International Society for Computational Biology Honors Goncalo Abecasis with Top Bioinformatics/Computational Biology Award for 2013

Christiana N. Fogg[1] and Diane E. Kovats[2]

[1]Freelance Science Writer, Kensington, MD, USA and [2]International Society for Computational Biology, La Jolla, CA, USA. Email: dkovats@iscb.org

INTRODUCTION

The International Society for Computational Biology (ISCB; http://www.iscb.org) honors a scientist each year for their outstanding achievements. The ISCB Overton Prize honors an early or mid-career scientist who has already made significant and enduring contributions to the field of computational biology. Dr Goncalo Abecasis of the University of Michigan is the 2013 recipient of the Overton Prize.

Dr Abecasis was selected the by ISCB's awards committee, which is chaired by Dr Alfonso Valencia of the Spanish National Cancer Research Center (CNIO) in Madrid. Dr Abecasis will receive his award and deliver keynote address at the ISCB's 21st annual Intelligent Systems for Molecular Biology (ISMB) meeting. This meeting is being held jointly with the 12th European Conference on Computational Biology and will take place in Berlin, Germany on July 21–23, 2013 (http://www.iscb.org/ismbeccb2013).

2013 ISCB Overton Prize: Goncalo Abecasis

Goncalo Abecasis was drawn to biology ever since he was a child. 'From a young age, I have always been fascinated with understanding how life works', said Abecasis. He fondly recalls spending Sundays at a bookstore with his parents and gradually collecting a small library of wildlife books.

But it was his experiences in a high school computer programming club that opened his eyes to an entirely different field.

'Although I didn't know it at the time, a key skill that later contributed to my success in genetics was my interest in computer programming', recalled Abecasis. 'The club was meant to keep us busy and out of trouble, but they did encourage us to try programming and pointed us in the direction of very useful techniques, like object oriented programming and the like'.

Human genetics appealed to Abecasis as he pursued his undergraduate studies at the University of Leeds, and he landed a position in the laboratory of Dr Mary Anne Shaw studying 'how genetic variation in the interleukin-1 gene cluster, a set of immune genes where variation was easy to measure with then available techniques, was related to infection by *Leishmania* and other tropical parasites'. This experience proved invaluable for helping Abecasis to receive funding for his PhD training in the laboratory of Dr William Cookson at the University of Oxford.

Cookson's laboratory at the Wellcome Trust Center for Human Genetics at Oxford was studying genes that contribute to asthma susceptibility. In the late 1990's and early 2000's, Abecasis described this Center as 'a mecca for human geneticists at the time, with great support from the Wellcome Trust, and lots of smart people trying new ways to run genetic studies and looking to make rapid progress in many different traits'. Abecasis also recalled that 'as we pushed the limits of the sequencing and genotyping technologies of the time, we were soon generating datasets that were beyond the reach of existing analysis tools and methods'.

Abecasis saw that, 'It was easy to realize that new analysis methods and computer software were needed—and being in Oxford, working at the Wellcome Trust Center, was just the right place to be'. With Cookson's support, and under the co-mentorship of statistical geneticist Dr Lon Cardon, Abecasis developed software to tackle the analysis of large genetic datasets. Abecasis remembered dealing with many software bugs along the way, but then, as now, he repeated the mantra to himself that 'all software is buggy, and this is no exception!'

Abecasis was pursuing his PhD at the same time as the race to sequence the first human genome was wrapping up. As the field of genomics was emerging, he realized that several of the methods he had developed could be used to look at how 'individual genomes differed from this initial sequence and to understand how these differences contribute to the great diversity we see among people today'. The application of these methods to genome data also shifted his research focus away from 'laboratory methods, technology and data generation', and towards 'issues related to study design and analysis'.

Abecasis's unique knowledge and training in human genetics, biostatistics and computational analysis landed him a faculty

position in the Biostatistics Department at the University of Michigan. Abecasis recounted the support and mentorship of Dr Michael Boehnke in the department. 'Mike somehow convinced the Biostatistics Department at the University of Michigan to take a flutter on me, when I had just finished my PhD and had much less formal training in statistics than most of my colleagues. He has always been generous with his time, and I probably can't count the times that I have interrupted him in his office, bounced some ideas off him, and came out energized and thinking about something new to try'.

