Hereditary Fundus Dystrophies

By

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Generalized Photoreceptor dystrophy

They include the following:

- Retinitis pigmentosa.
- Progressive cone dystrophy.
- Leber congenital amaurosis.
- Stargardt disease and fundus flavimaculatus
- Bietti crystalline corneo-retinal dystrophy.
- Alport syndrome.
- Familial benign flecked retina.
- Pigmented paravenous chorio--retinal atrophy.
- Congenital stationary night blindness.
- Congenital monochromatism.

Retinitis Pigmentosa

It may be typical or atypical

Typical retinitis pigmentosa

Presentation: starts with nyctalopia & ends by drop of central vision.

Signs:
- Arteriolar attenuation.
- Pigmentary disturbances.
- Beaten-metal glistening reflex at the macula (ILM changes).
- Consecutive optic atrophy.
- Maculopathy e.g epimacular membrane, cystoid macular oedema & atrophic maculopathy.

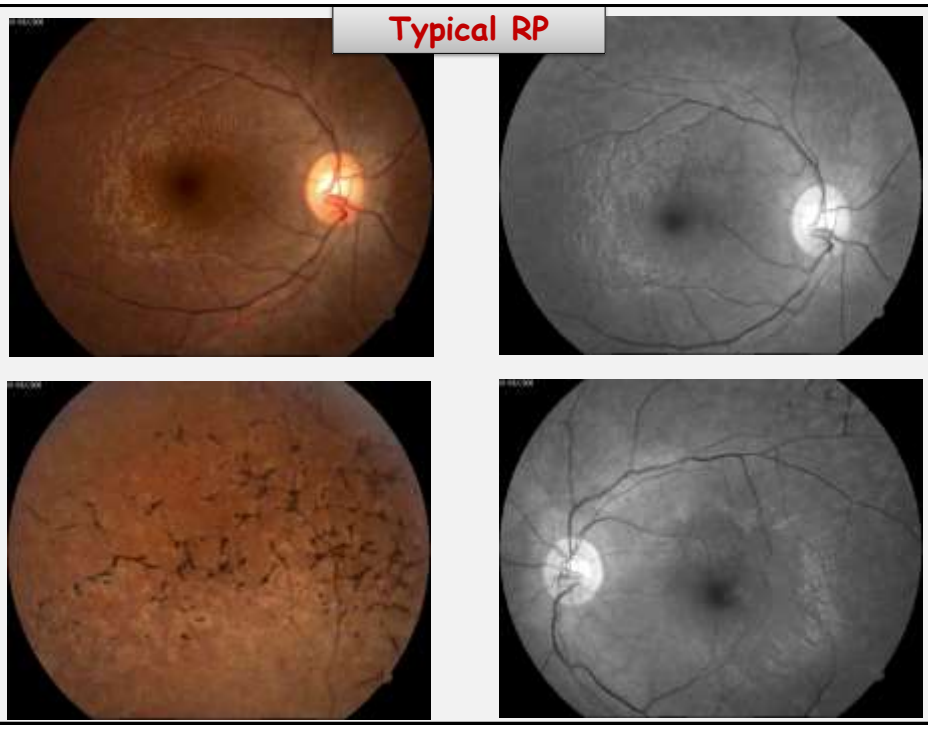
Investigations:

- ERG, EOG, perimetry, dark adaptometry, fluorescein angiography & OCT.

