



WP4: BIOINFORMATIC TOOLS. MAIN ACHIEVEMENTS AND FUTURE PLANS



WP4 Main achievements (AMU)

2

- **Pathogenicity prediction systems**
 - UMD-Predictor (<http://umd-predictor.eu>)
 - Human Splicing Finder - HSF (<http://umd.be/HSF3/>)
- **NGS data analysis system**
 - VarAFT (<http://varaft.eu>)
- **Bioinformatics systems for translational research**
 - Skip-E (<http://skip-e.geneticsandbioinformatics.eu>)
 - Crawfish (<http://crawfish.geneticsandbioinformatics.eu>)
 - NR-Analyzer



WP4 Main achievements (IBS)

3

- **ALFA: ALamut Functional Annotations**
 - Database of non-coding regulatory regions annotations
 - Includes promoters, transcription factor binding sites, transcription start sites, enhancers, microRNA target sites
 - Graphical web interface, REST API, command-line interface
 - Data also available in upcoming Alamut[®] Genova application



WP4 Main achievements (LUMC)

4

- Research support
 - Multi-omic biomarkers for disease progression/severity
 - Blood transcriptomics biomarkers reflecting changes in disease tissue



Drives tool development



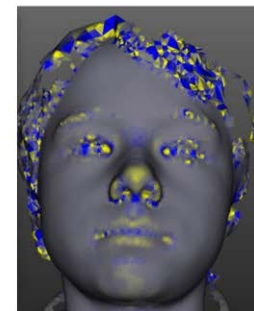
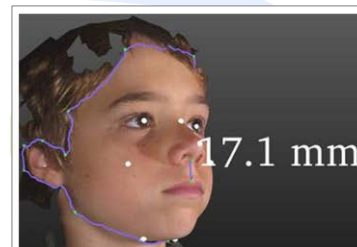
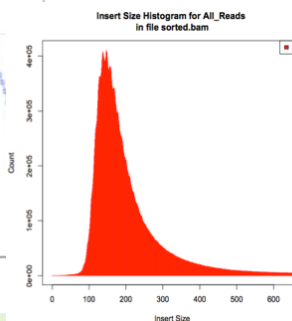
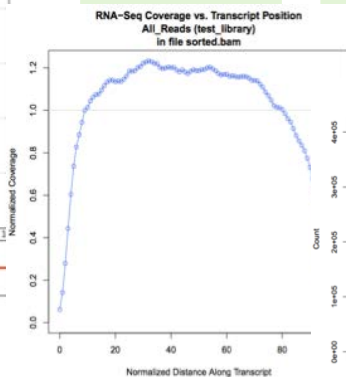
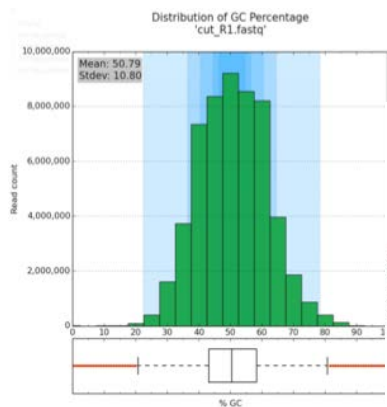
- Tool development
 - Multi-omics analysis and integration
 - Annotation services based on literature
 - Drug prioritization services



WP4 Main achievements (Murdoch)

5

- ❑ **RDRF:** Rare Disease registry framework
- ❑ **YABI:** integrative *-omics* Analysis Suite
- ❑ **MASTR-MS:** metabolomics suite
- ❑ **CliniFace:** 3D facial analysis tool



Government of Western Australia
Department of Health

The Undiagnosed
Diseases Program,
Western Australia (UDP-WA)
Information for parents and caregivers



HealthyWA
healthy.wa.gov.au



WP4 Main achievements (UPAT)

6

ePGA: Electronic Pharmacogenomics Assistant

Variant



Disease/Phenotype



“Patients with the **GG genotype** and **lymphoma** who are treated with **methotrexate** may have a **reduced risk of myelosuppression**”

