BHARATHIDASAN UNIVERSITY

Tiruchirappalli- 620024,
Tamil Nadu, India

Programme: M.Sc., Biomedical science

Course Title : Molecular medicine

Course Code : BM48C16M

Unit-I

TOPIC: RED-GREEN COLOR BLINDNESS & TAY SACH'S DISEASE

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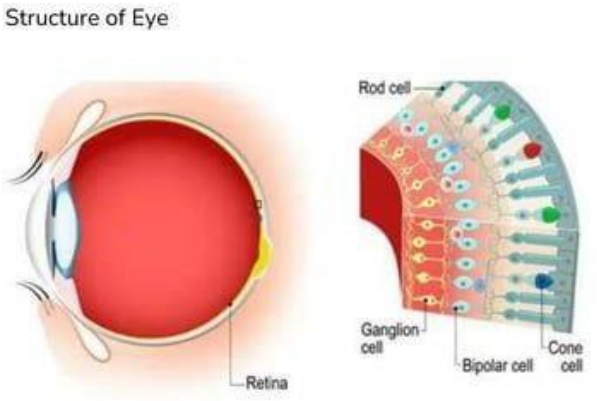
RED-GREEN COLOR BLINDNESS

- Color blindness occurs when you are unable to see colors in normal way.
- Color blindness often happens when someone cannot distinguish between certain colors. This usually happens between greens and reds.
- It is also known as X-linked inheritance.
- In the retina there are two types of cells that direct light. They are called rods and cones.
- Genes responsible for color blindness are present on X- chromosome. It is also called as Red-Green color blindness.

- Cone cells- Responsible for color vision
- Opsins-Photopsins in cone.
- X-linked inheritance – X-linked recessive. Both alleles for a character should be mut



CLASSIFICATION

- There are three types of cones:

    1.S-type-Blue sensitive opsin present in chromosome 7.

    2.M-type-Green sensitive opsin present in X-chromosome.

    3.C-type-Red sensitive opsin present in X-chromosome.

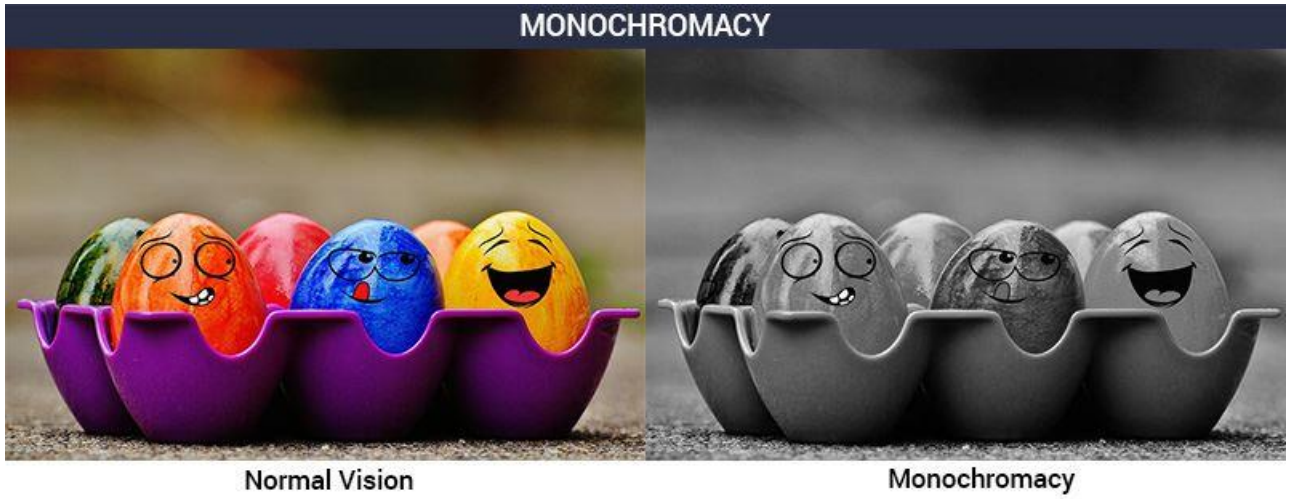
>Individuals who have all three types cones working at full capacity are called Trichromats. Likewise normal vision can be referred as Trichromacy.

