

The origin and genetic mystery of blue eyes

Evolutionary Physiology Lab. Professor
Yoko Satta

Q. What is the one thing you want to know the most right now?

Because I am greedy, it is hard for me to pinpoint one thing I want to know the most. Therefore, I list things that I want to know right now.

Firstly, I would like to know when and where the ancient human, such as Neanderthal and Denisovan, genomes have introgressed into our genome. And I know the effect of this introgression to our genome and the modern human evolution.

Secondly, I would like to know the origin of blue eye. Most of our Japanese have black or brown eyes, but many European have blue eyes. Why? Was there any advantage of having blue eyes for European survival? Or Was there any chance to have high frequency of blue eyes in an ancestor of European? The relationships between genetics and morphological traits was not so easy.

Q. Could you share your thoughts on the future prospects of this field?

It is important to define “a reference” in comparison, for example, what is different between individuals, what is different compare to chimpanzees, what is different compared to gorillas, and so on.

Currently, the analysis to identify what is different among individuals in the human genome and how these differences at the genome level relate to differences in appearance is called genome-wide association analysis (GWAS). These GWAS were

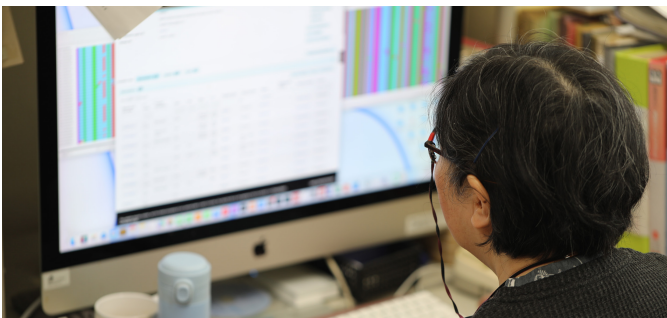
developed to identify genetic changes that cause human diseases. However, GWAS analyses are currently being performed on a variety of human traits. Many GWAS analyses have been performed to identify mutations (SNPs) associated with various traits. It has also been estimated how much each mutation contributes to the trait. However, there is one thing that is still unresolved. That is the functional relationship between SNPs thus identified. It would be nice if software could be developed that could quickly show the relationships among these SNPs, but it will probably be a long time before such software becomes available.



Q. What was the most enjoyable moment and the most challenging moment during your research?

The most enjoyable moment for me is when I feel that I am the only one who knows about this result now. The results of my research can be made known to many people in the world in the form of a published paper. But when it is in the form of a

paper, my data starts to walk on its own as new knowledge. However, I was the first to find the data, there is always a moment when I am the only one in the world who knows about it. When I have that moment, it is the most enjoyable and happy moment. On the other hand, a difficult moment is when I found out that my thought was “wrong” . In my research, I spend a lot of time working with computers. The advantage about computers is that they can tell me what will happen if a population evolve under a particular condition that I thought. The difficult part is producing those conditions. When the results are different from what I thought, is there something wrong with the conditions? Or, there is basically something wrong with what I am thinking. Such are the times when I find it difficult. Yet, when I overcome this difficult time and get the new result, I conversely feel “enjoyment”.



Q. Do you have any message for undergraduate and graduate students aspiring this field of research?

First, look at the data carefully, and if you see something in the data that differs from your intuition, that is a chance to find a “new” fact. It is important to be different from your intuition. When you see data that is different from your intuition, you need to find out what caused it and think about why it happened.

—But you also need to accumulate the background knowledge to believe that your intuition is correct.