



IZMIR BIOMEDICINE AND GENOME CENTER RAREBOOST PROJECT

LET'S LEARN ABOUT MACULAR CORNEAL DYSTROPHY (MCD

WHAT IS CORNEA?

2

The cornea is the transparent tissue at the front of our eye, responsible for admitting light and plays an important role in clear vision.





CORNEAL DYSTROPHY

Corneal dystrophies are inherited, progressive diseases that affect both eyes. These conditions compromise the cornea's transparency, resulting in visual disturbances. Macular Corneal Dystrophy (MCD) is one of the most prevalent forms of this disorder.

MCD DISEASE GENETICS -

MCD is usually inherited through recessive genes. In order for an individual to develop the disease, they must receive the mutated gene from both of their parents. In this case, both parents become "carriers." Carriers usually do not show symptoms but can pass them on to their children.





— MCD AND CONSANGUINEOUS MARRIAGE -

Although the rate of this genetic disease being carried in society is low, the risk of these genes coming together increases, especially in consanguineous marriages. As a result of consanguineous marriages, the probability of having children with serious vision impairment.

WHY AND HOW DOES MKD OCCUR?

In MCD disease, the cornea's transparency decreases due to the accumulation of keratan-sulfate in the cornea, and patients usually begin to experience vision loss in their 20s or 30s.



This project has received funding from the European Union's Horizon 2020 research and innovation program (grant agreement no. 952346).



IZMIR BİOMEDICINE AND GENOME CENTER RAREBOOST PROJECT

LET'S LEARN ABOUT MACULAR CORNEAL DYSTROPHY (MCD)

IS THERE A TREATMENT?

Currently, corneal transplantation is the most effective treatment method known. However, there is a possibility of recurrence of the disease even after this transplant.





- HOW IS IT DIAGNOSED? -

MKD can result from different changes (mutations) in the same gene. A simple blood test can confirm the diagnosis.

WHAT CAN I DO?

- HOW CAN THE RISK BE REDUCED?

While researchers explore non-surgical treatments for MCD, preventing consanguineous marriages is crucial to reducing the risk of this and similar diseases.

• You can have genetic tests before marriage.

g

• If you are a surrogate parent, you can seek genetic counseling from your doctor during the process of having a child.



- You can pay attention to routine eye examinations starting from childhood.
- You can follow the Ministry of Health screening programs.

CONSULT YOUR PHYSICIAN



If you experience any changes in your eye health, please see an eye doctor right away.

10

We appreciate your trust in our treatment and your belief in the power of science

- Izmir Biomedicine and Genome Center (IBG) is a leading research center in Türkiye that aims to develop advanced technologies and products for diagnosis, diagnostics, and transportation in life sciences.
- RareBoost is a project by IBC to enhance capacity in rare disease research and innovation.

