

2023 Dossier Bioinformatics





Contents

- 1. Mission, Vision and Values
- 2. Quality
- 3. Genome One Easy 3.1. Genome One Reports
- 4. Genomics
 - 4.1. Sequencing + Bioinformatic analysis4.2. CNVs analysis
- 5. Transcriptomics
 - 5.1. Sequencing+ Bioinformatic analysis
- 6. Research
- 7. Contact

1. Mission, Vision and Values

Dreamgenics is formed by a **group of people passionate about our work**. We are aware about the impact that our performance can have on the health and quality of life of the people and therefore we make an effort to improve every day, revising all the processes that we do, looking for improvements to offer our clients a wider catalogue of services and more advanced products. In the same way, we feel committed to take advantage of our technology and great knowledge of our team to contribute to the development of Personalized Medicine, which enables the individual characterization of the disease of each patient and therefore receive the treatment according to its necessities.



Mission

Provide **clinicians and researchers** with genomic and bioinformatic solutions that help to understand the molecular basis of the diseases.



Vision

Each person working in Dreamgenics is proud of being able to contribute to **Precision Medicine** and we hope to continue improving every day to keep helping people.



Values

We try our best to perform our activities **respecting the environment**, following the established standards in the corresponding norm, minimizing our impact by reducing the residues produced and decreasing energy consumption.



2. Quality

The ISO certifications are documents that prove that a specific company is following the ISO norm. In this way, it is demonstrated that the stardards required by this legislation have been implemented and prove that the company is reliable in relation to the products and services that it offers. In Dreamgenics, **the Quality is the base of what we develop in our daily work.** We are certified since 2018 by the Asociación Española de Normalización y Certificación (AENOR) in the ISO 9001:2015, UNE-EN ISO 13485:2018 and UNE-ISO/IEC

marking (License number: 7157-PS).

27001:2017 norms, which allow us to offer to our clients products and services that fulfill the most demanding quality standards.

Sanitary registry

EIBT certification

Base (EIBT).

Dreamgenics is registered as a diagnostic center in the Registry of Centers, Services and Sanitary establishments of the Principado de Asturias with the **number C.2.5.6./6466.**

ISO Certifications and CE-IVD marking

ISO 9001:2015 International standard of certifiable character that regulates the Quality Management Systems.	UNE-EN ISO 13485:2018 Quality management norm that has been especially designed for manufacturers of sanitary devices and products.
UNE-ISO/IEC 27001:2017	CE-IVD marking Our software Genome One has CE-IVD

This recognition is awarded to companies whose practical knowledge and skills are based on the **application of new technologies**, through the development of basic research.

Dreamgenics has been recognized by the National Association of Spanish CEEIs (ANCES) as an Innovative Company of Tehnological

Dossier Bioinformatics 2023

information.

assurance, confidentiality and

integrity of the data and of the

3. Genome One Easy

Genome One Easy is a simplified version of our Genome One software which works with VCF files obtained by Illumina[®], MGI and Ion Torrent sequencing platforms. Genome One Easy has been designed to facilitate the daily work to its users through the automatization of multiple processes, thus **it is very easy and intuitive to use and does not require previous experience in bioinformatics**. Moreover, all the analyses performed with Genome One Easy follow the necessary requirements of the CE-IVD marking and the UNE-EN ISO 13485:2016 norm.

Simplify your daily work with Genome One Easy



Filter and revise the variants

Create a database with the revised variants

Main advantages

- It has an intuitive interface.
- It offers an analysis and samples repository.
- It allows the automatization of processes.
- Tracking the analyses is possible.
- Compative analysis of previous analyses is possible.
- It is compatible with any sequencing platform and library kit.
- It allows the generation of own databases by variant revision.
- It allows the configuration of views and filters.

