



Co-financed by Greece and the European Union

ARISTEIA II Programme - A genomic reference map of Greece. Studying the structure and history of Greek sub-populations and the Hellenic diaspora.

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Website- <http://genomap.mbg.duth.gr>

SNPs: databases, programs and applications

1. GENOME BROWSER - University of California Santa Cruz <http://genome.ucsc.edu/>
2. Select genome browser (link appears on the left)
3. You can search the GENOME BROWSER by gene names, physical position (in bp) or SNP names (rs numbers).
4. Select build March 2006 (build 36) – What are genome builds? Which is the most recent genome build?

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5. Search for CFTR
6. Select cystic fibrosis transmembrane conductance
7. A map of the region comes up. The image is interactive. If selected, you can view all SNPs in the exonic and intronic regions of the gene.
8. SNPs are colored according to their function. What color corresponds to missense SNPs?

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9. What genes are found before and after the CFTR gene on this same chromosome

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10. From the UCSC GENES section select the link to CFTR
 11. What is the function of CFTR. What phenotypes is it linked to?
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12. Name a type of tissue with very high expression of CFTR

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13. Go back to the previous page with the interactive map

14. Search for rs766874. Click on the name of the SNP.

15. What is the physical position of rs766874? (in base pairs – bp)

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16. What are the alleles observed at this SNP?

17. Click on the dbSNP link

18. You are now at dbSNP – all SNPs are deposited in this database and are assigned a code – their “rs number”.

19. At FASTA SEQUENCE look for the sequence around this SNP. R stands for the polymorphism using the IUPAC nucleic acid code system

20. What are the IUPAC nucleic acid codes?

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21. Return to dbSNP for rs766874. Give the sequence before and after the SNP (10 nucleotides before – 10 after).

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22. From the GeneView section, select cSNPs and GO

23. You can now investigate all the coding SNPs across the gene.

24. What is the function of rs374705585?

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25. What is the function of rs369464175?

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26. What is the function of rs1800136?

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27. Click on the link for rs1800136.

28. From INTEGRATED MAPS select the icon for the 1000 Genomes project.

29. What is the frequency of the A allele in Finns?

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30. Open a new tab. Go to <http://hapmap.ncbi.nlm.nih.gov/>

31. Go to Genome Browser (release 28) – search for CFTR

32. Here are the different SNPs that have been typed on the HapMap populations.

33. What is the allele frequency for allele A of SNP rs1800136 in Mexicans, Europeans and Maasai?

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34. Compare allele frequencies of SNPs across the CFTR gene in Greeks versus other Europeans (Greek data available at <http://genomap.mbg.duth.gr>)

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35. Why do allele frequencies differ in different populations?

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