

# The distribution of single nucleotide polymorphisms across consecutive exons and introns of the human genome



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## Conclusions

distribution of SNPs among introns and exons is highly non uniform

1st exons → significantly more SNPs

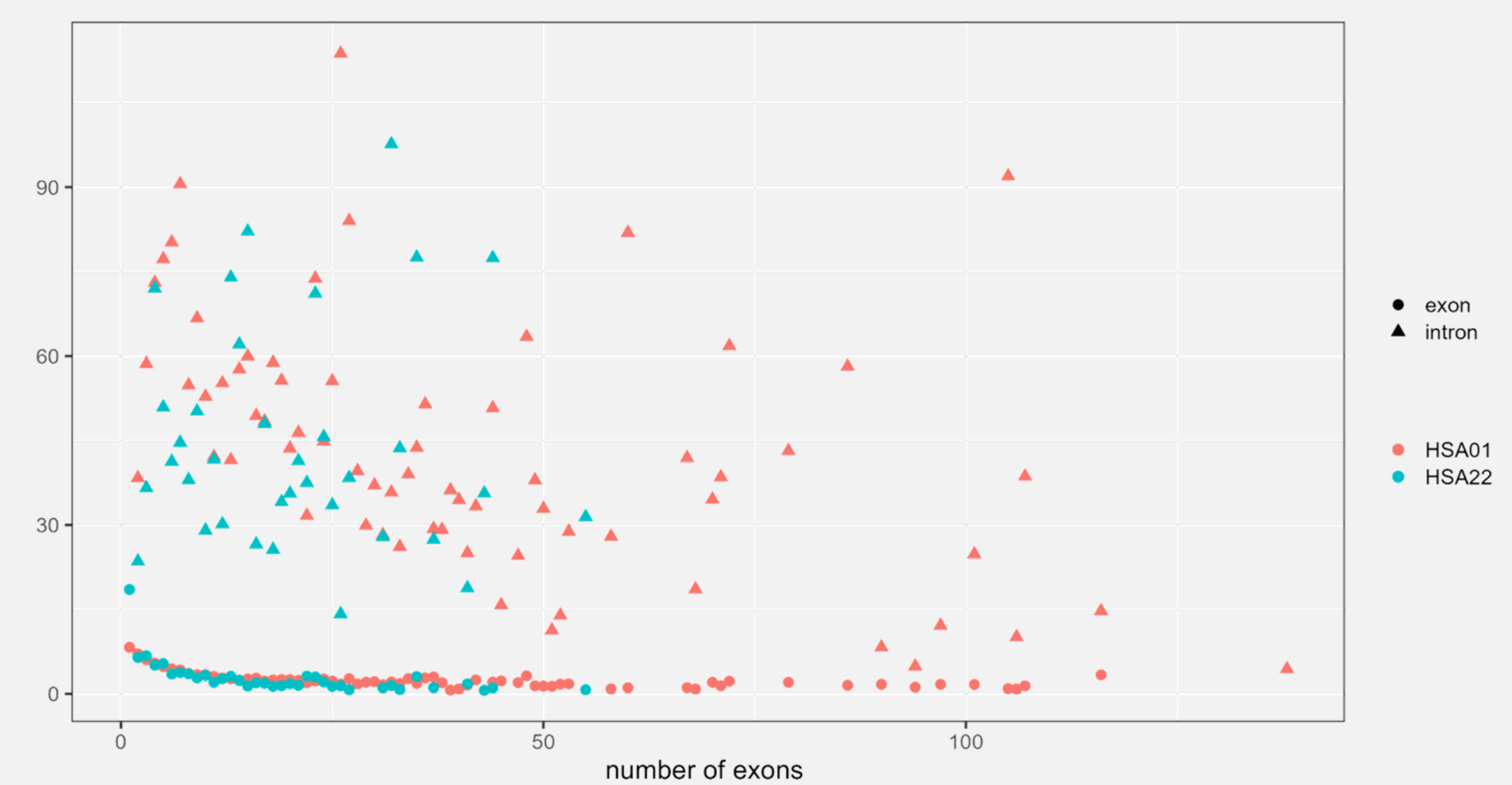
1st introns → significantly more SNPs

## Motivation

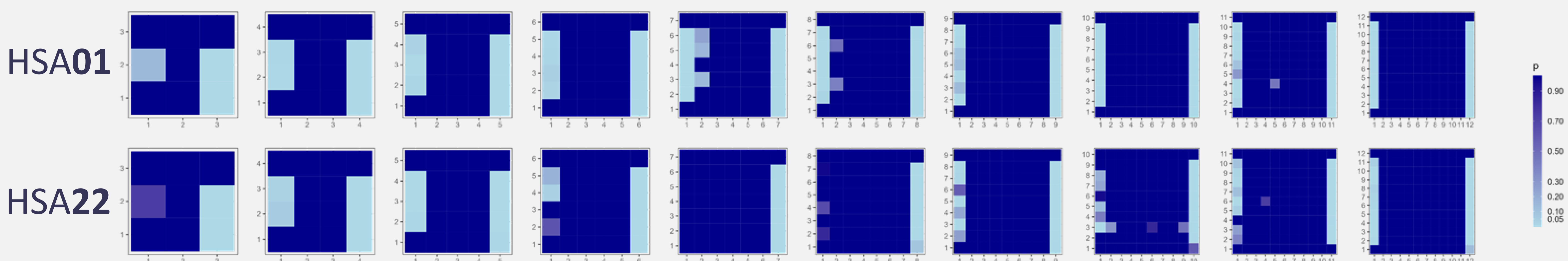
examine the numbers of SNPs in consecutive introns / exons

## Material

- 1,222 individuals of Polish origin  
→ 41,836,187 SNPs from WGS
- HSA01 (5,177 genes) & HSA22 (1,313 genes)  
→ 1,705,575 SNPs

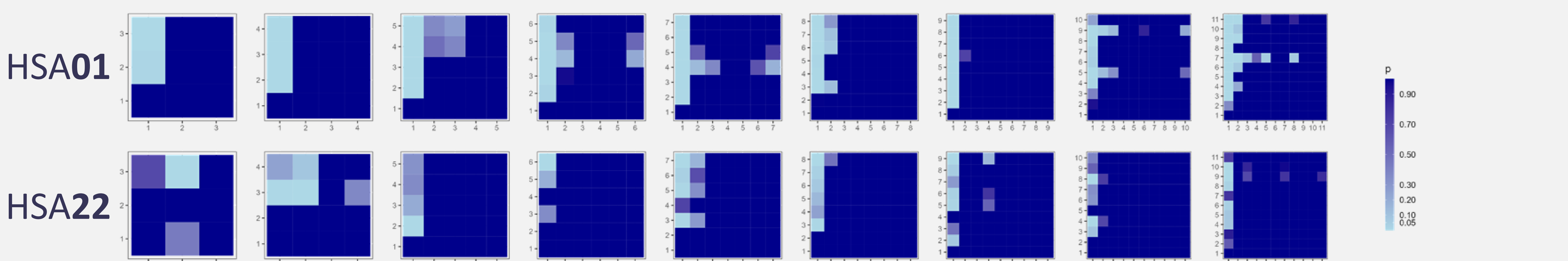


## Results



significance of pairwise comparisons of SNP counts in particular **exons**

P values → number of SNPs within the i-th exon (rows) < number of SNPs within the j-th exon (columns) → Conover test



significance of pairwise comparisons of SNP counts in particular **introns**

P values → number of SNPs within the i-th exon (rows) < number of SNPs within the j-th exon (columns) → Conover test