

Pinpointing the genetic basis of disease

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The human genome contains 3.2 billion nucleotides, chained together in long, linear sequences of DNA. Differences in single nucleotides known as single-nucleotide polymorphisms (SNPs) account for a lot of the genetic variation in a population, and can be associated with disease. Jianjun Liu, head of the Human Genetics group at the A*STAR Genome Institute of Singapore (GIS), is using genome-wide association studies to scan large volumes of genetic data for that single unit out of place in

people suffering psoriasis, a condition characterized by scaly, flaky or itchy skin that affects approximately three per cent of the world's population.

"Genome-wide association studies can sharpen our understanding of disease mechanisms, and help us to develop new treatments or strategies for identifying people at high risk of developing a disease," says Liu.

Liu joined the GIS in 2002, becoming head of the Human Genetics group in 2007 and deputy director of GIS research programs in 2012. He has spearheaded the institute's involvement in huge international collaborations to identify the genetic basis of disease inheritance and susceptibility. "Human genetics analysis is a powerful tool for dissecting many different kinds of disease with significant family risk, and thus genetic susceptibility."

Liu searches for observable traits, known as phenotypes, which clearly demonstrate a disease's [genetic basis](#), and then sets about finding doctors with expertise in that disease and clinical resources. "Once those two conditions are in place, I apply my tools for human genetic analysis to help doctors gain a first-hand understanding of the kind of variants and related genes that are involved in the disease environment," he explains. Liu has taken this approach to study a wide range of diseases, from cancer and neurological conditions, such as schizophrenia, through to autoimmune disorders.

Liu's recent interest in psoriasis was piqued when he noted that the condition has an estimated heredity of up to 80 per cent. "It was very natural for me as a geneticist to pick psoriasis," he explains.

Many genetic studies have looked at the disease, but Liu was the first to conduct a trans-ethnic genome-wide meta-analysis across different ethnic populations for psoriasis. Liu and his team compared [genetic data](#)

from seven independent Chinese and Caucasian population samples, consisting of information on about 4 million SNPs in more than 5,000 patients and 8,000 healthy controls, to see if there were any significant psoriasis-related genetic differences between patients and healthy controls as well as the Chinese and Caucasian cohorts.

Surprisingly, the team discovered that of the 45 genetic loci associated with psoriasis susceptibility—four of which were discovered during the course of the study—ten were only found in the Caucasian population. This goes some way towards explaining why Caucasians are approximately ten times more likely to experience psoriasis than the ethnic Chinese population.

Liu notes that this type of cohort-based study is extremely useful for understanding differences in [genetic susceptibility](#) to [disease](#) at the ethnic-population level and for identifying [high risk](#) groups.

Moving forward, Liu emphasizes the need for more studies focusing on non-European populations, including Asian populations, and for collaborative research involving molecular biologists, physicians and geneticists. He hopes to work closely with clinicians to design more effective, targeted treatments that account for ethnic differences.

More information: Xianyong Yin et al. Genome-wide meta-analysis identifies multiple novel associations and ethnic heterogeneity of psoriasis susceptibility, *Nature Communications* (2015). [DOI: 10.1038/ncomms7916](#)

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