

Single Nucleotide Polymorphisms (SNPs)

By
Amira
Jhelum
Rahul
Shweta



Recap of the Project Goal

- The goal of this project is to compare the distribution of **SNPs** over certain genes for two different organisms(Humans and Chimpanzees).





Papers to be discussed..

“Selection on Human Genes as Revealed by Comparisons to Chimpanzee cDNA”.

Ines Hellmann, Sebastien Zollener,
Wolfgang Enard, Ingo Ebersberger,
Birgit Nickel and Svante Paabo

Abstract

- For better understanding of the evolutionary forces that affect human genes
- 5055 ESTs were sequenced from the Chimpanzee and compared to the human counterparts
- Comparison with intergenic Chimpanzee DNA shows that 39% of silent sites in protein coding regions are deleterious and subjected to negative selection

Abstract continued

- Divergence between human and chimpanzee compared with the extent of nucleotide polymorphisms among humans in same sequences.
- Positive selection has a considerable influence on 5' UTRs
- CpG dinucleotide exhibits a different substitution pattern within 5'UTRs as compared with other parts of the genome.

Highlights of the paper

- This approach leads towards the understanding of the evolutionary forces that shape the human genome.
- Chimpanzees differ from humans by an average of 1.2% in overall genomic DNA and are estimated to have shared a common ancestor with humans 4.6-6.2 million years ago.
- It gives us an insight how the genes have diverged between humans and chimpanzees

Abstract of Paper 2

“The Essence Of SNPs”

By Anthony J. Brookes

- Single Nucleotide Polymorphisms (SNPs) are abundant forms of genetic variation.
- Distinguished from rare variations by a requirement for the least abundant allele to have frequency of 1% or more.

Abstract..continued

- SNP definition and its basics
- SNP discovery and scoring
- Population genetics and linkage equilibrium
- Complex phenotypes and genome variation
- SNP based association studies
- The characterization of human SNPs and their role in phenotypic determination represents a truly milestone project, for while humans beings are clearly much more than just “bags of DNA”, perhaps on the individual level we are little more than “sacks of SNPs”.



Paper 3

**“Human-Chimpanzee DNA Sequence
Variation in the Four Major Genes of the
Renin Angiotensin System”**

**Cecile Dufour, Didier Casne, Derek Denten, Jean
Wickings, Pierre Corvol and Xavier
Jeunemaitre**

Abstract

- The Renin Angiotension System(RAS) is involved in blood pressure control and water/sodium metabolism
- Genes encoding the proteins of this system are AGT,REN,DCP1 and AGTR1
- Observations within and between the four chimpanzees genes
 - o Nucleotide diversity at noncoding sites was similar
 - o Nucleotide diversity at nonsynonymous sites was low except for the AGT gene
 - o Nucleotide diversity at the synonymous sites, which was dependent upon the G+C content was high except for the AGTR1 gene

Importance of the paper

- Analysis of the polymorphisms within the species and divergence between species shed light on the evolutionary constraints on the genes
- Comparison of the pattern of mutation at polymorphic and fixed sites between humans and chimpanzees suggested that the high G+C content of the DCP1 gene was maintained by positive selection at its silent sites
- 68 ancestral alleles for the human RAS genes are proposed and the implications for their use in future hypertension susceptibility association studies is discussed.

Steps ahead..

- Analysis of Chimpanzee and Human data
- Initial observations and calculations
- Refining of calculations
- Biological significance of conclusions and inference



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www.angelfire.com/sk3/compbio601

Thank you..