

Genetic susceptibility in thyroid autoimmunity.

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Abstract

The autoimmune thyroid diseases (AITD) include Graves' disease (GD) which manifests in hyperthyroidism and Hashimoto's thyroiditis (HT), manifesting as hypothyroidism. Genetic susceptibility in combination with external factors (e.g. dietary iodine) are believed to initiate the autoimmune response to thyroid antigens in AITD. Indeed, there is solid epidemiological data to support a strong genetic influence on the etiology of AITD including family and twin studies. Recently, there has been significant progress toward the identification of the AITD susceptibility genes. Several loci (genetic regions) that are linked with AITD have been mapped and in some of these loci putative AITD susceptibility genes have been identified. Some of these loci predispose to a single phenotype (GD or HT), while other loci are common to both diseases, indicating that there is a shared genetic susceptibility to GD and HT. The putative GD and HT susceptibility genes include both immune modifying genes (e.g. HLA, CTLA-4) and thyroid specific genes (e.g. TSHR, Tg) and it is likely that the final disease phenotype is a result of an interaction between these loci, as well as environmental influences.