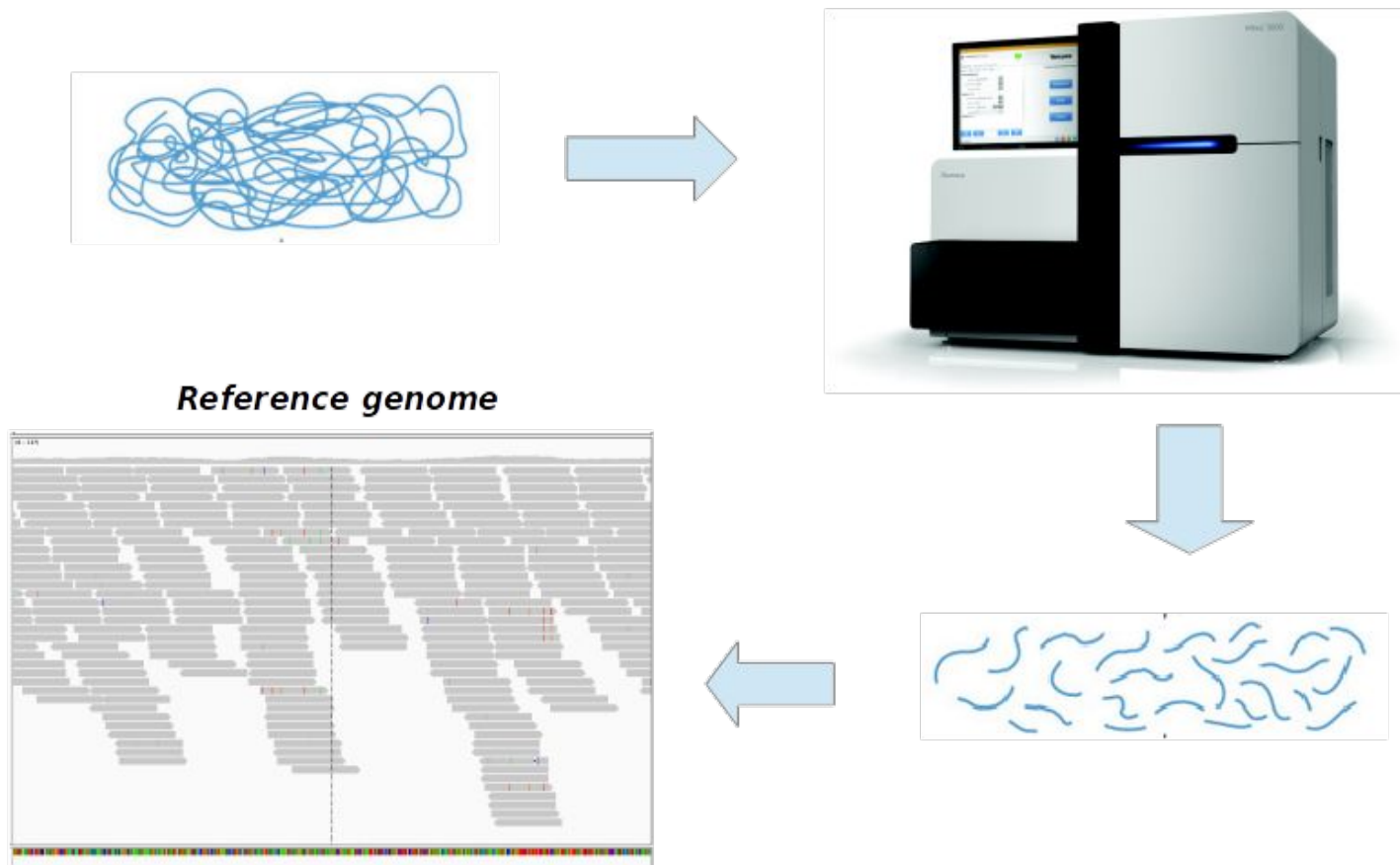
High throughput technologies: microarrays



