Junta de Andalucía
Consejería de Salud y Familias
FUNDACIÓN PROGRESO Y SALUD

ADVANCED GENOMIC DATA ANALYSIS
CLINICAL BIOINFORMATICS AREA
FUNDACIÓN PÚBLICA PROGRESO Y SALUD (JUNTA DE ANDALUCÍA)

The **Clinical Bioinformatics Area** of the Fundación Pública Progreso y Salud (Junta de Andalucía) offers a data analysis service (single, trios or familiar groups) coming from NGS technologies (panels, exomes or genomes), in order to facilitate the clinician's decision making process about patient diagnosis and treatment, not only in rare and/or inherited diseases, but also in cancer cases.

In this context, the Clinical Bioinformatics Area has participated in projects such as NAGEN (Proyecto Genoma 1000 Navarra) and ENOD (rare diseases program of CIBERER), in addition to collaborating with clinicians from many hospitals and nationwide research groups.

Sequencing data (FASTQ or VCF files) are processed to extract and annotate all variants with information from different databases (ENSEMBL, OMIM, ClinVar...). Then, a prioritization of these variants is carried out based on different criteria (inheritance model, population frequency, phenotype, pathogenicity, conservation...), which allows to select finally those variants more likely to explain the disease.

Types of analyzed variants include:

- **SNVs and small INDELs.**
- **Structural variants** (large insertions or deletions, duplications, inversions, CNVs and translocations).
- **Mitochondrial variants.**

In certain cases (see table), and always respecting the anonymity, confidentiality and patient's consent, the following analyses are also available:

- **STRs (Short tandem repeats):** the number of repeats of certain sequences responsible for 25 diseases is analysed.
- **SMA (Spinal muscular atrophy):** the number of SMN1 copies is evaluated to detect affected or carrier individuals.
- **Secondary findings:** the presence of pathogenic or likely pathogenic variants related to pathologies different from the reason for consultation is analysed. They could entail:
    - Personal predisposition to disease.
    - Reproductive risk: cystic fibrosis and X-fragile syndrome.
    - Pharmacogenomic risk.
- **Familiar genetic counselling:** the presence of pathogenic variants for recessive diseases is assessed in couples interested in knowing about risks in future pregnancies.

Type of analysis	Sample type				
	Normal tissue genome		Tumoral tissue genome	Exome	
	Single	Familiar		Single	Familiar
SNVs and small INDELs	*	*	*	*	*
SV	*	*	*	(*)	(*)
STR	*	*			
SMA	*	*		(*)	(*)
Mitochondrial DNA	*	*	*	(*)	(*)
Predisposition to disease	*	*		*	*
Reproductive risk	*	*		*	*
Familiar genetic counselling		*			*
Pharmacogenomics (**)	*	*	*	*	*

(*) For exomes, SV analysis is limited. Equally, SMA and mitochondrial DNA analyses will depend on the exome capture kit used.
(**) Single nucleotide polymorphisms related to Pharmacogenomics.

Although the analysis can be fully carried out in our area, an analysis tool (MMP, Personalized Medicine Module) is completely available for clinicians, if necessary. MMP facilitates the filtering, analysis and interpretation of variants.

CONTACT INFORMATION

For further information, you can visit our webpage (http://www.clinbioinfosspa.es) or send an email to clinbioinfoarea@gmail.com.