



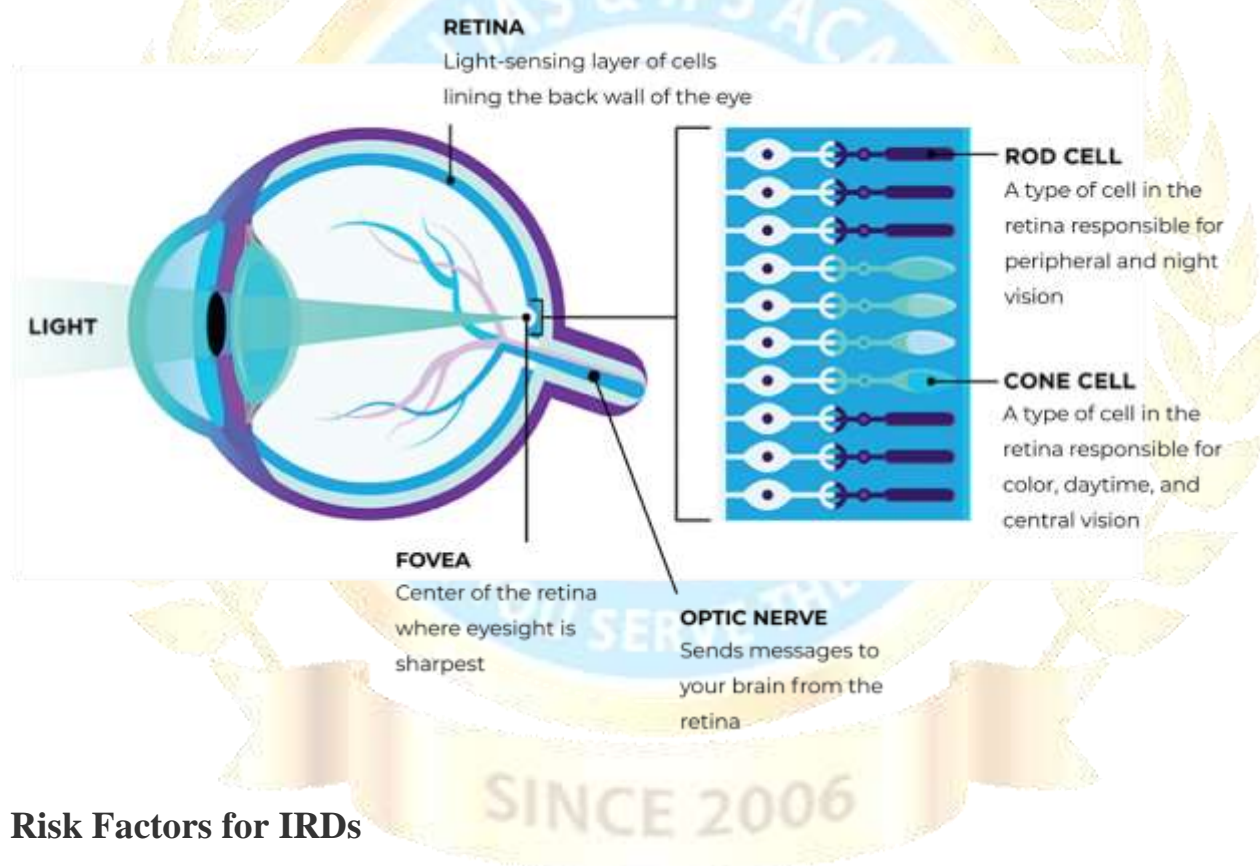
INHERITED RETINAL DISEASES - DISEASES

NEWS: Over 2.2 billion people worldwide suffer from some form of vision impairment, with inherited retinal diseases (IRDs) being a leading cause.

WHAT'S IN THE NEWS?

What are Inherited Retinal Diseases (IRDs)?

- Inherited Retinal Diseases (IRDs) are a group of **genetic conditions** that cause **progressive vision loss or blindness** due to **damage to the retina**, the **light-sensitive tissue** at the back of the eye.
- These diseases are passed down from parents to their children through inherited gene mutations.
- The retina's primary role is to capture light and send visual signals to the brain.



