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Pathways and genes under positive selection in metabolic diseases

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Uncovering signatures of positive selection has been a long-standing interest in the field of genomics. The high prevalence of metabolic diseases such as diabetes has been suggested to be associated with positive selective pressures. Advantageous alleles increase in frequency and linked surrounding deleterious mutations rise infrequency as well, therefore the high prevalence of many diseases. High-density SNP maps of the human genome enable us to look for such regions involved in the susceptibility to diseases, particularly diabetes, obesity and metabolic syndrome. Firstly, we conduct a sensitivity analysis to evaluate the performance of several existing methods to detect positive selection. Out of the 7 methods (EHHST, XPEHHST, XP-EHH, iHS, nSL, XPCLR and hapFLK) that were compared under various demographic scenarios, XPCLR and iHS were found to perform best. These two methods were used for a genome scan of the HapMap Phase II database. Based on these results, we carried out an enrichment analysis to uncover signals enriched for positive selection. Two methods to conduct the enrichment analysis were used: the SUMSTAT statistic and Gowinda, an already available tool. String, Intact and Bio4j databases were also used to extract information about possible Protein-Protein Interactions associated with the 'interesting genes'. Our results indicate that selection has affected in a large percentage the evolution of diseases in the human history. More specifically, 64 pathways were discovered to have undergone selection and a total of 16 positively selected genes were found to have a direct or indirect links with diabetes, obesity or metabolic syndrome.

Biography

Alexandra Vatsiou is in her final year of PhD undertaken in the University Joseph Fourier in Grenoble. Her PhD is a Marie Curie program and the University Joseph Fourier is in collaboration with Era7, a Bioinformatic company in Granada, Spain and the Scottish Oceans Institute in the University of St Andrews. She completed her bachelor degree in University of Thessaly in Greece and her Master of research degree (MRes) in Computational Biology in the University of York, where she was awarded with a BBSRC scholarship.

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