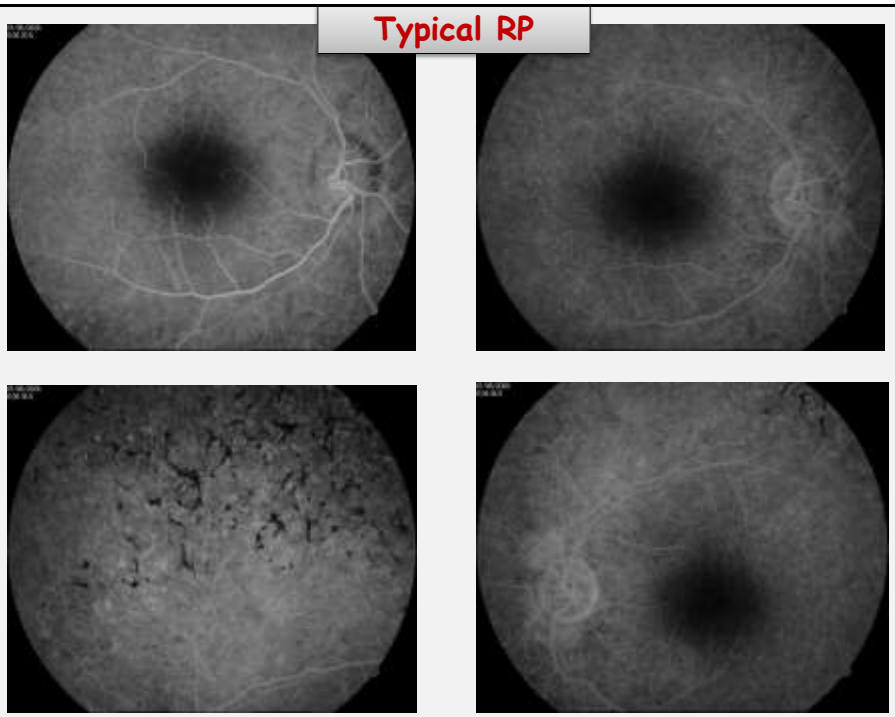
Atypical retinitis pigmentosa

- Cone-rod dystrophy: with earlier cone affection, starts with loss of central vision.
- RP sine pigmento: with paucity or absence of pigment accumulation which may appear later in the disease.
- Retinitis punctate albesens: characterized by scattered whitish – yellow spots., most numerous at the equator, usually sparing the macula.
- Sector RP: affecting sector of the retina.

