SORS: Characterization of regulatory variants in promoters with enhancer activity and their relation with human diseases

Objectives

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Abstract: Gene regulation is driven by the interaction of regulatory sequences, commonly categorized as either enhancers or promoters. Recently, using a modification of the STARR-seq assay, we identified sets of promoters with enhancer potential. Given that the majority of genetic variants associated with human diseases and traits (93.7%) have been found to be located in non-coding DNA, in this follow up analysis we set out to characterize regulatory variants in ePromoters. Using genetic variants associated with traits and disease (GWAS catalog), we found a significant enrichment of GWAS variants associated to Hematological Measurements ePromoters found in HeLa.

We hypothesize that genetic variants within ePromoters are likely to affect transcription factor (TF) binding. Therefore, we aimed to identify the relevant TFs interacting with these regulatory regions and look for variants disrupting TF binding. Particularly, we found variants affecting binding of TFs associated to inflammatory response.

Understanding ePromoters and the regulatory mechanisms that affect their dual function will help identify the causes of human diseases and traits.

Alejandra obtained her Ph.D. in 2012 from the Biomedical Sciences Program at the National Autonomous University of Mexico (UNAM), with a project developed under a co-tutorship between Dr. Collado, at UNAM, and Dr. van Helden at the Université libre de Bruxelles. Later on, she continued her postdoctoral training in The Hospital for Sick Children in Toronto with Dr. Michael Wilson, during this time she focused on developing comparative genomics projects. Dr. Medina has been committed to strengthen bioinformatics training in Mexico and Latin America. Since 2012 she has been collaborating on the organization of the International Workshops in Bioinformatics in Cuernavaca, Mexico, and now she participates as a trainer in the Wellcome Genome Campus Next Generation Sequencing Bioinformatics Course held in Santiago, Chile. Since her Ph.D., she has been focused on developing bioinformatic tools, and strategies to study gene regulatory mechanisms, most of the developed tools are now part of the Regulatory Sequence Analysis Tools suite (RSAT, http://rsat.eu/), where she is now one of the lead developers. Alejandra joined the International Laboratory for Human Genome Research at the National Autonomous University of Mexico as Junior Faculty in 2015, where using computational approaches, her research is focused on leveraging functional genomics data with genetic variation, aiming to identify variants and regulatory mechanisms related to disease.

Speakers

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