

CIBERER Spanish Variant Server

Valencia, 28-30 Sep 2015



Alejandro Alemán
aaleman@cipf.es

Genomics Data Analysis CIBERER

Introduction

- 1) CSVS provides information about the variability of the Spanish.
- 2) It is useful for filtering polymorphisms and local variations.
- 3) CSVS currently stores information on 578 unrelated Spanish individuals.
- 4) URL: csvs.babelomics.org
- 5) Github: <https://github.com/babelomics/pvs>

CSVS

CIBERER Spanish Variant Server Search Stats

BIER **CSVS**

[➔ Try it now](#)

Overview

Welcome to the CIBERER Spanish Variant Server. CSVS was created to provide information about the variability of the Spanish population to the scientific/medical community. It is useful for filtering polymorphisms and local variations in the process of prioritizing candidate disease genes. CSVS currently stores information on 578 unrelated Spanish individuals. We accept submissions from WES or WGS. See the protocol for sending samples.

[Submission Protocol](#)

Supported by

Note:

CSVS web application makes an intensive use of the HTML5 standard and other cutting-edge web technologies such as Web Components, so only modern web browsers are fully supported, these include Chrome 36+, Firefox 32+, IE 10+, Safari 7+ and Opera 24+.

CSVS: created by Computational Genomics Department
Principe Felipe Research Center
2015

CSVS

Clear Search

Position

Chromosomal Location:
1:1-100000,2:1-100000

Gene:
BRCA2, PPL

Subpopulations

- MGP (267 healthy controls)
- IBS (107 Spanish individuals from 1000genomes)
- I Certain infectious and parasitic diseases
- II Neoplasms
- III Diseases of the blood and blood-forming organs and certain disorders involving the immune mechanism
- IV Endocrine, nutritional and metabolic diseases

Chr	Position	Alleles	Id	MAF						1000G MAF (phase 1)					1000G MAF (phase 3)					ESP 6500		SIFT	POLYPHEN	PhastCons	phyloP	Clinvar	
				Genotypes			Freq.			ALL	AME	ASI	AFR	EUR	ALL	AME	South ASI	East ASI	AFR	EUR	Eur. Ame.						Afr. Ame.
				0/0	0/1	1/1	./.	0 Freq	1 Freq																		

Page 1 of 0 items 1 - 0 of 0

Genomic context **Frequencies** **Phenotype** **Effect**

Min Max 159 1:1004529-1004687 Go! << >> >>>

Region overview Window size: 1271 nts
1003973 1004608 1005243

Detailed information Window size: 159 nts
1004529 1004608 1004687

Sequence - v ^
CCCTGCCGTGCCGGCACCTTCCCCCATCCGTAGCCCCCAGCCGCGCTTGTCCGAGGGCTGGACTGCGACACCGACGTCGCCGAGACGATCCAGCTGGTTTGGGGCAGCACCTGGGAATGGTGCAGACGGAGGCTCCGTACAAGTTCACGTACC

Gene - v ^

SNP - v ^
rs189753683 rs371314094 rs75134978

CSVS - v ^

CSVS

Clear Search

Position

Chromosomal Location:
1:1-100000,2:1-100000

Gene:
FAT3

Subpopulations

- MGP (267 healthy controls)
- IBS (107 Spanish individuals from 1000genomes)
- I Certain infectious and parasitic diseases
- II Neoplasms
- III Diseases of the blood and blood-forming organs and certain disorders involving the immune mechanism
- IV Endocrine, nutritional and metabolic diseases

Chr	Position	Alleles	Id	MAF							1000G MAF (phase 1)					1000G MAF (phase 3)					ESP 6500		SIFT	POLYPHEN	PhastCons	phyloP	Clinvar	
				Genotypes				Freq.			ALL	AME	ASI	AFR	EUR	ALL	AME	South ASI	East ASI	AFR	EUR	Eur. Ame.						Afr. Ame.
				0/0	0/1	1/1	/.	0 Freq	1 Freq	MAF																		
11	92085508	G>T	COSM545692	577	1	0	0	0.999	0.001	0.001	0.21	0.016	0.978	0.533	
11	92085623	C>T	rs76685441	566	11	0	1	0.99	0.01	0.01	0.010	0.010	0.000	0.000	0.010	0.002	0.007	0.000	0.000	0.000	0.007	0.005	0.002			0.992	0.655	
11	92085905	G>A	.	577	1	0	0	0.999	0.001	0.001			0.994	0.655	
11	92086301	C>A	.	576	2	0	0	0.998	0.002	0.002			0.708	-0.444	
11	92086458	G>A	.	577	1	0	0	0.999	0.001	0.001	0.23	0.063	0.538	-0.768	
11	92086472	A>C	rs139731877	577	1	0	0	0.999	0.001	0.001	0.001	0.000	0.000	0.002	0.001	0.002	0.000			0.256	0.533	
11	92086496	T>A	.	576	1	0	1	0.999	0.001	0.001	0.66	0.131	0.841	0.533	
11	92086513	C>T	rs10830902	523	47	2	6	0.955	0.045	0.045	0.050	0.040	0.000	0.130	0.040	0.055	0.039	0.004	0.001	0.150	0.044	0.056	0.138	0.01	0.182	0.929	0.533	
11	92086649	G>A	rs10830903	531	44	1	2	0.96	0.04	0.04	0.050	0.040	0.000	0.110	0.040	0.050	0.036	0.004	0.001	0.135	0.044	0.056	0.124			0.056	0.533	
11	92086662	A>G	rs16917409	577	1	0	0	0.999	0.001	0.001	0.030	0.010	0.000	0.140	0.000	0.036	0.009	0.000	0.000	0.133	0.000	0.001	0.098	0.75	0.032	0.167	0.655	