- Anomalous trichromacy-one color is seen weakly.
    1.Protanomaly(L-cone defect)-Red weak
    2.Deuteranomaly(M-cone defect)-Green weak(common)
    3.Tritanomly(S-cone defect)-Blue weak(Rare).

>Dichromacy- Only two of three primary colors are seen.One is totally dysfunctional or

1.Protanopia(no L-cone/no red cone)
2.Deuteraponia(no M-cone/no green cone)
3.Tritanopia(no S-cone/no blue cone)

- Monochromacy(achromatopsia)-1 in every 33,000 people. People with monochromacy see no color at all. For these individuals, the world exists in black and white.



GENES RESPONSIBLE

- Mutations in the ***OPN1LW,OPN1MW*** and ***OPN1SW*** genes cause the forms of color deficiency.
- The OPN1LW,OPN1MW,and OPN1SW genes provide instructions for making opsins pigments in the cone.
- **OPN1LW-**Long wavelength, located on X-chromosome at position **Xq28.**
- **OPN1MW-**Medium wavelength ,position **Xq28**
- **OPN1SW-**Short wavelength chromosome 7 position**-7q32.1**

ACQUIRED COLOR BLINDNESS

- Age
- Alcohol consumption
- Brain trauma
- Chronic illness such as Alzheimer's disease, leukemia
- Macular degeneration

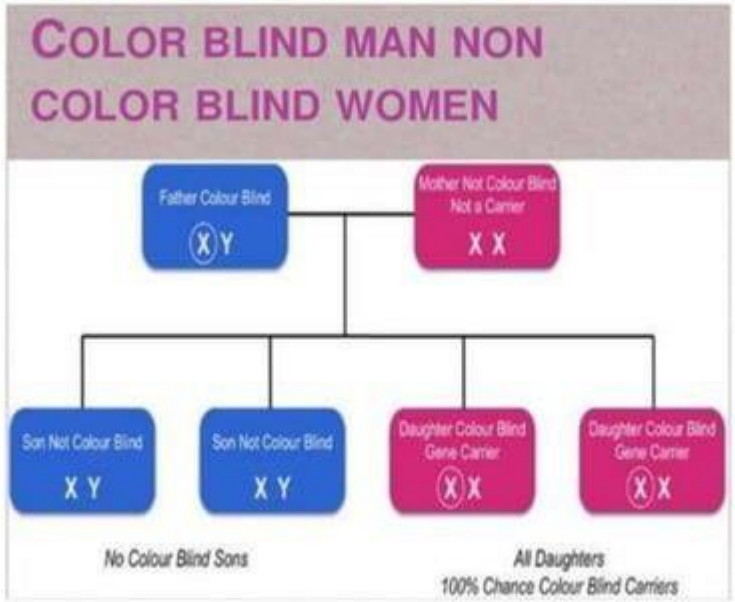
SYMPTOMS

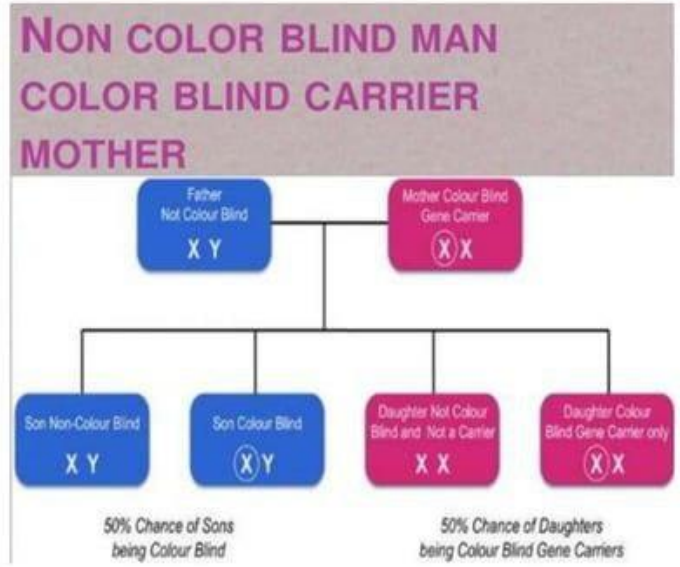
- The symptoms of color blindness can range from mild to severe
- Many people have such mild symptoms that are unaware that they have a color deficiency
- This symptoms include