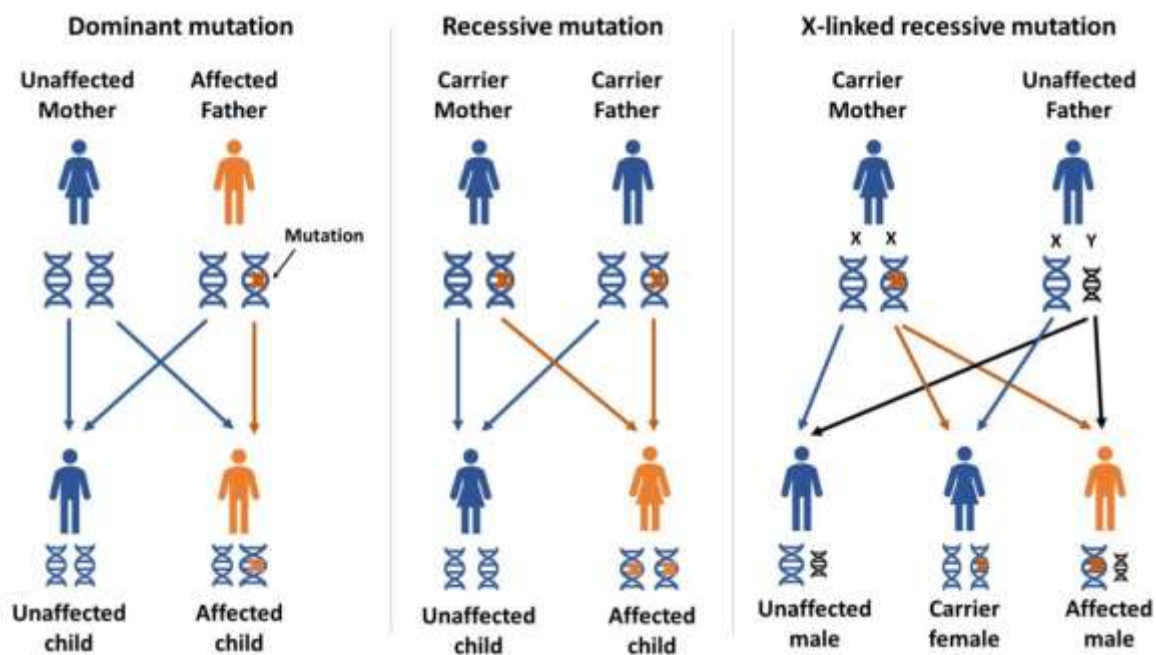
Risk Factors for IRDs

IRDs follow different **inheritance patterns**:

- **Autosomal Dominant:** The defective gene is located on an autosome (non-sex chromosome).
 - A person inherits one faulty dominant gene from a parent and one normal gene from the other.
 - The faulty dominant gene causes the disorder.



- **Autosomal Recessive:** Both copies of the gene (one from each parent) must be defective.
 - Parents are usually asymptomatic carriers.
 - 25% chance of inheriting the disease if both parents are carriers.
- **X-linked Disorders:** The defective gene is located on the X chromosome.
 - Males (XY) are more severely affected as they have only one X.
 - Females (XX) may be carriers or show milder symptoms.



- **Mitochondrial Inheritance:** Mutations occur in mitochondrial DNA (mtDNA), inherited exclusively from the mother (**sperm do not contribute mitochondria**).
 - Can affect multiple organs, including the eyes.
- **Common IRDs include:**
 - **Retinitis Pigmentosa:** A group of diseases that cause the breakdown of the retina's cells, leading to gradual vision loss, often starting with night blindness.
 - **Stargardt Disease:** A form of macular degeneration that usually affects children and causes central vision loss.
 - **Leber Congenital Amaurosis:** A severe form of vision loss that typically manifests at birth or in infancy.
 - **Cone-Rod Dystrophy:** A condition that affects both the central and peripheral vision, often leading to color blindness or light sensitivity.



