The Genetics of Multiple Sclerosis: 4 Key Points

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• **Relatives of patients with MS are at increased risk for this disease.**

  Population based studies have firmly established that relatives of patients with MS are at increased risk for the disease. For example, children of MS patients have a lifetime risk of 3% to 4% for MS compared with 0.1% for unrelated individuals. About 20% of MS patients report having an affected relative. Non-biologically related individuals living with an MS patient (spouses, step-siblings, adopted relatives) are not at increased risk for MS. This demonstrates that genetic factors are almost entirely responsible for the increased risk of MS seen in relatives.

• **The HLA-DRB1 gene is a strong biomarker for MS.**

  In the early 1970s, MS was shown to be associated with the human leukocyte antigen (HLA) region on chromosome 6—in particular the HLA-DRB1 gene. We know now that this locus exerts the single strongest genetic effect in MS (a single copy of the HLA-DRB1*15 risk allele increases risk by 3-fold).

• **Many other genes, all involving the immune system, are also involved.**

  More recently, through developments in technology, scanning the genome with hundreds of thousands of markers in thousands of MS patients and controls has shown that approximately 60 other genes confer a more modest effect on MS risk (10% to 20% risk increase). These genes all have an immune function, providing overwhelming evidence that MS is an immune-mediated disease.

• **More information is on the way.**

  Despite recent success in identifying genes, we still cannot fully explain the genetic basis of MS. This suggests that more genetic factors remain to be found. While this will likely not help us predict those in whom MS will develop, it may aid in further understanding the disease’s pathogenesis and designing therapeutics.

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