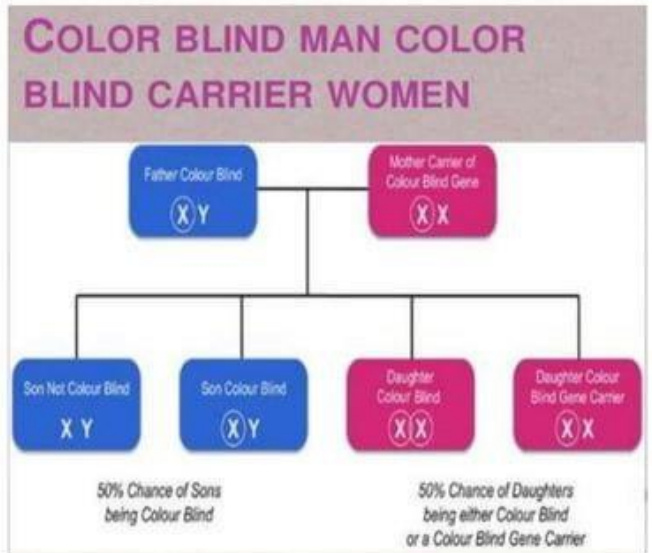
>trouble seeing colors and the brightness of colors in usual way

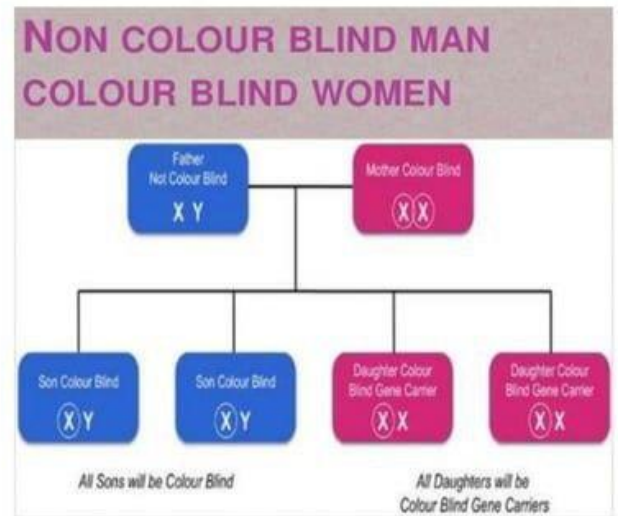
>Inability to tell the difference between shades of the same or similar colors.

INHERITANCE PATTERN



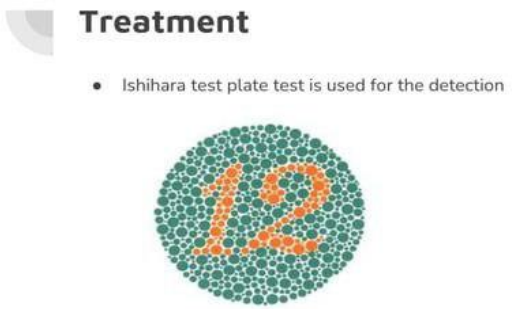



COLOR BLIND MAN COLOR BLIND CARRIER WOMEN
Father Colour Blind
X Y
Mother Carrier of Colour Blind Gene
X X
Son Not Colour Blind
X Y
Son Colour Blind
X Y
Daughter Colour Blind
X X
Daughter Colour Blind Gene Carrier
X X
50% Chance of Sons being Colour Blind
50% Chance of Daughters being either Colour Blind or a Colour Blind Gene Carrier