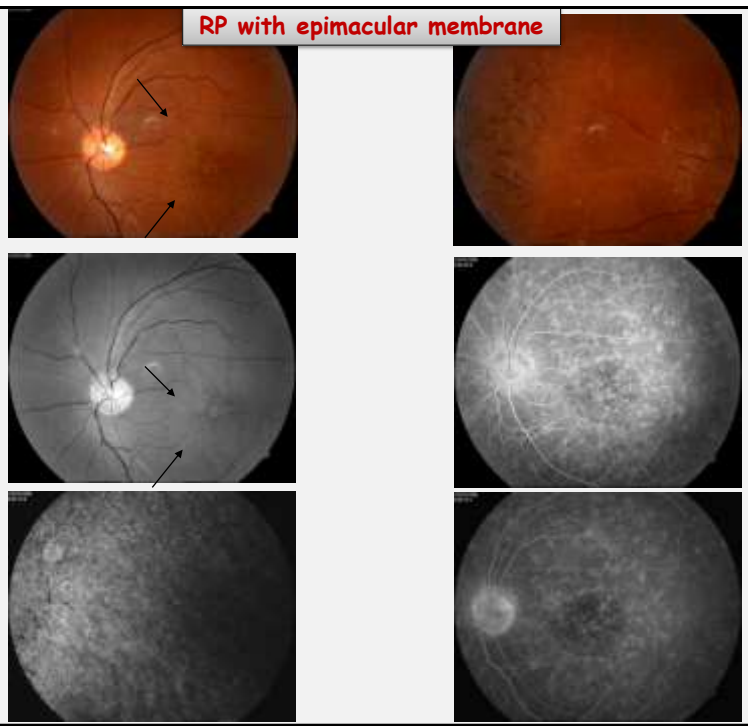
Typical RP



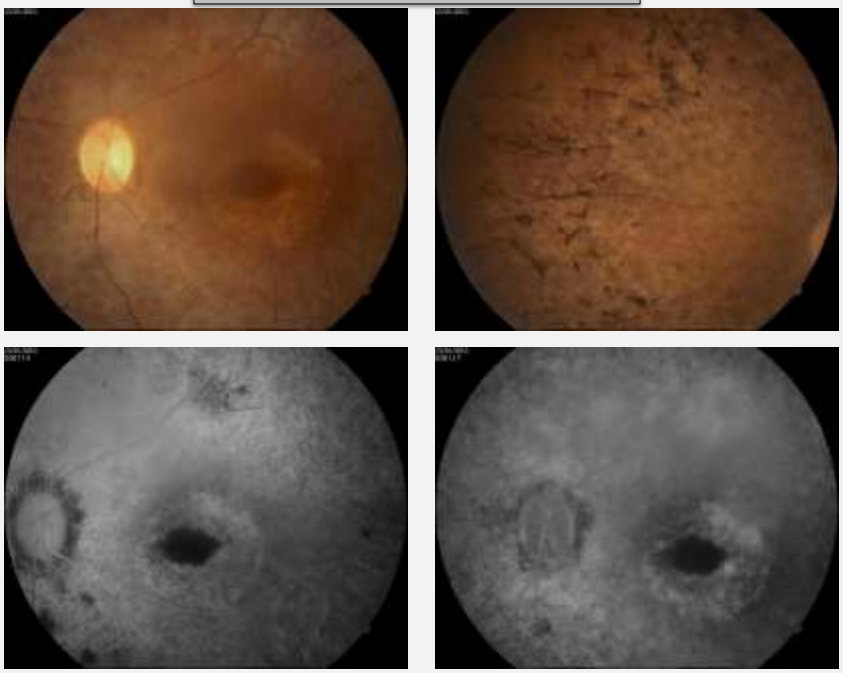
Typical RP



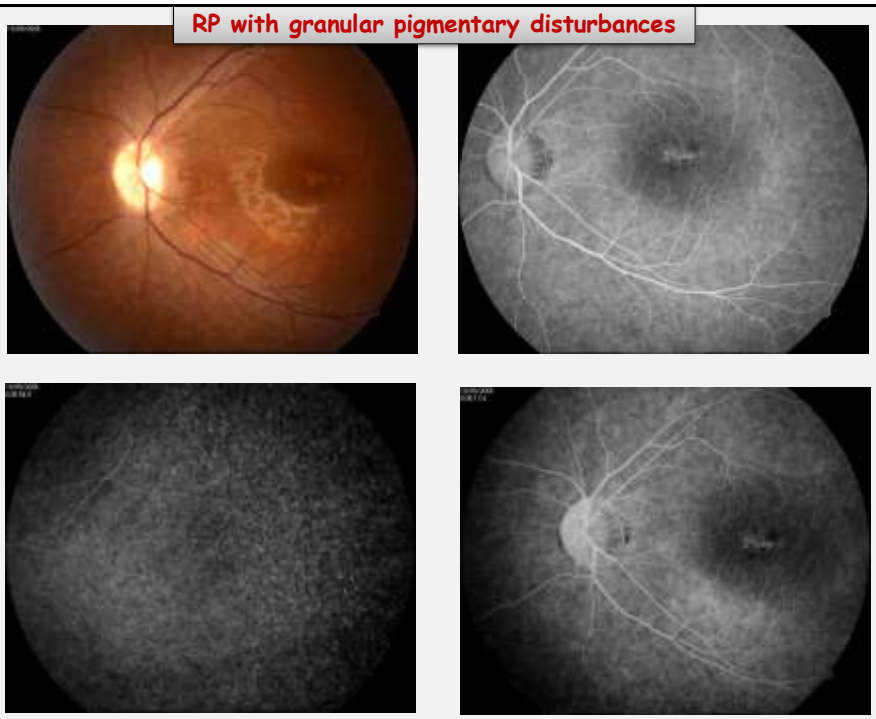
RP with epimacular membrane



