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Characterization of Genetic variation in the Fc gamma receptor locus in the Ecuadorian population

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Human Fcγ receptors (FcγRs) are glycoproteins that bind the Fc region of IgG. These proteins are essentially receptors for immune complexes and are primarily, though not exclusively, expressed on cells of the innate immune system, thereby linking the humoral immune system with cellular effectors. Based on their structure and affinity for monomeric IgG three classes of FcγRs are distinguished, FcγRI (CD64), FcγRII (CD32) and FcγRIII (CD16). Low-affinity FcγRs (CD32 and CD16) are encoded in a single FCGR locus on chromosome 1, a region characterized by both genetic polymorphism and copy number variation (CNV). The combination of gene duplication, multiple common single nucleotide polymorphisms (SNPs) and CNVs is characteristic of regions involved in immune regulation and studies of this CNV and these SNPs in different human populations can provide insight into the evolution of this locus. We studied FCGR genetic variation in a cohort of 69 individuals of Ecuadorian origin, using both a multiplex ligation-dependent probe amplification (MLPA) method (MRC-Holland, Amsterdam, The Netherlands) and direct sequencing. In line with previously published data analyzing other South American populations, we observed a high frequency of individuals carrying 3 copies of FCGR3B (coding for a CD16B receptor) and the great majority of the currently studied donors were of the FCGR3B HNA1a allotype, which is found more frequently in South American populations than in Caucasoids. In contrast, the common *FCGR2A R131H*, *FCGR3A V158F*, *FCGR2B I232T* and *FCGR2C Q57X* polymorphisms were found in distributions similar to those observed in Caucasoids. We also identified 4 intronic SNPs in the FCGR3A gene, two of them not previously described, that might affect splicing. The implications of these data will be discussed.

Biography

Adriana Pérez Portilla studied Biochemistry and Pharmacy at the University of Cuenca (Ecuador). She has a Master's degree in Biotechnology at the Technical University of Madrid (UPM), and she is in a PhD program in Molecular Biosciences at the Autonomous University of Madrid (UAM).

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