NON COLOUR BLIND MAN COLOUR BLIND WOMEN
Father Not Colour Blind
X Y
Mother Colour Blind
X X
Son Colour Blind
X Y
Son Colour Blind
X Y
Daughter Colour Blind Gene Carrier
X X
Daughter Colour Blind Gene Carrier
X X
All Sons will be Colour Blind
All Daughters will be Colour Blind Gene Carriers

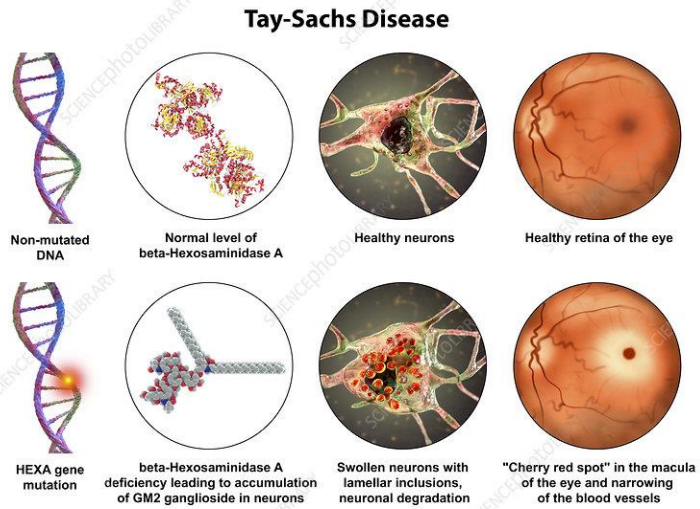
TREATMENT

- Ishihara test plate test is used for the detection.
- Color filter or contact lens can be used to enhance the brightness of some colors
- For acquired color blindness once the cause has been established and treated the vision may be restored back to normal.



TAY-SACHS DISEASE

- Rare disorder passed from parents to child.
- Absence of an enzyme that helps breakdown of fatty substances called Gangliosides.
- Build up to toxic levels in the child's brain and affect the function of the nerve cells.