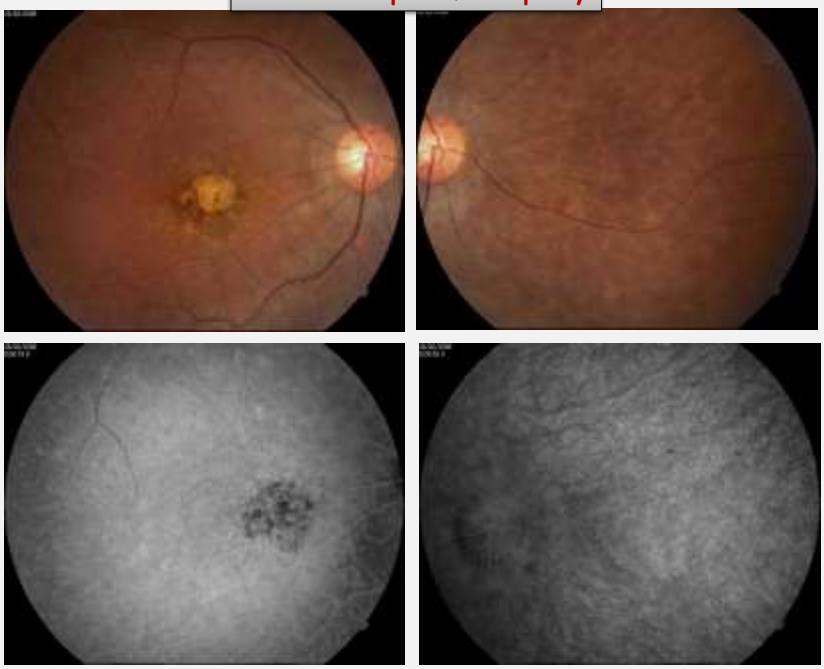
RP with Bull's eye like maculopathy



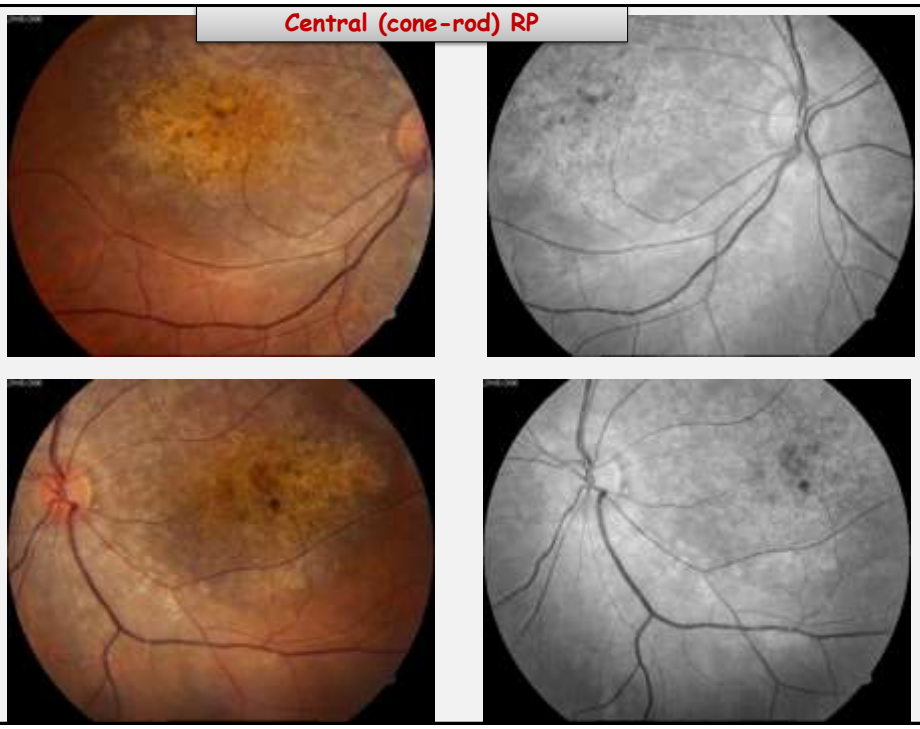
RP with granular pigmentary disturbances



