

The Role of Genetics in Diabetic Retinopathy

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Diabetic retinopathy is one of the most severe complications that can occur due to the abnormal changes in the microvascular in diabetic patients. Diabetes is known to affect the blood flow to the vital organs including the heart and brain. It can also affect the tiny capillaries supplying blood to the eyes, especially the retina, causing Diabetic retinopathy.

If left unchecked, it can affect the eyesight of the person to the extent of causing complete or partial blindness. With the worldwide prevalence of diabetes being projected to rise substantially with every passing year, diabetic retinopathy is expected to pose a major health concern. [\[1\]](#)

Hence, efforts are being made to unravel the role of genetics in the development of this condition. The findings of research studies conducted to evaluate the role of genetics are expected to help patients take appropriate measures to inhibit the progress of this disease.

Epidemiological studies have indicated that the risk of diabetic retinopathy is higher in patients who suffer from uncontrolled diabetes. At the same time, intensive clinical measures to bring about glycaemic control have been shown to delay the development of this complication. [\[2\]](#)

However, it is noteworthy that some diabetic patients may develop diabetic retinopathy in spite of having good glycaemic control. Also, at times, patients with poor control over blood sugar levels have been spared of this complication.

These contradictory observations have led to the belief that the genetic susceptibility could be responsible for making a person prone to develop diabetic retinopathy. This means that the risk of developing diabetic retinopathy may still persist even in patients who have good control over their blood sugar levels simply due to their genetic make-up.

An effort was made to evaluate the functional role of genes in the pathogenesis of diabetic retinopathy using scientific research methods such as genetic analysis and mapping the genes to biologic processes or pathways. It is found that most of the genes involved in the

development of this condition contribute to its development by altering biological processes as given below:

- Insulin signaling pathways can affect the receptors in the retina, which respond to the blood sugar-regulating hormone called insulin. This can result in inflammation or damage to the retina and trigger the development of retinopathy. [3]
- Genes that control the process of formation of new blood vessels called angiogenesis can contribute to the risk of developing retinopathy. It can cause abnormal changes in the process of regulation of the size of blood vessels due to which the blood supply to the retina is reduced.
- The genes involved in regulating the functions of the immune system can cause an increased production of pro-inflammatory substances like interleukin-6, as well as tumor necrosis factor and transforming growth factor-B. These chemicals can cause damage to the retina resulting in an increased risk of diabetic retinopathy. [4]
- Other than these, the genes that play a role in regulating lipid metabolic processes, differentiation of neural cells and neurotrophin signaling can also contribute to a high risk of retinopathy.

Further analysis of these biological processes will help reveal the functions these genes affect and trigger the development of diabetic retinopathy. The identification of specific genetic factors as well as their contribution to the pathogenesis of diabetic retinopathy is expected to allow diabetologists to recommend a customized treatment plan to patients based on his or her specific genetical assessments. This would also help to prevent complications related to diabetic retinopathy such as visual disability and blindness.

References:

1. <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4398904/>
2. <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1705856/>
3. <https://www.ncbi.nlm.nih.gov/pubmed/16567541>
4. <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5870958/>