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**MATRIC NUMBER: 18/MHS02/192**

**COURSECODE: PHS212**

**COURSE TITLE: PHYSIOLOGY**

**QUESTION**

Write short notes on any two eye defects

**ASTIGMATISM:** Astigmatism is a common vision problem caused by an error in the shape of the cornea. With astigmatism, the lens of the eye or the cornea, which is the front surface of the eye, has an irregular curve. This can change the way light passes, or refracts, to your Retina. This causes blurry, fuzzy, or distorted vision. Farsightedness and Nearsightedness are two other types of problems with the way light passes to your retina. Farsightedness is called hyperopia. Nearsightedness is called myopia.

The two main types of astigmatism are corneal and lenticular. A corneal astigmatism happens when your cornea is misshapen. A lenticular astigmatism happens when your lens is misshapen.

Causes: It's not known what causes astigmatism, but genetics is a big factor. It's often present at birth, but it may develop later in life. It may also occur as a result of an injury to the eye or after eye surgery. Astigmatism often occurs with nearsightedness or farsightedness.

## **Genetics**

Genetics, based on twin studies, appear to play only a small role in astigmatism as of 2007.

Genome-wide association study (GWAS) have been used to investigate the genetic foundation of astigmatism. Although no conclusive result has been shown, various candidates have been identified. In a study conducted in 2011 on various Asian populations, variants in the PDGFRA gene on chromosome 4q12 were identified to be associated with corneal astigmatism. A follow-up study in 2013 on the European population, however, found no variant significantly associated with corneal astigmatism at the genome-wide level (single-nucleotide polymorphism rs7677751 at PDGFRA). Facing the inconsistency, a study by Shah and colleagues in 2018 included both populations with Asian and Northern European ancestry. They successfully replicated the previously identified genome-wide significant locus for corneal astigmatism near the PDGFRA gene, with a further success of identifying three novel candidate genes: CLDN7, ACP2, and TNFAIP8L3. Other GWAS studies also provided inconclusive results: Lopes and colleagues identified a susceptibility locus with lead single nucleotide polymorphism rs3771395 on chromosome 2p13.3 in the VAX2 gene (VAX2 plays an important role in the development of the dorsoventral axis of the eye); Li and associates, however, found no consistent or strong genetic signals for refractive astigmatism while suggesting a possibility of widespread genetic co-susceptibility for spherical and astigmatic refractive errors. They also

found that the TOX gene region previously identified for spherical equivalent refractive error was the second most strongly associated region. Another recent follow-up study again had identified four novel loci for corneal astigmatism, with two also being novel loci for astigmatism: ZC3H11B (associated with axial length), NPLOC4 (associated with myopia), LINC00340 (associated with spherical equivalent refractive error) and HERC2 (associated with eye color).

## **PATHOPHYSIOLOGY**

### Axis of the principal meridian

- Regular astigmatism – principal meridians are perpendicular. (The steepest and flattest meridians of the eye are called principal meridians.)
  - With-the-rule astigmatism – the vertical meridian is steepest (a rugby ball or American football lying on its side).
  - Against-the-rule astigmatism – the horizontal meridian is steepest (a rugby ball or American football standing on its end).
  - Oblique astigmatism – the steepest curve lies in between 120 and 150 degrees and 30 and 60 degrees.
- Irregular astigmatism – principal meridians are not perpendicular.

In with-the-rule astigmatism, the eye has too much "plus" cylinder in the horizontal axis relative to the vertical axis (i.e., the eye is too "steep" along the vertical meridian relative to the horizontal meridian). Vertical beams of light focus in front (anterior) to horizontal beams of light, in the eye. This problem may be corrected using spectacles which have a "minus" cylinder placed on this horizontal axis. The effect of this will be that when a vertical beam of light in the distance travels towards the eye, the "minus" cylinder (which is placed with its axis lying horizontally – in line with the patient's excessively steep horizontal axis/vertical meridian) will cause this vertical beam of light to slightly "diverge", or "spread out vertically", before it reaches the eye. This compensates for the fact that the patient's eye converges light more powerfully in the vertical meridian than the horizontal meridian. Hopefully, after this, the eye will focus all light on the same location at the retina, and the patient's vision will be less blurred.

In against-the-rule astigmatism, a plus cylinder is added in the horizontal axis (or a minus cylinder in the vertical axis).

Axis is always recorded as an angle in degrees, between 0 and 180 degrees in a counter-clockwise direction. Both 0 and 180 degrees lie on a horizontal line at the level of the center of the pupil, and as seen by an observer, 0 lies on the right of both the eyes.

Irregular astigmatism, which is often associated with prior ocular surgery or trauma, is also a common naturally occurring condition. The two steep hemimeridians of the cornea, 180° apart in regular astigmatism, may be separated by less than 180° in irregular astigmatism (called *nonorthogonal* irregular astigmatism); and/or the two steep hemimeridians may be asymmetrically steep—that is, one may be significantly steeper than the other (called *asymmetric* irregular astigmatism). Irregular astigmatism is quantified by a vector calculation called topographic disparity.