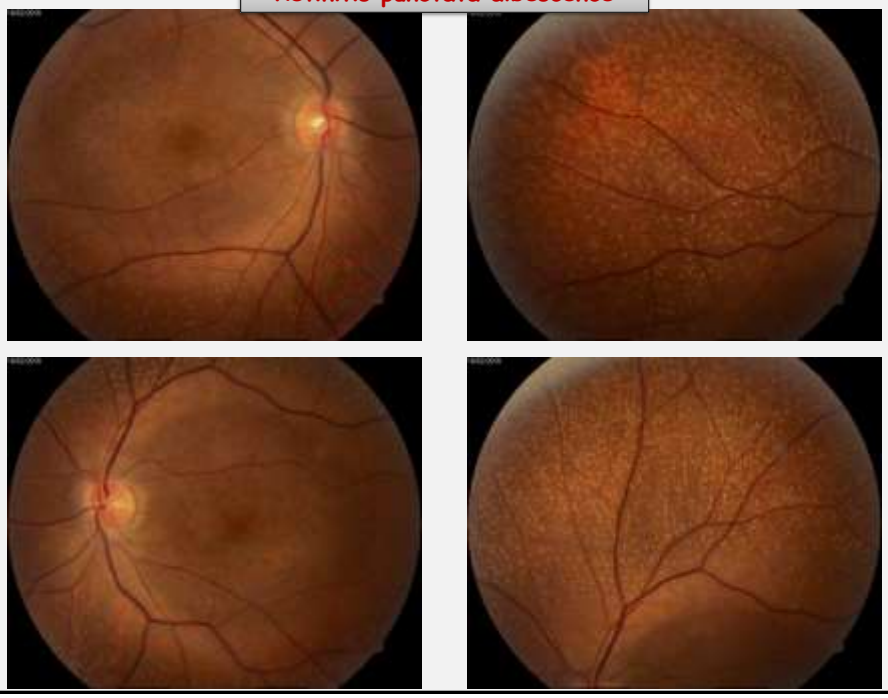
RP with atrophic maculopathy



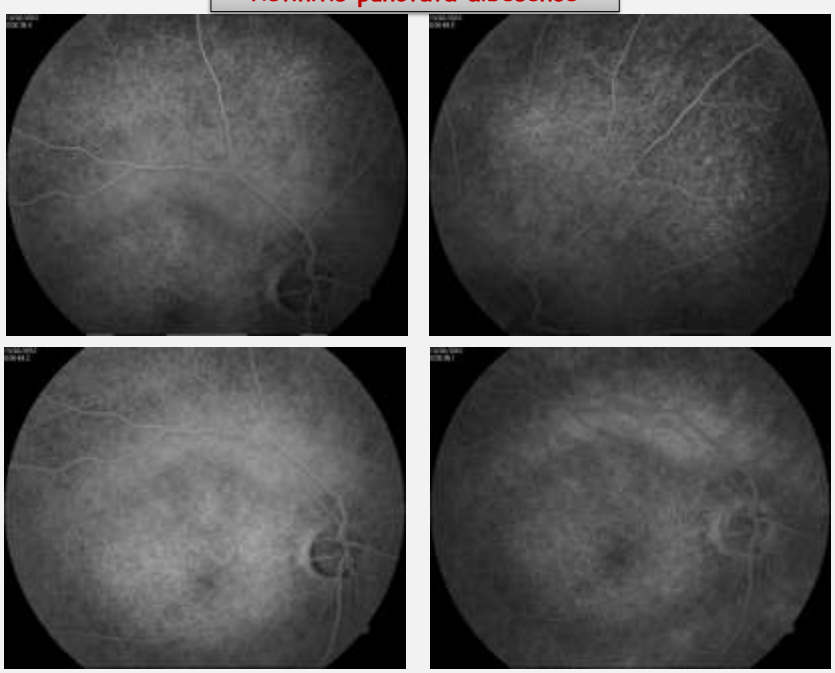
Central (cone-rod) RP



Retinitis punctata albescence



Retinitis punctata albesence



Progressive cone dystrophy

Presentation: gradual bilateral change of central and colour vision which may be followed by photophobia.