UID	NAME	GWEIGHT	spo0	spo30	spo2	spo5
EWEIGHT			1	1	1	1
YAL003W	EFB1	1	0.23	-1.79	-1.29	-1.56
YAL004W		1	0.41	-0.38	-0.89	-1.06
YAL005C	SSA1	1	0.61	-0.07	-1.29	-1.29
YAL010C	MDM10	1	0.16	-0.15	-0.76	-1.25
YAL012W	CYS3	1	0.03	1.39	-0.84	-1.64
YAL015C	NTG1	1	-0.18	-0.18	-0.62	-1.32
YAL018C	YAL018C	1	-0.51	-0.62	-0.76	3.74
YAL025C	MAK16	1	-0.14	-3.32	-1.84	-1.12
YAL034C	FUN19	1	0.19	-0.03	-1.03	-1.29
YAL035W	FUN12	1	0.01	-1.47	-1.15	-0.69
YAL036C	FUN11	1	-0.15	-2.74	-1.79	-1.32
YAL038W	CDC19	1	-0.06	-1.89	-1.69	-2.32
YAL040C	CLN3	1	-0.17	-2.25	-1.69	-2.25
YAL054C	ACS1	1	0.51	2.6	1.9	1.7
YAL055W	YAL055W	1	-0.32	0.83	0.58	0.82

Computational Genomics

High throughput technologies: Next Generation Sequencing



Computational Genomics

Biological
knowledge

KEGG
pathways

Gene
Ontology

Regulatory
elements

MiRNA, CisRed
Transcription Factor
Binding Sites

Biocarta
pathways

InterPro
Motifs

Gene
Expression
in tissues

Bioentities from
literature

Clinical
knowledge

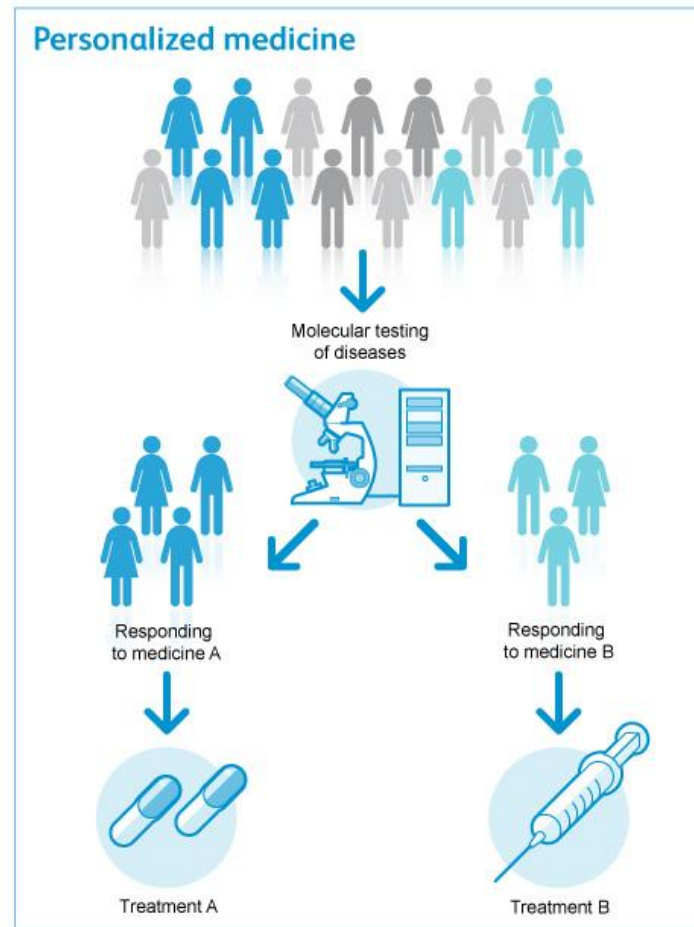
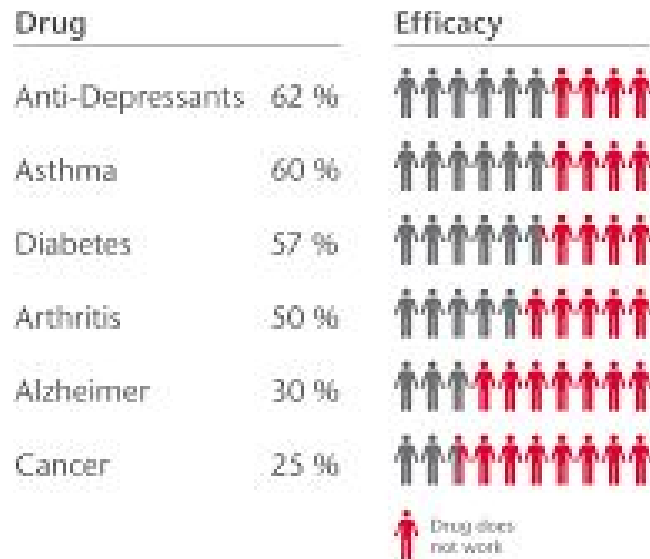
HUMSAVAR

