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FROM GENOME SCANS TO THE IDENTIFICATION OF FUNCTIONAL GENETIC VARIANTS ASSOCIATED WITH MALARIA RESISTANCE

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The contribution of host genetic factors to resistance or susceptibility to *Plasmodium falciparum* malaria has been widely studied. Nevertheless, a few genome scans have been performed, and few of them led to the discovery of loci significant at the genome level and to the identification of functional variants potentially causal. Here we describe loci genetically linked to malaria phenotype at the genome level and genetic variants located within those loci and associated with malaria phenotype in two independent populations. Furthermore, we provide evidence of a cis-regulatory effect of the genetic variants, suggesting that those variants are causal. We mainly focus on genes and genetic variants located within chromosome 6p21, which has been linked to mild malaria. These include TNF and NCR3, which encode a major actor of inflammation and a receptor of natural killer cells involved the cytotoxicity function, respectively. Also, the results are in line with those supporting the role of TNF in malaria on the one hand and allow us to propose a new biological model to explain the association of a cis-regulatory variant of NCR3 with mild malaria, on the other hand. Also, the genetic variation that alters the activation of natural killer cells may influence human malaria resistance.

Biography

Pascal Rihet has a long lasting experience of research in the field of genetics and genomics of infectious diseases. He has mapped malaria and sepsis predisposing genes by using genetic linkage or association approaches. Furthermore, he has identified many variants associated with the disease; most of those genetic variants are located within noncoding regions. He has provided evidence that several variants have a cis-regulatory effect, suggesting that the regulation of gene expression is critical for the pathogenesis. In this way, he has investigated gene expression profiles in patients or in mouse models, and identified a number of genes whose expression is up- or down-regulated before or at the onset of the disease. He was the Deputy Director of the TAGC laboratory. Currently he is the Director of TAGC laboratory. The research scope of the laboratory is Genetics, Genomics and Bioinformatics. He is a Professor of Genomics and Immunology at AMU.

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