

Lupus Genetic Markers

Lupus is an autoimmune disease that causes chronic inflammation and destruction of multiple body organs. The antibodies get activated against its body organs and start destroying them. It has clinical heterogeneity which means that it has a variety of disease manifestations. It may involve one body organ in one person and multiple organ systems in the other. It shares its signs and symptoms with other autoimmune diseases like rheumatoid arthritis. Same is true regarding its severity.

Lupus and Genetics

Lupus is a multifactorial disorder with an elusive etiology; it has a strong association with multiple factors, genetic and non-genetic factors. Clinical studies have established that ultraviolet radiation, specific drugs (isoniazid, hydralazine, procainamide) and infections (Epstein-bar virus) can also lead to SLE (systemic lupus erythematosus).

Currently, there is an explosion of information regarding the possible role of genetic markers in this disease. Hundreds of studies are being done in many parts of the world to establish its genetic predisposition. This new evidence has profound effects on understanding the etiology and planning an effective remedy.

What makes Lupus a genetic disease?

Lupus has been found to exert some typical etiological features similar to the known hereditary disease. It has a clear predilection for affecting women in their young ages. It

is so common in females that it is being labeled as a women disease. About 9 out of 10 lupus patients are young women. Apart from having a strong link with a specific age and gender, its runs in families too. About 10% of lupus patients have some relevant bearing the same disease. It was found to have a remarkable 40% concordance in monozygotic twins and 4% concordance in dizygotic twins. In addition, it is prevalent in certain races like Hispanic, African-Americans, and Asian-Americans. All of these factors make it clear that lupus has a strong genetic predisposition; faulty genes being the real culprit.

Study approaches for complex genetic causes:

In order to understand the genetic side of lupus, you must also have an idea about methods that are being applied to find the truth. Currently, two major approaches are being used:

- Linkage studies
- Association studies

Linkage studies involve gathering of families in which 2 or more than two persons have lupus. The genome of such families is scanned using microsatellite markers and their genetic makeup is studied. In association studies, one single gene is selected and hypotheses are formed against it. Most common practice is to study a single code using case-control cohort method. In order to refine results, trio design is being formed which includes testing of both patient and his/her parents. With the latest advances in technology, these methods are also getting quick, easy and improved.

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lupus genetic markers

Important Genetic markers in Lupus:

Before naming the exact genes related to lupus, it is important to know that lupus is a genetically complex trait. It means that several genes, coding regions or loci are involved in causing susceptibility in this disease. There are no single genes but a group of many loci implicated in lupus. There is also the possibility of their mutual interaction and interaction with hormonal or non-genetic factors. The important lupus genetic markers can be listed as:

▪ HLA region genes:

HLA (human leukocyte antigen) genes are a topmost group of genetic markers that are strongly associated with lupus. These genes maintain the body immune system. The deletion of C4A allele has been found closely related to increased risk of lupus for many years. However, HLA region contains hundreds of genes and it has not been possible to pinpoint a single culprit gene yet. It is quite clear that HLA is the strongest genetic association of lupus among all other genes. HLA class 1&2 have been shortlisted and further studied for finding exact code.

▪ FCγR 2A and 3A:

The Fc receptors for immunoglobulin G protein are located at membranes of certain immune cells. These are responsible for the clearance of immune complexes and have been found to be linked with lupus. Several meta-analyses have shown their presence in patients of kidney lupus.

▪ STAT4:

STAT4 is a newly discovered agent and polymorphism in these genes is related to lupus and rheumatoid arthritis both.

▪ IRF-5:

Interferon regulatory factor 5 is a part of innate immunity and is said to increase the risk of lupus according to many cohort studies. Many groups of successive cohort studies established its possible role in causing lupus and other autoimmune diseases.

Conclusion

Unfortunately, no genetic marker or group of genes has been finally confirmed and are being further investigated. Much has to be done yet to solve the genetic mysteries of lupus. After years of studies, we have an idea about the possible culprit genes and their mechanisms. With advancing technology and refined tools, we should be hoping for some breakthrough in the future.