Mutational load in signaling pathways of human populations and its functional consequences

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Signaling pathways constitute a formal representation of the knowledge existent on the consequences that the combined effect of gene activity has over the cell functionality in response to different stimulus. A non-negligible number of genes of these pathways are affected by the extensive mutational load recently uncovered by large scale genome sequencing projects. Nevertheless, to what extent such variation affects signaling pathways remains still unknown. Whole exome sequencing (WES) data of 1,092 individuals belonging to 14 populations from the 1000 genomes project have been used to derive a catalog of deleterious variants in genes involved in human signaling pathways. Probabilistic models of signal transmission along with gene expression data on 66 tissues were used to analyze the effect of the deleterious variants found in normal population has over the functionality of the different pathways studied. A comprehensive catalog of the effects that naturally occurring deleterious mutations cause in different pathways, measured in different populations and in 66 different tissues have been produced. The proportion of stimulus-response signaling circuits active in all the tissues are around 5% of the total number. It is very frequent that the genes carrying deleterious mutations have ultimately not an effect on signal transmission in the pathways in which they are located.

Biography
Rosa D Hernansaiz Ballesteros is a PhD Student at the Principe Felipe Research Center, at the Bioinformatics and System Biology Department. Licensed (BSc+MSc) in Biotechnology by the Polytechnic University of Valencia, she obtained an MSc in Bioinformatics by the University of Valencia. She also holds the position of board member in the Spanish Federation of Biotechnologists (FEBiotec), where she is responsible of the institutional and corporate communications, working with national and international entities in order to create and maintains satisfactory ties.

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