Along with Boehnke, Abecasis acknowledged how fortunate he has been in the mentorship he received throughout his training, including the volunteers who taught him to code in his high school club. 'As I knew them, I remember my mentors as demanding, generous with their time, unrelentingly positive and encouraging, and totally transparent. It is obviously a standard I'd like to meet, although I doubt I am there yet.'

Their example also motivates Abecasis to be a good mentor. 'It is great to set a student free on an interesting open problem and have them solve it. You can do so much more with a few good trainees than you could ever accomplish on your own.'

Abecasis's research, and the field of human population genetics in general, has been transformed by the advent of high-throughput genetics. 'We now have very clear answers about the degree and structure of genetic variation in the world today, but have also gained a lot of detail on human population history—including very ancient events, like admixture with Neanderthals', said Abecasis.

Abecasis's laboratory is now focused primarily on identifying genetic variants relevant to human disease. They look at linkage disequilibrium within human genomes to describe, 'how groups of variants are shared among individuals'. One of the observations that Abecasis's group (and others) made several years ago, which he recalled as being surprising, was 'that much of the genetic variation in any individual could be recovered accurately by comparing each individual with a reference set of individuals, and, more recently, we have used the process to make it relatively inexpensive to sequence large numbers of individuals. At our last count, >30 000 human genomes had been sequenced using our 'low-coverage' linkage disequilibrium-based approach.'

One of the highlights of Abecasis's career was being invited to the White House in 2010. 'I was thrilled. I remember I had very short notice (perhaps a couple of days) and had to rush and find something to wear, recalled Abecasis. Although it is cheesy, it is really amazing to live in a country that functions so much like a meritocracy. I didn't have to write a check, join a committee, vote—anything. I had a good idea about how to sequence a lot of genomes more rapidly, proposed it, and not only did I get funded to try it out (it worked, by the way), but my work was selected as one of the highlights for Vice President Biden's speech on the importance of technology development and biomedical research.'

Abecasis described the importance of collaborations to his research and is a strong proponent of sharing data and software tools. 'So many great discoveries and advances come from bringing in insights, ideas and approaches from a different field', said Abecasis.

But Abecasis also agreed that data sharing is not without challenges. 'There are legitimate concerns about protecting the identity and privacy of research subjects and, once in a while, people do use data you share pre-publication to gain an advantage', said Abecasis. 'Still, there is no doubt we are moving in the right direction—expectations for data sharing and collaboration are so much more open than when I started'.

Abecasis has felt fortunate to work with so many great collaborators. One of his most interesting collaborations has been his work with Dr David Schlessinger, Dr Francesco Cucca, Dr Serena Sanna and many others on the 'SardiNIA project'. 'When I first met David, and he described the idea of conducting a thorough genetic study in an isolated valley in Sardinia, I never thought it would happen', remembered Abecasis. 'It seemed so ambitious. But David and our Sardinian colleagues have boundless energy and real dedication, and the study probably accounts for most of my highly cited papers!'

'The work of Goncalo underscores the importance of the theoretical developments and their implementation in computational methods for the progress in current biomedical research, bringing genomic information closer to the study of the complex genetic basis of common diseases', said Alfonso Valencia, chair of the ISCB's Awards committee.

Abecasis feels truly honored and humbled to be the 2013 recipient of the Overton Prize. Abecasis also hopes that 'If this award encourages members of the ISCB to bring some of their considerable expertise to bear on the big open problems in genetics, that would be an amazing outcome.'

Additional Information

During the past 20 years, the ISMB conference has grown to become the world's largest bioinformatics/computational biology conference, and ISMB/ECCB 2013 will be the year's most important computational biology event globally.

The ISMB conferences provide a multidisciplinary forum for disseminating the latest developments in bioinformatics/computational biology. ISMB/ECCB brings together scientists from computer science, molecular biology, mathematics, statistics and related fields. Its principal focus is on the development and application of advanced computational methods for biological problems. ISMB/ECCB 2013 offers a strong scientific program and the broadest scope of any international bioinformatics/computational biology conference.

ISMB/ECCB 2013 takes place July 19–23, 2013, at the Messe Berlin (ICC Berlin), Germany. For 2 days preceding the conference, additional opportunities including Satellite Meetings, Junior Principal Investigator Symposium, Student Council Symposium and a selection of Special Interest Group Meetings and Tutorials are all offered to enable registered participants to learn more on the latest methods and tools within specialty research areas.