- **Treatment:** Currently, there is no cure for most IRDs, but treatments like **gene therapy and RNA-based therapies** are showing promise in slowing or even halting disease progression.
- Despite global advances, there is limited awareness of gene therapy options like RPE65 gene therapy in India. While gene therapy is a breakthrough, it is not effective for all genetic conditions, paving the way for RNA-based therapies.
- **RNA-based Therapies:**
 - RNA-based therapies, including **antisense oligonucleotides (ASOs)**, are emerging as safer and more precise alternatives to gene therapy.
 - These therapies make temporary changes at the RNA level without altering DNA, reducing the risk of long-term effects.
 - ASO therapy is already successfully used for diseases like spinal muscular atrophy and Duchenne muscular dystrophy, and it is being explored for retinal conditions such as **Stargardt disease and retinitis pigmentosa**.

According to the World Health Organisation, more than 2.2 billion people worldwide experience **some forms of vision impairment**.

- An estimated 5.5 million people suffer from IRDs around the world, with a prevalence rate of one in 3,450.
- Studies have revealed significantly higher prevalence of such cases in India
 - one in 372 individuals in rural South India,
 - one in 930 in urban South India,
 - and one in 750 in rural Central India affected by these conditions.

India's Role in Precision Medicine

What is Precision Medicine?

1. Tailored Treatments:

Precision medicine is an innovative approach to healthcare that customizes medical treatments based on an individual's unique genetic makeup, lifestyle, and environmental factors. Unlike traditional medicine, which often uses a generalized approach, precision medicine focuses on delivering personalized care.

2. Personalized Care:

The goal of precision medicine is to move away from a "one-size-fits-all" model and instead provide treatments that are specifically designed for each patient. This approach improves treatment efficacy, reduces side effects, and enhances overall patient outcomes.

Need for Genetic Research in India



1. Genetic Links to Inherited Retinal Diseases (IRDs):

Over 300 genes have been identified as being associated with Inherited Retinal Diseases (IRDs), which are a group of genetic disorders causing vision loss. However, India lacks large-scale studies to understand the genetic mutations specific to its population.

2. Lack of Large-Scale Studies:

No major study involving more than 500 patients has been conducted in India to map the genetic variations in IRD patients. This gap in research limits the understanding of the genetic landscape of IRDs in the Indian population.

3. Importance of Identifying Mutations:

Identifying common genetic mutations among Indian patients is crucial for developing effective and targeted treatments. Without this knowledge, it is challenging to create therapies that are tailored to the genetic profile of the Indian population.

Challenges in India

1. Diverse Genetic Variations:

India's population is highly diverse, with significant genetic variations across different communities. This diversity makes genetic research more complex, as findings from one community may not apply to another.

2. Barriers to Precision Medicine:

Several challenges hinder the progress of precision medicine in India:

- **Low Awareness:** Many doctors and patients are unaware of the benefits and availability of genetic testing and precision medicine.
- **Limited Access to Genetic Counseling:** Genetic counseling services, which are essential for interpreting genetic test results and guiding treatment decisions, are not widely available.
- **Insufficient Research Funding:** There is a lack of adequate funding for genetic research, which limits the scope and scale of studies.
- **Diagnostic Infrastructure Gaps:** Diagnostic facilities, particularly in rural areas, are inadequate, making it difficult for patients to access genetic testing and related services.

Conclusion

Precision medicine holds immense potential for transforming healthcare in India by providing personalized treatments based on genetic, lifestyle, and environmental factors. However, realizing this potential requires addressing significant challenges, including the lack of large-scale genetic studies, low awareness, limited access to genetic counseling, insufficient funding, and inadequate diagnostic infrastructure. By overcoming these barriers, India can play a pivotal role in advancing precision medicine and improving healthcare outcomes for its diverse population.



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Source: <https://www.thehindu.com/sci-tech/science/inherited-retinal-diseases-rna-therapeutics-promise-india/article69143245.ece>



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