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Single Nucleotide Polymorphisms associated with Asthma Phenotypes

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Asthma is an inflammatory airway disease associated with intermittent airflow obstruction and bronchial hyperresponsiveness (BHR) based on eosinophilic airway inflammation. The phenotype of asthma is so much heterogeneous as the disease expression is extremely variable both within and between families. However, patterns of clustering and segregation analyses in asthma families have confirmed a genetic component to asthma. Although much progress has been made toward defining the molecular genetics of asthma over the past decade, the intricacy of the numerous genetic and environmental factors involved has made genetic dissection of this and other complex disease difficult. Genome wide screenings for asthma and related phenotypes have been conducted by collaborative study groups such as Collaborative Study on the Genetics of Asthma (CSGA) in USA, Epidemiologic study for the Genetics and Environment of Asthma (EGEA) in French and in UK and other countries. Genome-wide scanning has revealed that multiple loci on several chromosomes (1p, 2p, 2q, 5p, 6p, 9q, 12q, 13q, 14q, 17q, 19q, 21q) are linked to asthma and related phenotypes. The precise definition of which genes in these regions are responsible for the reported linkages to asthma and the phenotypes may await completion of high- resolution physical maps of the human genome. The number of biologically plausible candidate genes that might be involved in the determination of asthma and associated trait is very large. To date, research into the molecular genetics of asthma has generously focused on candidate genes with clearly defined roles in the allergic process. Several of these candidate genes have been investigated; especially the genes in the loci linked to asthma phenotypes. In this study, screening of candidate gene - SNPs has been performed in order to identify genetic factors contributing to asthma susceptibility and the phenotypes. The analyzing phenotypes were asthma, skin prick test response, total IgE, and bronchial hyperreactivity and peripheral blood eosinophils. In addition, we have studied the functional effects of SNPs of eotaxin genes on the expression of eotaxin m-RNA and protein.