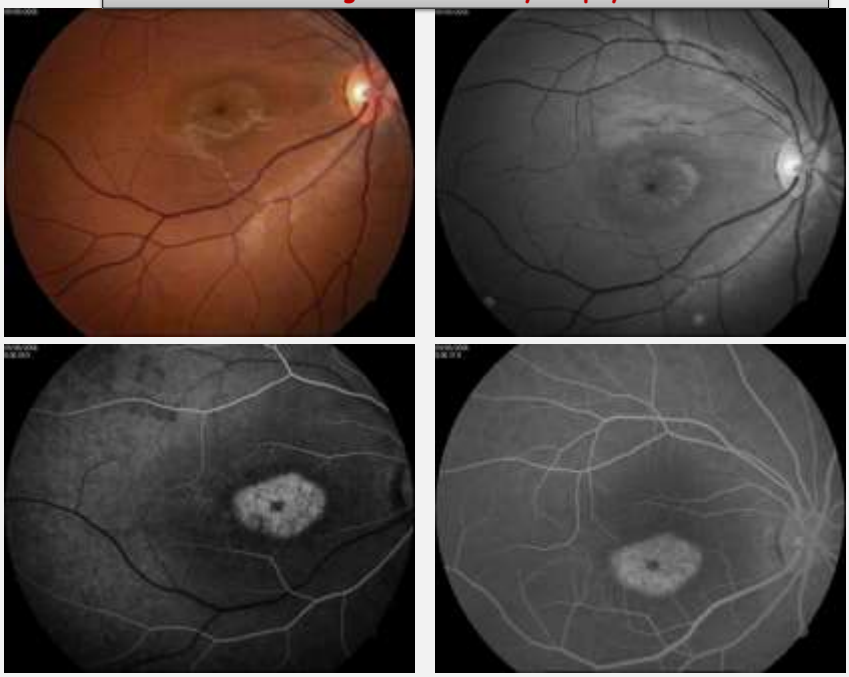
Signs (chronologically):

- Normal appearance or subtle pigmentary changes at the macula.
- Golden sheen at the macula.
- Bull's eye maculopathy (usually hypopigmntation with hyperpigmented center).
- Geopgraphic atrophy of the macula.

Investigations:

- ERG, EOG, dark adaptometry & fluorescein angiography.

Progressive cone dystrophy



Leber congenital amaurosis

Presentation: blindness at birth or shortly after with roving eye movement or nystagmus.

Signs, variable and may include:

- Absent or diminished light reflex.
- Arteriolar attenuation.
- Severer pigment proliferation at the macula.
- Peripheral pigmentary changes.
- Oculodigital syndrome.
- Coloboma like atrophic maculopathy

Investigations:

- ERG.

Stargardt disease &funudus flavimaculatus

They are considered as same disease category with different presentation and prognosis. Characterized by diffuse lipofuscin pigment accumulation in RPE with vermilion fundus appearance and dark choroid in fluorescein angiography.

Fundus changes take any of the following appearance:

- Bull's eye maculopathy.
- Bull's eye maculopathy with perifoveal flecks (yellow white deposits at the level of RPE, of different sizes and shapes).
- Bull's eye maculopathy with scattered flecks allover the retina.
- Scattered retinal flecks without Bull's eye maculopathy.