ClinVar

HGMD

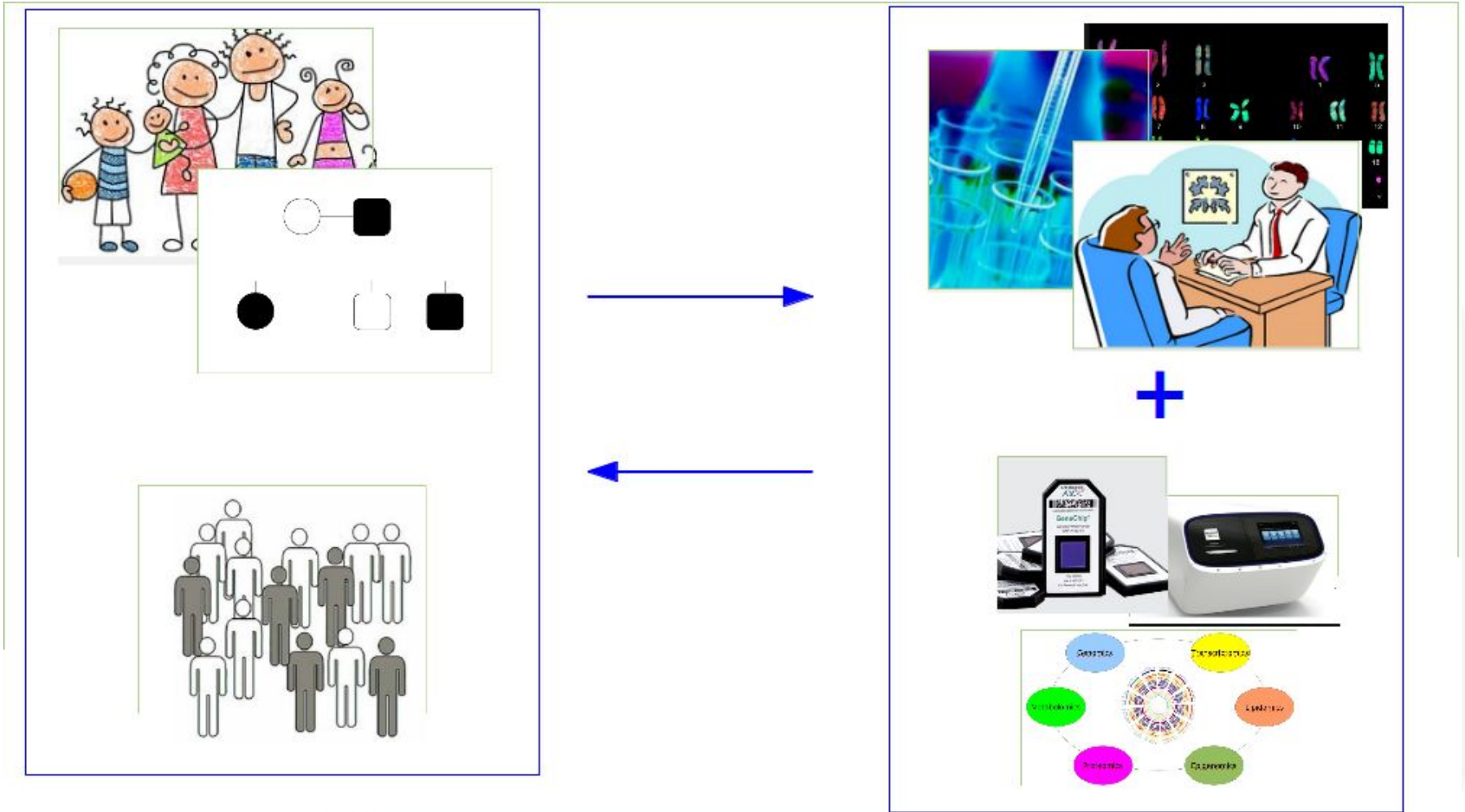
COSMIC

Computational Genomics



New molecular and diagnostic technologies can be used to match select groups of patients with treatments that may give them the best results

Computational Genomics



How do we work?


