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Genetic risk assessment of type 2 diabetes appropriate for individualized health care in Japan

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Our personal genome potentially contains tremendous amount of valuable information for our health but we are currently using only a very small portion of them. Especially for polygenic diseases, it is important to make use of the data on multiple SNPs derived from GWAS, processing these integrated data into genetic risk scores, which are then returned to the individuals. Type 2 diabetes mellitus (T2DM) is one of the representative polygenic diseases with its prevalence currently increasing worldwide. T2DM is preventable as long as the preemptive cares begin at the very early phase or even before onset. Values of genetic tests, though not satiable compared to those of laboratory tests in disease prediction at a certain point in given time, are indicated to have power in evaluating the lifetime onset. Several lines of evidence show the effectiveness of risk assessment by the post-test probability using genotype-based likelihood ratio for the T2DM management in healthy persons. Such risk assessment method has been developed originally based on SNP genotype-disease database of Caucasian population. Taking into consideration the significant differences in risk allele frequency and risk effects between Caucasian and Japanese, we thought it inevitable that we build a Japanese version of SNP genotype-T2DM database. As expected, the SNP combination used for risk calculation for Japanese turned out to be different from the Caucasian set. Pre-evaluation of our risk assessment in a Japanese local cohort (derived from Takahata district) revealed that our risk scoring has better discrimination ability (C-statistics, 0.70; 95% CI, 0.66 to 0.74) than the simple risk allele count scoring (C-statistics, 0.62; 95% CI, 0.57-0.67). Future prospective cohort study should be taken to validate this risk assessment method. Furthermore it will also be important to assess the influence brought by the disclosure of the genetic risk score to the individuals on their behaviors in a sense of prevention of the disease.

Biography

Noriko Sato has received her M.D. and Ph.D. degrees from the University of Tokyo. Her specialty is Molecular Biology and Molecular Epidemiology. She is an Associate Professor, Department of Molecular Epidemiology/Department of Epigenetic Epidemiology, Medical Research Institute, Tokyo Medical and Dental University.

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