### **Focus of the principal meridian**

- Simple astigmatism
  - Simple hyperopic astigmatism – first focal line is on the retina, while the second is located behind the retina.
  - Simple myopic astigmatism – first focal line is in front of the retina, while the second is on the retina.
- Compound astigmatism
  - Compound hyperopic astigmatism – both focal lines are located behind the retina.
  - Compound myopic astigmatism – both focal lines are located in front of the retina.
- Mixed astigmatism – focal lines are on both sides of the retina (straddling the retina)

### **Throughout the eye**

Astigmatism, whether it is regular or irregular, is caused by some combination of external (corneal surface) and internal (posterior corneal surface, human lens, fluids, retina, and eye-brain interface) optical properties. In some people, the external optics may have the greater influence, and in other people, the internal optics may predominate. Importantly, the axes and magnitudes of external and internal astigmatism do not necessarily coincide, but it is the combination of the two that by definition determines the overall optics of the eye. The overall optics of the eye are

typically expressed by a person's refraction; the contribution of the external (anterior corneal) astigmatism is measured through the use of techniques such as keratometry and corneal topography. One method analyzes vectors for planning refractive surgery such that the surgery is apportioned optimally between both the refractive and topographic components.

There are three primary types of astigmatism: myopic astigmatism, hyperopic astigmatism, and mixed astigmatism. Cases can be classified further, such as regular or irregular and lenticular or corneal.

### **Treatment**

Astigmatism may be corrected with eyeglasses, contact lenses, or refractive surgery. Glasses are the simplest and safest, although contact lenses can provide a wider field of vision. Refractive surgery can eliminate the need to wear corrective lenses altogether by permanently changing the shape of the eye but, like all elective surgery, comes with both greater risk and expense than the non-invasive options. Various considerations involving eye health, refractive status, and lifestyle determine whether one option may be better than another. In those with keratoconus, certain contact lenses often enable patients to achieve better visual acuity than eyeglasses. Once only available in a rigid, gas-permeable form, toric lenses are now also available as soft lenses.

In older people, astigmatism can also be corrected during cataract surgery. This can either be done by inserting a toric intraocular lens or by performing special incisions (limbal relaxing incisions). Toric intraocular lenses probably provide a better outcome with respect to astigmatism in these cases than limbal relaxing incisions.

**MYOPIA: Near-sightedness**, also known as **short-sightedness** and **myopia**, is an eye disorder where light focuses in front of, instead of on, the retina. This causes distant objects to be blurry while close objects appear normal. Other symptoms may include headaches and eye strain. Severe near-sightedness is associated with an increased risk of retinal detachment, cataracts, and glaucoma.

The underlying cause is believed to be a combination of genetic and environmental factors. Risk factors include doing work that involves focusing on close objects, greater time spent indoors, and a family history of the condition. It is also associated with a high socioeconomic class. The

underlying mechanism involves the length of the eyeball growing too long or less commonly the lens being too strong. It is a type of refractive error. Diagnosis is by eye examination.

Tentative evidence indicates that the risk of near-sightedness can be decreased by having young children spend more time outside. This may be related to natural light exposure. Near-sightedness can be corrected with eyeglasses, contact lenses, or surgery. Eyeglasses are the easiest and safest method of correction. Contact lenses can provide a wider field of vision, but are associated with a risk of infection. Refractive surgery permanently changes the shape of the cornea.

Near-sightedness is the most common eye problem and is estimated to affect 1.5 billion people (22% of the population). Rates vary significantly in different areas of the world. Rates among adults are between 15% to 49%. Rates are similar in females and males. Among children, it affects 1% of rural Nepalese, 4% of South Africans, 12% of Americans, and 37% in some large Chinese cities. Rates have increased since the 1950s. Uncorrected near-sightedness is one of the most common causes of vision impairment globally along with cataracts, macular degeneration, and vitamin A deficiency.

### **Signs and symptoms**

A myopic individual can see clearly out to a certain distance (called far point), but everything further becomes blurry. If the extent of the myopia is great enough, even standard reading distances can be affected. Upon routine examination of the eyes, the vast majority of myopic eyes appear structurally identical to no myopic eyes.

Onset is often in school children, with worsening between the ages of 8 and 15.

### **Causes**

The underlying cause is believed to be a combination of genetic and environmental factors. Risk factors include doing work that involves focusing on close objects, greater time spent indoors, and a family history of the condition. It is also associated with a high socioeconomic class.

A 2012 review could not find strong evidence for any single cause, although many theories have been discredited. Identical twins are more likely to be affected than non-identical twins which

indicate at least some genetic factors are involved. Myopia has been increasing rapidly throughout the developed world, suggesting environmental factors are involved.

## **Genetics**

A risk for myopia may be inherited from one's parents. Genetic linkage studies have identified 18 possible loci on 15 different chromosomes that are associated with myopia, but none of these loci is part of the candidate genes that cause myopia. Instead of a simple one-gene locus controlling the onset of myopia, a complex interaction of many mutated proteins acting in concert may be the cause. Instead of myopia being caused by a defect in a structural protein, defects in the control of these structural proteins might be the actual cause of myopia. A collaboration of all myopia studies worldwide identified 16 new loci for refractive error in individuals of European ancestry, of which 8 were shared with Asians. The new loci include candidate genes with functions in neurotransmission, ion transport, retinoic acid metabolism, and extracellular matrix remodeling and eye development. The carriers of the high-risk genes have a tenfold increased risk of myopia.

Human population studies suggest that contribution of genetic factors accounts for 60–90% of variance in refraction. However, the currently identified variants account for only a small fraction of myopia cases, suggesting the existence of a large number of yet unidentified low-frequency or small-effect variants, which underlie the majority of myopia cases.

## **Treatment**

The National Institutes of Health says there is no known way of preventing myopia, and the use of glasses or contact lenses does not affect its progression. There is no universally accepted method of preventing myopia and proposed methods need additional study to determine their effectiveness. Optical correction using glasses or contact lenses is the most common treatment; other approaches include orthokeratology, and refractive surgery. Medications (mostly atropine) and vision therapy can be effective in addressing the various forms of pseudomyopia.