PRINCIPE FELIPE
CENTRO DE INVESTIGACION

Computational Genomics



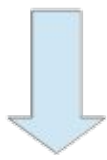
snow

TEAM



Network Miner

BierApp



Outline

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Primary Analysis

Secondary

1. Sequence preprocessing

2. Mapping

3. Variant calling

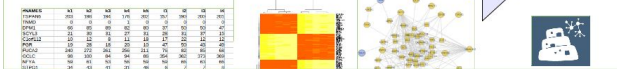
4. Variant prioritization

UPLOAD DATA

EDIT DATA

NORMALIZATION
+
DIFFERENTIAL
EXPRESSION

FUNCTIONAL
PROFILING



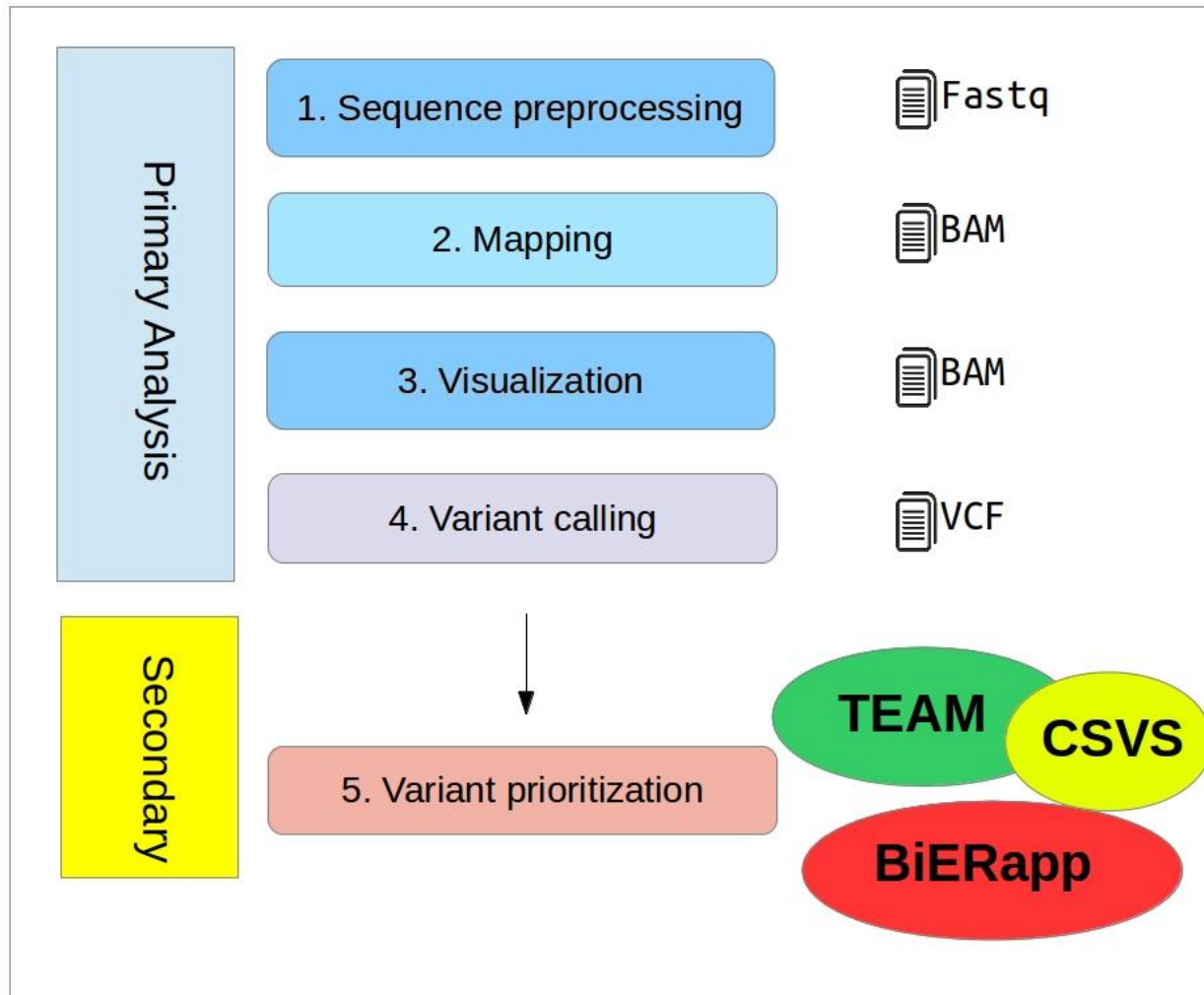
TEAM

BierApp

CSVs

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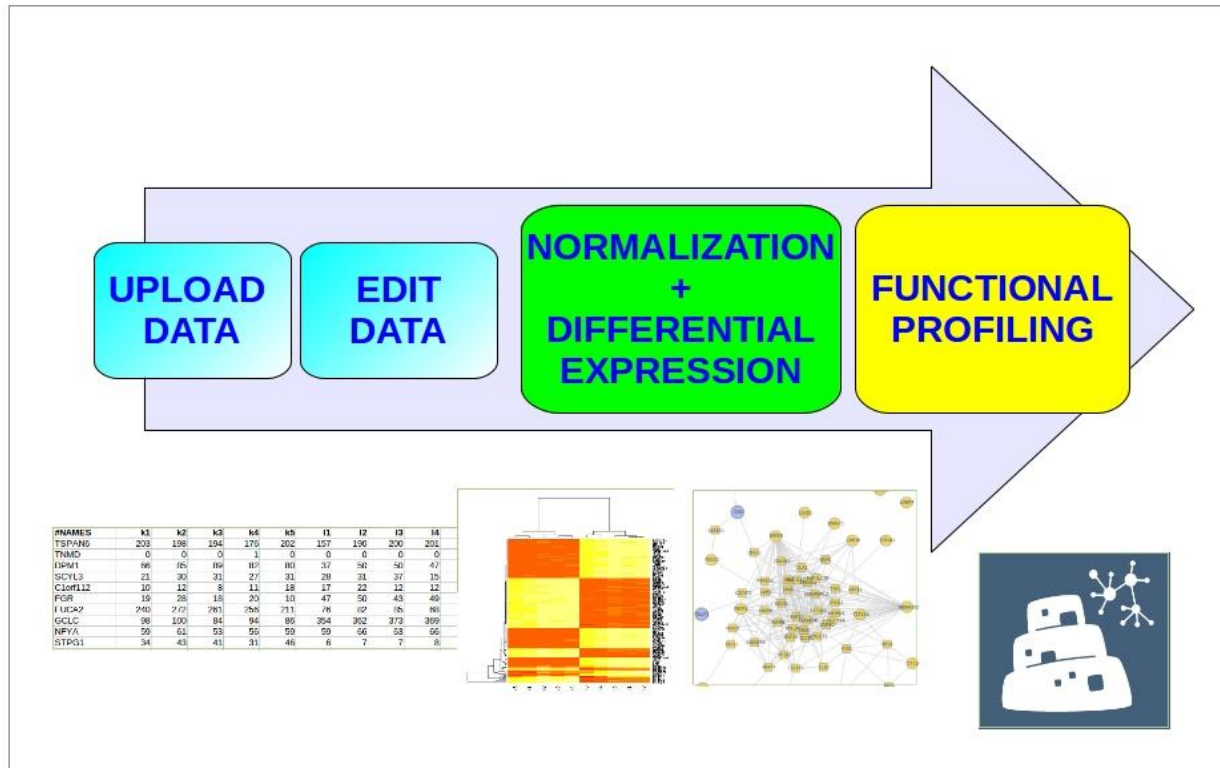
Day 1: Genomic variation studies



Wednesday 28
Registration + coffee
Course presentation
Introduction to NGS Technologies
NGS Data Analysis Pipeline
Lunch
Prioritization of variants and genes: <i>BiERapp</i> , <i>CSVs</i> and Genome Browsers (<i>Genome Maps</i> ,...)

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Day 2: Transcriptomic studies + functional profiling



Thursday 29

Panel of genes: design and analysis for clinical applications. *TEAM*

Coffee break

RNA-Seq Data Analysis from *Babelomics 5*

Lunch

Functional Profiling: *FatiGO* and Gene Set Analysis

Practical session

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<http://bioinfo.cipf.es/gda16ciberer/>