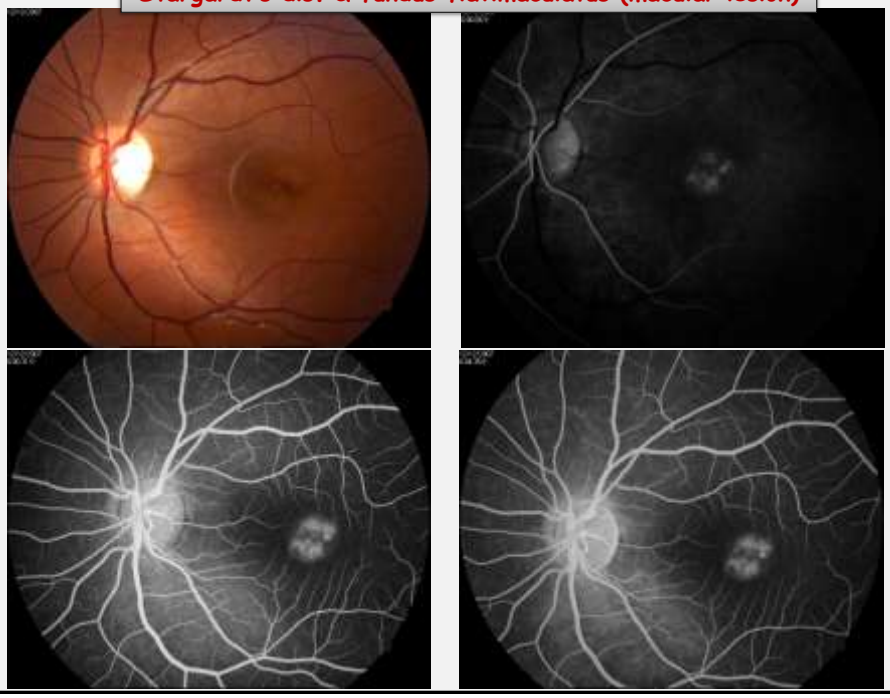
Visual acuity depends on state of the macula.

Macular lesion ends by geographic atrophy & may be complicated by CNV

Investigations:

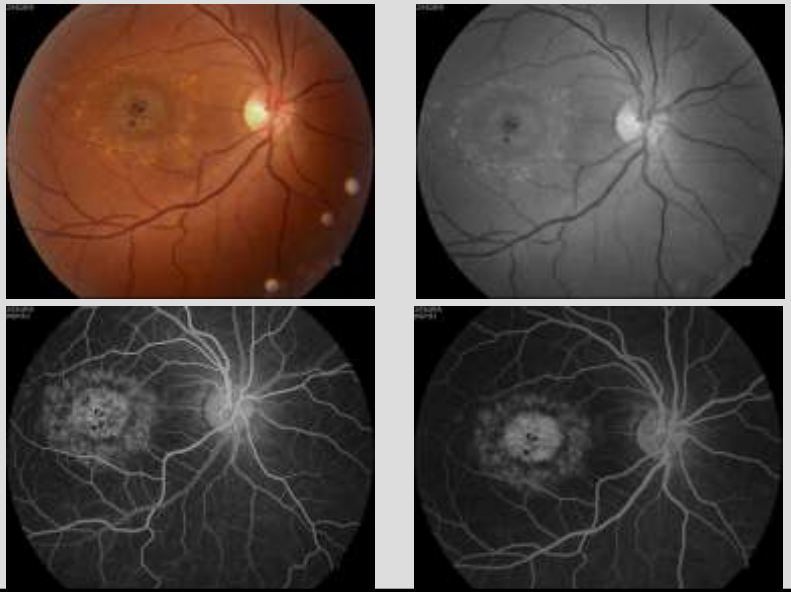
- ERG, EOG, FA, ICGA, & autofluorescence.

Stargardt's dis. & fundus flavimaculatus (macular lesion)

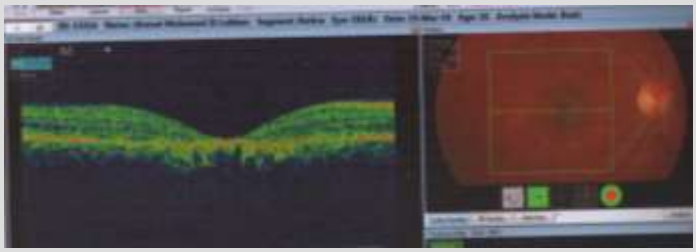


Stargardt's dis. & fundus flavimaculatus (macular lesion with perifoveal flecks)

