

UNIT-II

NUCLEAR RADIATION AND ITS EFFECTS ON THE BODY

2.2 CATARACTS GENETIC EFFECTS

Cataracts, which cause clouding of the eye's natural lens, can have genetic causes alongside environmental factors like aging, UV exposure, smoking, and certain medications. Here's a breakdown of the genetic influences:

1. Congenital Cataracts

These are present at birth or develop in early childhood and are often linked to genetic mutations. They can be inherited in different ways:

- **Autosomal Dominant:** A mutation in just one copy of a gene is enough to cause cataracts. This is the most common inheritance pattern for congenital cataracts.
- **Autosomal Recessive:** Both copies of a gene need to be mutated for cataracts to develop. This is less common.
- **X-linked:** Rare cases where mutations are carried on the X chromosome, affecting mostly males.

2. Genes Associated with Cataracts

Several genes have been implicated in cataract development:

- **CRYAA, CRYAB, CRYBB2:** These genes code for crystallins, the structural proteins in the lens. Mutations can lead to protein clumping, causing cloudiness.
- **MIP (Major Intrinsic Protein):** Mutations affect water balance in the lens, contributing to cataract formation.
- **GJA8:** Codes for connexin proteins important in cell communication within the lens. Mutations can disrupt this process, leading to cataracts.

3. Syndromic Cataracts

Some genetic disorders include cataracts as part of a broader syndrome:

- **Down Syndrome:** Increased risk of cataracts in early life.
- **Galactosemia:** A metabolic disorder that can cause cataracts due to accumulation of galactose.
- **Marfan Syndrome:** Connective tissue disorder associated with lens dislocation and cataracts.
- **Lowe Syndrome:** Affects the eyes, brain, and kidneys, with cataracts being an early sign.

4. Age-related Cataracts and Genetics

**While aging is the primary factor in most cataracts**, genetic predisposition can influence when and how they develop. Variants in genes related to oxidative stress, metabolism, and lens structure can increase susceptibility.

### 5. Genetic Testing and Counseling

For families with a history of congenital or early-onset cataracts, genetic testing can identify mutations and help guide treatment decisions. Genetic counseling may also be beneficial for understanding inheritance patterns and risks for future generations.