						C46_VCF_Functions	eles vol annotation hg38 [048;	CF, Functionales human/GRCh38(primary,assembly	human(18/08Ch37 CARDIOL06IA I	Inne,Panel, PANy381CA8010	1			
itorie > Anatysis > 02200	0													
									1					
Analysis 17172	and I fillen	and Address of Concession and												
	the second second	and the second s							-					9
D Analysis 🗄 Varia	m 21	Report @ Outpute 02 C	Comments											
		F Grant and same						000	n Databases Punctional annotation	Functional scores	A1			
C Default with Vara	me w	FACETED BY Pressies effect to dis	ether STN AND Zypesity is HCM2.	ALT PRITERED BY Variation ACMO	Premium REVENED INCOME				Varsome	Giobal Review	Clinitar Disease	Clinitar Clinical Significance	Clinivar Conflict	
V	~	Variant Samples Cancer o	Antabases Clinical Database	n Population Databases	Functional annetation Fu	nctional scores All								
	~	ACMS Becondary Findings	Variana Premium	Varuime ACMO Premium	VP Classifications	VP User Exp	Variante Planutypes	Variante InterNance VP CIII	11647373		Absol Normation, Tamitol, 6 / Absol	BENION / LIKELY_BENION		
and the second second	- 8°			REVIEWED -					25563141	10 10 04	Cardiovascular phenetype	INTHODENIC		
C O Hetz	541	70		D danage (ant) des	GrunAD perumes aliale Reques	. No matching phonesype found to	r A0/49, tasket on gene information	25563141	10 O 10	Familial hypercholesterolemia / H	5 BENION		
0 00 cmar		Page 1	0	D Likely Barrige	8P4 PM2	The position is not conserved by	ry. No matching planningse hund h	er Allohk, beami en jeree information	25563140	(2° ×01,40	Hypercholesterolemia, familiai, 4	PATHODENIC		
	1000	-		The Design and	BAD BPA	GruntAD pertornes allele frequee	. No rist ting planetsise band 5	er AR based at gene effertuation for	55030879	9 BUUET	Hypercholestorolonia, familiai, 1	CONFLICTING_INTERPRETATIONS	Cliniver Conflict	
				D brief	antiera	Country and and the second	· No matching physicities based by	AD based on some information for	55039901	O NOME				
+ Cete	191	24		the second se	Antisesterpipes	Country along white frequency	v . Non-institution photoschicke Assett II	ADM. Search on service information 202012907 / 2	55110970		Eardial human halastaniania (b)	DENITY (1970) V DENITY		
- Ponsin affect	1141	Ter		a loss	Bet Me Inte	Graniff, range align frames	. In particul chances in all	a AD hand on man Alternation for						
Deliver OF	1601		-		and an elana lana lana	Design of the second			237042552					
🗋 war				and the second se	[] and heat mail fact has	and prove and repar		Active participation of the second second	21009015		Familial hypercholesterolemia / H	6 CONFLICTINGUNTERPRETATIONS	Clinive Conflict	
- without				o beilt	1) EC/EP+/EPE	CONCERNENT AND TREAT	 Restation provides and a 	P AN, Desire of give information inc	21045879		Hepatocellular carcinoma / Hyper	1 BENION		
C C C C C C C C C C C C C C C C C C C	1.1	. 10	0	C Bergs	ENITER'S SAME	GrankD exones allee hepano	y . No matching phanotype found to	 ACINE, based on gene information. \$7823772 (2). 	73448097		Alstrom syndrome / not specified	BENION		
C BROALDER	- 2	***	•	D Likely Serieph	1) SP4 (PM2	CADO + 139 is less than 103 is a	is the matching phanotype hand h	P AR, besid on providential to	73450719					
Can Shicks		10	•	D WR	1 1942	Variant nut found in provAD get	 No matching phenotype found to 	er AA, takket on gene information fra	73451027					
C Consectory		24	0	C Sarage	841(\$P4)\$P6	DisnAD econes alive hopiers	y. No matching planning in hand h	er All, based on gene information for	73490689					
	1	24	•	D Berrigs	1 801 802 (8F4	DionAD essnes alive Texpens	y No matching phanotype hand h	er AD, based on gene tofurnation the						
		14	0	D Bengs	() BATJAPA	DrumAD persities allele frequer	 No mething photospipe based by 	at AD, loased on gene information fro.					C Page	_
E2 01%		194	•	D Bergs	BAD (AP-4) BP1 (\$P\$	GranAD econes alter frequency	y. No matching phanologie hume h	r All, based on periorithermation by 29227844						

7. Contact

3.1. Genome One Reports

Visualization is essential for **a correct interpretation of the results** after the bioinformatic analysis. However, the quantity and complexity of the data obtained is a challenge for many users. Therefore, we hand in our bioinformatic results with our **Genome One Reports platform,** an intuitive web interface capable of managing a lot of data allowing the revision and visualization of the results in a simple way.

Support



4. Genomics

The NGS data analysis is a complex process that requires a lot of bioinformatic knowledge and great processing and storage resources. In Dreamgenics, we have **our own software and pipelines** for the analysis of NGS data obtained from the sequencing of **genomes**, **exomes**, **exome trios and gene panels**. Moreover, our service of personalized counseling allows us to offer the best option according to the necessities of each person.

The input files can be FASTQ, BAM and VCF from Illumina® or MGI and VCF files in case of Ion Torrent

Our analysis includes

- · Quality control of the sequences.
- · Alignment of reads against the reference genome.
- Detection of high-quality variants (SNVs, translocations and CNVs).
- · Filtering of variants that are common in the population.
- Annotation of the variants obtained with multiple databases and prediction algorithms.
- · Comparison between samples and identification of recurrent variants.





Res<u>ults</u>

The results are handed in using our web platform called **Genome One Reports** for the advanced visualization and interactive revision. Moreover, Genome One Reports allows the automatic generation <u>of custom reports</u>.



