SNP-SIG 2013: the state of the art of genomic variant interpretation

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The success of SNP-SIG 2013 (Berlin, Germany), as confirmed by the number of participants and the interesting discussions, indicated the great interest of the community in the automatic annotation of genomic variants. SNP-SIG 2013 sessions focused on the annotation and prediction of structural/functional impacts of SNPs (morning session) and on the disease and evolution-related SNP perspectives (afternoon session).

In the morning session, chaired by Yana Bromberg, the two keynote papers were Ruth Nussinov, National Cancer Institute (Frederick, MD), and Manolis Kellis, Massachusetts Institute of Technology (Cambridge, MA). Dr Nussinov discussed her investigation of non-synonymous variants in cancer pathways (here, networks proteins of known 3D structure) to understand the relationship among linked phenomena, e.g. inflammation and cancer. Dr Kellis addressed the effects of genomic and epigenomic changes on gene regulation. Particularly, he talked about the making predictions of regulatory activity using epigenomic maps of human tissues and specific cell types to expand the annotation of non-coding regions and to provide mechanistic hypotheses of complex disease. This session also hosted four original work presentations by Christopher Yates, Imperial College of London (London, UK), Lukas Folkman, Griffith University (Brisbane, Australia), Martin Kircher, University of Washington (Seattle, WA), and Bjoern Stade, Christian-Albrechts-University (Kiel, Germany). All presentations described different methods for the annotation and prioritization of single nucleotide variants.

After the company presentation (Frank Schacherer, BIOBASE GmbH) and the poster session, Emidio Capriotti chaired the second session to discuss SNPs as effectors of change in disease and evolution. Paul Flicek, European Bioinformatics Institute (Hinxton, UK), and Alon Keinan, Cornell University (Ithaca, NY), presented highlights talks in this session. Dr Flicek focused on the use of comparative genomics analysis to reduce the search space of regulatory variants associated with rare diseases. Particularly, he presented a newly developed method for identifying the thousands of variants in regulatory regions associated with genetic disorders. The major novelty of this method is in the use of functional conservation of regions rather than sequence conservation, which is difficult to detect in non-coding stretches of DNA. Dr Keinan’s talk presented the results of a population genetics study describing the abundance of rare variants as an effect of the explosive growth of the human population since the Neolithic. The developed theoretical model based on the spectrum of allele frequencies is able to recapitulate the human demographic history. In addition, the model has been used to estimate the effects of the load of individual genetic variants in terms of complex disease risk. In the second session, there were also three selected presentations by Graham Ritchie, European Bioinformatics Institute (Cambridge, UK), Andrey Grigoriev, Rutgers University (Camden, NJ), and John Moult, University of Maryland (Rockville, MD). These talks addressed different topics, including the annotation and visualization of non-coding variants and the identification of potential drug targets using GWAS.

In the last part of the meeting, Steven Brenner, UC Berkeley (Berkeley, CA), summarized the results of the last edition of the Critical Assessment of Genomic Interpretation. This was an in-depth report on the discussions taking place in the community of scientists, involved in the development of new computational methods for genomic interpretation. The report mostly addressed issues of proper testing datasets selection in the assessment of the available techniques.

Finally, all speakers and participants were invited to discuss the ‘hot’ open topics in the field as part of a round-table session chaired by Sean Mooney, Buck Institute (Novato, CA). The participation of leading scientists in this discussion was an opportunity to define most important challenges for the field. This session also addressed the role of computational biology and bioinformatics in delivering algorithms into clinical settings—no small task for the near future of personalized medicine.

In the next few months, a SNP-SIG 2013 BMC Genomics Special Issue, edited by Yana Bromberg and Emidio Capriotti, will be published. This collection of selected works presented in Berlin will be the third issue in the series of proceedings highlighting the lessons learned at the SNP-SIG meeting.

After 3 years of meetings, we believe that our SIG has become a well-established venue for the discussion of major advances in the field of genomic interpretation. This is evident from the enthusiastic participation of colleagues and young scientists who strongly contribute to the success of this meeting. In the near future, we expect that our SIG could play a central role in the organization of an interdisciplinary Community Of Special Interest (COSI) to understand the relationship between genomic variation and disease.

See you soon at VarI-SIG

This year the SNP-SIG is undergoing some changes, which will further promote our efforts in genome interpretation. SNP-SIG
will change its name to VarI-SIG (Variant Interpretation Special Interesting Group) to reach out to scientists investigating all the different types of genetic variants. This expansion is likely to bring in more interesting people and discussions into the community. We are currently working on the organization of VarI-SIG meeting (July 12, 2014) that will be held in the context of the ISMB 2014 (Boston, MA). Further information about the meeting is available on our Web site (http://varisig.biofold.org).

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