« < Page 1 of 21 > » items 1 - 10 of 203

Genomic context Frequencies Phenotype Effect

Region overview Window size: 1271 nts

1003973 1004608 1005243

Detailed information Window size: 159 nts

1004529 1004608 1004687

Sequence - v ^

CCCTGCCCTGCCGGCACCTTCCCCCATCCGTAGCCCCGCCCAAGCGCGTTGTCCGAGGGCTGGACTCGACACCGACGTCCCGAAGACGATCCAGCTGGTTTGGGGCAGCACTGGGGAAATGGTGCAGACGGAGGCTCCGTACAAGTTCACGTAC

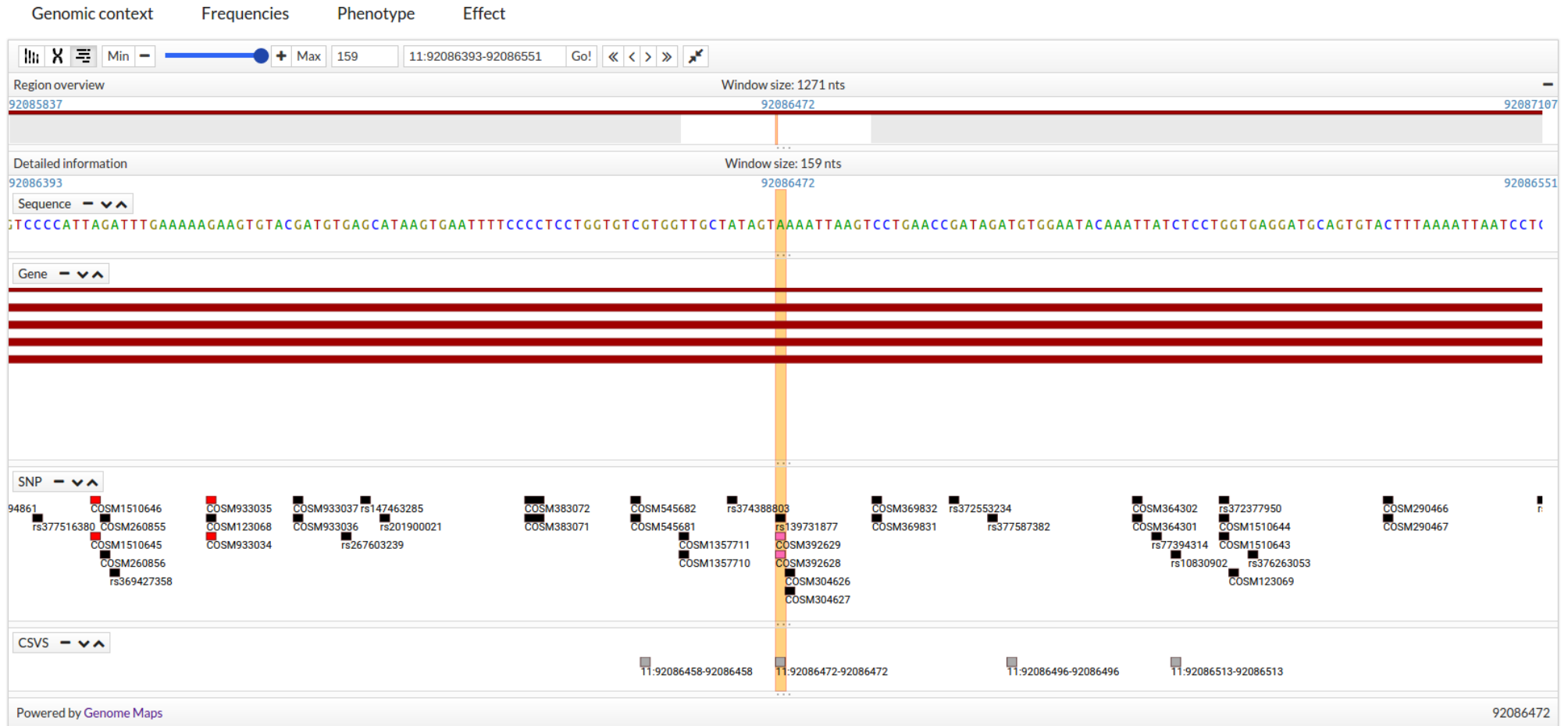
Gene - v ^

SNP - v ^

rs189753683 rs371314094 rs75134978

CSVS - v ^

CSVS



CSVS