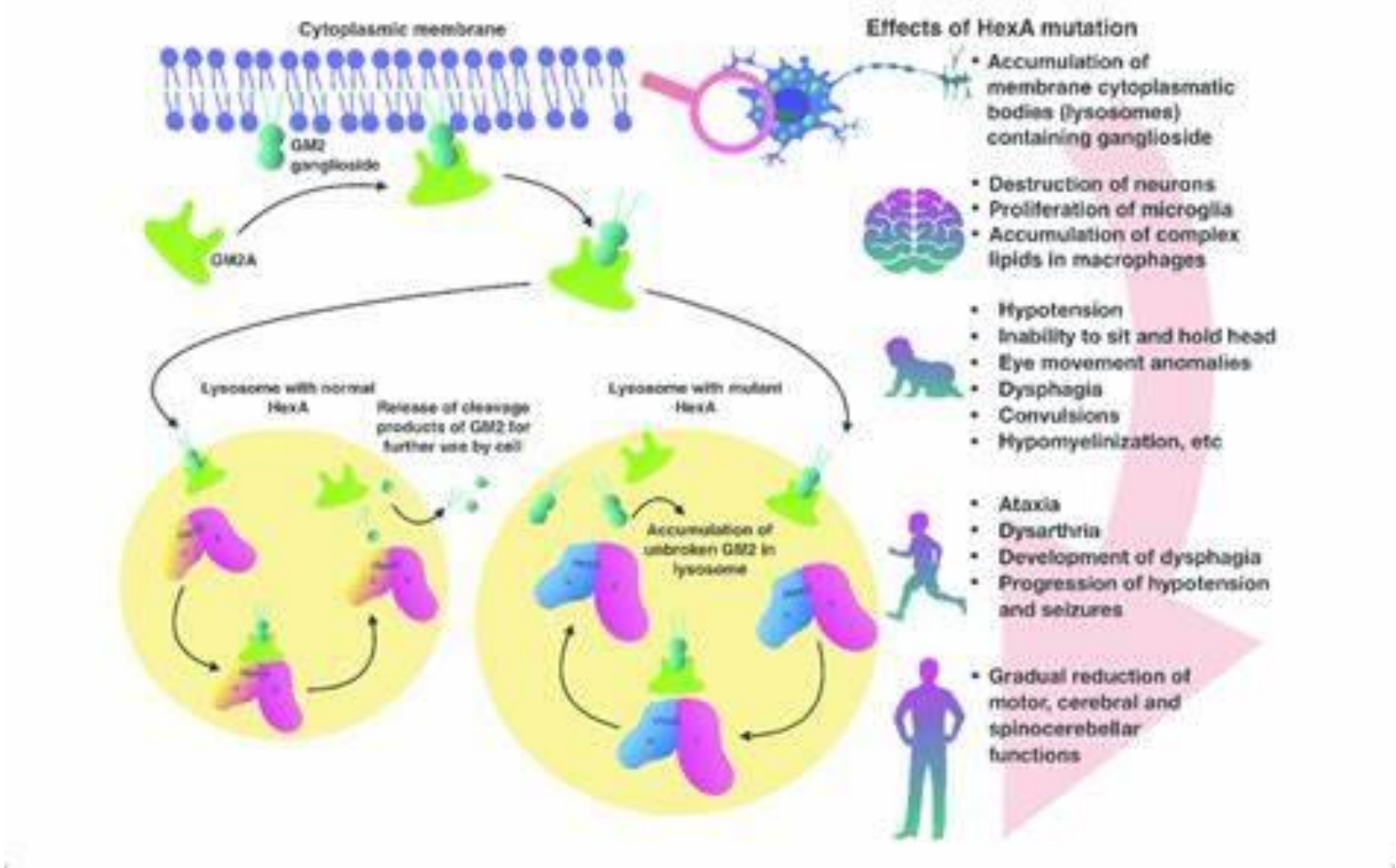


MECHANISM

- Tay-Sachs is an autosomal recessive disease caused by mutations in both alleles of a gene (HEXA) on chromosome 15
- HEXA codes for the alpha of the enzyme beta- hexosaminidase A.
- Normally beta-hexosaminidase A helps to degrade a lipid called CM2 ganglioside.
- In Tay-Sachs individuals, the enzymes is absent or present only in very reduced amounts.

TYPES OF TAY-SACHS DISEASE

- Tay-Sachs disease is classified based on the time of onset of neurological symptoms.
- Infantile TSD-Between 3 and 10 month(Death before the age of 4 to 5 years)
- Juvenile TSD-Between 2 and 10 months ,Extremely rare (die between 5-15 years)
- Adult/Late onset TSD-Between 20 and 30 years usually non fatal


Cytoplasmic membrane
GM2 ganglioside
GM2A
Lysosome with normal HexA
Release of cleavage products of GM2 for further use by cell
Lysosome with mutant HexA
Accumulation of unbroken GM2 in lysosome
Effects of HexA mutation
• Accumulation of membrane cytoplasmatic bodies (lysosomes) containing ganglioside
• Destruction of neurons
• Proliferation of microglia
• Accumulation of complex lipids in macrophages
• Hypotension
• Inability to sit and hold head
• Eye movement anomalies
• Dysphagia
• Convulsions
• Hyponyelinization, etc
• Ataxia
• Dysarthria
• Development of dysphagia
• Progression of hypotension and seizures
• Gradual reduction of motor, cerebral and spinocerebellar functions

DIAGNOSIS

- Simple blood test: To measure the body's level of hexosaminidase A. Tay-Sachs lack the most or all of this protein whereas levels are reduced in other forms of hexosaminidase A deficiency.
- Genetic testing
- Eye examination.

TREATMENT

- There is currently no cure or effective treatment for Tay-Sachs disease, but your health team can offer palliative care options to ease symptoms. This may include nutritional support and medicines to treat seizures.
- It can be challenging to care for a child with a life-limiting illness. Psychological support is recommended for the whole family.

REFERENCES

- https://medlineplus.gov/genetics/condition/color-vision-deficiency/
- Elles, R.,Mountfield,R.(2011).Molecular Diagnosis of Genetic Diseases. Springer Publication