Effect

Drug

- Efficiently browse PGx information
- Locate PGx variants in sequenced samples
- Support micro-attributions



WP4 Main achievements (UPAT)

7

A. Clinical annotations:

Pharmacogenomic associations

UNDER INVESTIGATION

As such, **NOT for direct clinical use!**

Format:

Gene/Variant + Drug + Disease

→ Annotation

B. Submission of novel PGx variants (same format as with clinical annotations)

Using the microattribution approach against an ORCID

C. Quality check of all PGx variants

Using the MutationInfo tool (Kanterakis et al., *submitted*)



WP4 Main achievements (UAVR)

8

- ❑ **Diseasecard:** Web-based IR tool (and API for developers) to access and relate diseases data already available on the Internet
- ❑ **COEUS 2.0:** Automated platform to integrate and publish biomedical data
- ❑ **Linked Registries:** Connection of the rare diseases patient registries through a Semantic Web layer
- ❑ **SCALEUS:** Semantic web migration tool that can be deployed on top of traditional systems in order to bring knowledge, inference rules, and query federation to the existent data
- ❑ **GeCo:** Statistical compressor for genomic sequences
- ❑ **Next 6 months:** Further improvements of the SCALEUS tool (query + visualization)



Next 6 months

9

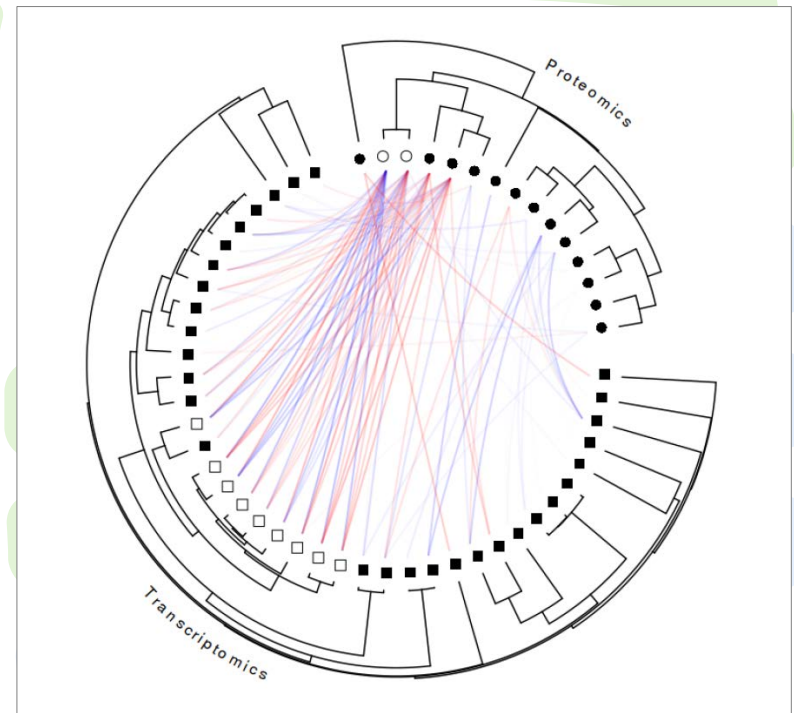
- **AMU: collaboration with the ELIXIR Human CNV community**
 - CNV analysis system
 - Interconnection of the RD-Connect platform with national CNV resources (BANCCO)
- **AMU: Bioinformatics systems for translational research**
 - Systems improvements (data, web access, validation)
- **IBS: Prediction of potential effects of regulatory variants on pathogenicity**
 - Machine-learning based on ALFA + allele frequency and classified variants databases



Next 6 months

10

- **LUMC: D4.19: Development of integrative -omics Analysis Suite (Multi-omics tool)**
 - Finish web tool user interface
 - Perform user testing
 - Integration / RD-Connect platform
 - Submit manuscript





Next 6 months

11

- **Murdoch**
 - **3D-facial system:** Integration into the RD-Connect platform

- **UPAT**
 - **ePGA:** Integration into the RD-connect platform

- **UVAR**
 - Further improvements of the SCALEUS tool (query + visualization)