4.1. Sequencing + Bioinformatic analysis

We work with the **sequencing leader Macrogen** to provide our clients with a complete service that includes the sequencing of the DNA samples and the posterior bioinformatic analysis. Depending on the sample and the analysis, different libraries can be used, therefore we **assess our clients to choose the best conditions for their project.**

Contact

- 🚱 985 088 180 / 634 524 714
- info@dreamgenics.com



Genome
Sequencing + Bioinformatic analysis
✓ Illumina® NovaSeq 6000 platform
✓ Illumina® TruSeq Nano DNA Library
✓ Mean coverage >30x
\checkmark >1 μg of genomic DNA or >100 ng of FFPE
✓ 75-180 Gb/sample

- ✓ SNVs, indels and CNVs analysis*
- ✓ Results in Genome One Reports

*CNVs whenever the experimental design allows it.



4.2. CNVs analysis

Structural variants in the genoma include **Copy Number Variations (CNVs),** mobile genetic elements, inversions and translocations. The correct identification and characterization of structural variants is essential for the diagnosis of many genetic diseases and to establish prognostic, preventive and treatment strategies. In Dreamgenics, we do CNVs analysis with our Genome One software and **using an algorithm designed by us**¹. The results of the analysis are handed in using our web platform Genome One Reports.

Circos plot representing the identified CNVs

Interactive table summarizing the detected CNVs



¹Valdés-Mas R, et al. PLoS One. 2012;7(12):e51422.

5. Transcriptomics

The transcriptomic analysis allows the **quantification of gene expression** using NGS platforms. The interpretation of data obtained from RNA sequencing (RNA-Seq) has become a very demanding service due to its high sensitivity and precision. In Dreamgenics, we have a wide experience in the bioinformatic analysis of RNAseq and we offer our clients a personalized assessment

during the experimental design and in the interpretation of results.

We perform transcriptomic analysis from Illumina[®] FASTQ files.

Our analysis includes

- · Quality control of the sequences.
- Alignment of reads against reference sequences.
- · Quantification of gene expresión in the sample.
- · Differential gene expresión analysis between different samples.
- Study of gene ontology enrichment and pathways.
- Study of isoforms formed by alternative splicing*.
- Identification of other RNAs (smallRNAs and ncRNA)*.

*When the experimental design and the coverage allows so.

Results

The report with the results is handed in through our web plastform **Genome One Reports**, which allows the advanced visualization and interactive revision.

Data is represented in tables and graphs easy to interpret. Moreover, all the information is exported in different formats for its use in scientific papers.





5.1. Sequencing + Bioinformatic analysis

The libraries used in an experiment of RNA-Seq depend on the sample and type of RNAs that will be studied. We work with the sequencing leader Macrogen to offer our clients a wide variety of sequencing options, therefore we assess them to always choose the best conditions for their project.

Contact

- R 985 088 180 / 634 524 714
- info@dreamgenics.com

We offer an integral service

- - Initial assessment for your project



Support in the delivery of samples to Macrogen



Direct reception of the FASTO files





Support after the handing in of the results in Genome One Reports

mRNA-Seq Sequencing + Bioinformatic analysis ✓ Illumina[®] NovaSeg 6000 platform Illumina® TruSeg stranded mRNA Library ✓ mRNA analysis ✓ >lug of RNA with RIN>7 ✓ 30-40 M/sample At least 3 samples per condition

- Differential expression, ontologies and pathways
 - Results in Genome One Reports

*Removes ribosomal RNA and globin mRNA in blood samples

Total RNA-Seq Sequencing + Bioinformatic analysis ✓ Illumina® NovaSeq 6000 platform Illumina® TruSeg stranded Total RNA Library ✓ mRNA and IncRNA analysis ✓ >lug of RNA with RIN>7 ✓ 40-60 M/sample Ribo-Zero Globin treatment* At least 3 samples per condition Differential expression, ontologies and pathways

✓ Results in Genome One Reports

6. Research

Our mission as a biotech company is to provide clinicians and researchers with genomic and bioinformatic solutions that will decipher the molecular basis of the diseases. To achieve so, we carry out different I+D+i projects with the objective of developing new projects and services to offer our clients. In the same way, we participate with hospitals and research centers in projects associated with human health to which we contribute by providing the researchers with our expertise in NGS data analysis..

Research projects

European I+D+i projects



Highlighted Projects

MOLECULAR PROFILE STUDY IN PERIPHERAL BLOOD TO PREDICT THE **RESPONSE TO ICIS IN PATIENTS** WITH ADVANCED NSCLC

Project done in collaboration with the Instituto de Investigación Sanitaria de Santiago de Compostela (IDIS) and the Service of Medical Oncology of the Hospital Universitario de Santiago.

INSPIRING THE INNOVATION IN THE EARLY DETECTION OF INFANT HEARING LOSS IN THE SUDOE SPACE: TOWARDS A PERSONALIZED MEDICINE BASED ON GENOMIC DIAGNOSTIC TOOLS

European Project done in collaboration with the Clínica Universidad de Navarra and other hospitals of France and Portugal.





7. Contact