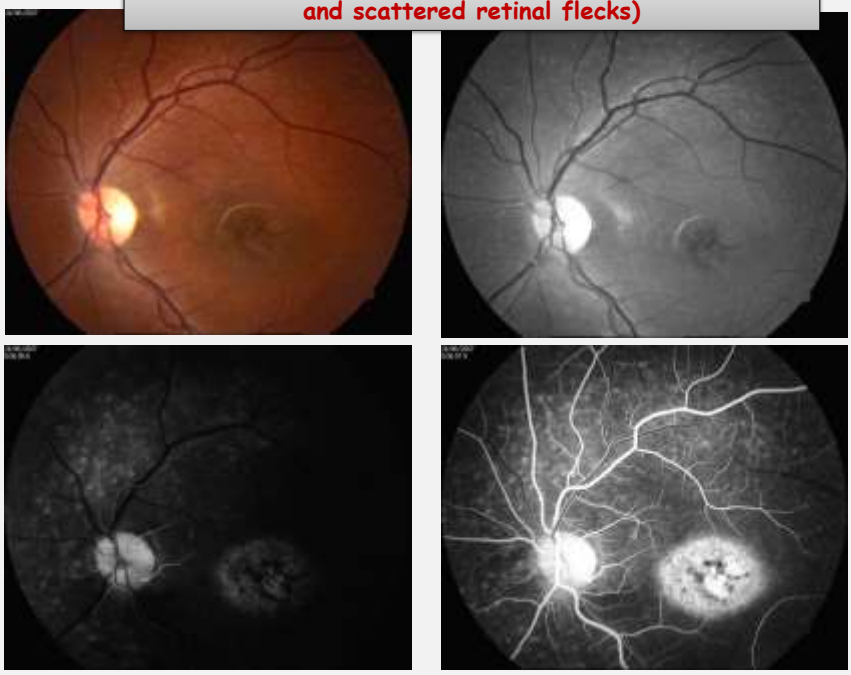
Stargardt's dis. & fundus flavimaculatus(macular lesion with perifoveal flecks)



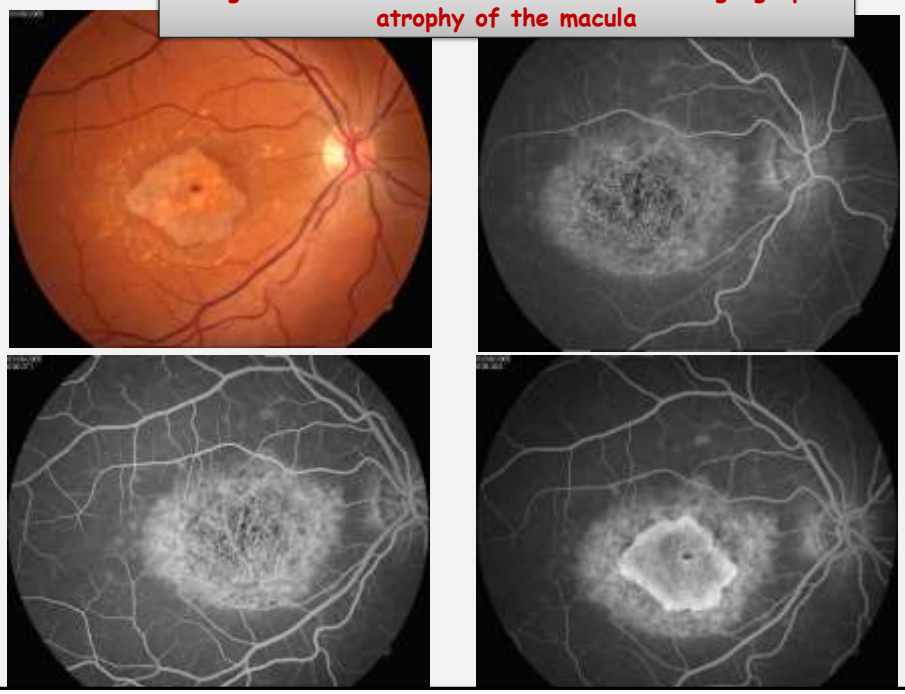
OCT of a case of Stargardt disease with atrophic maculopathy and perifoveal flecks



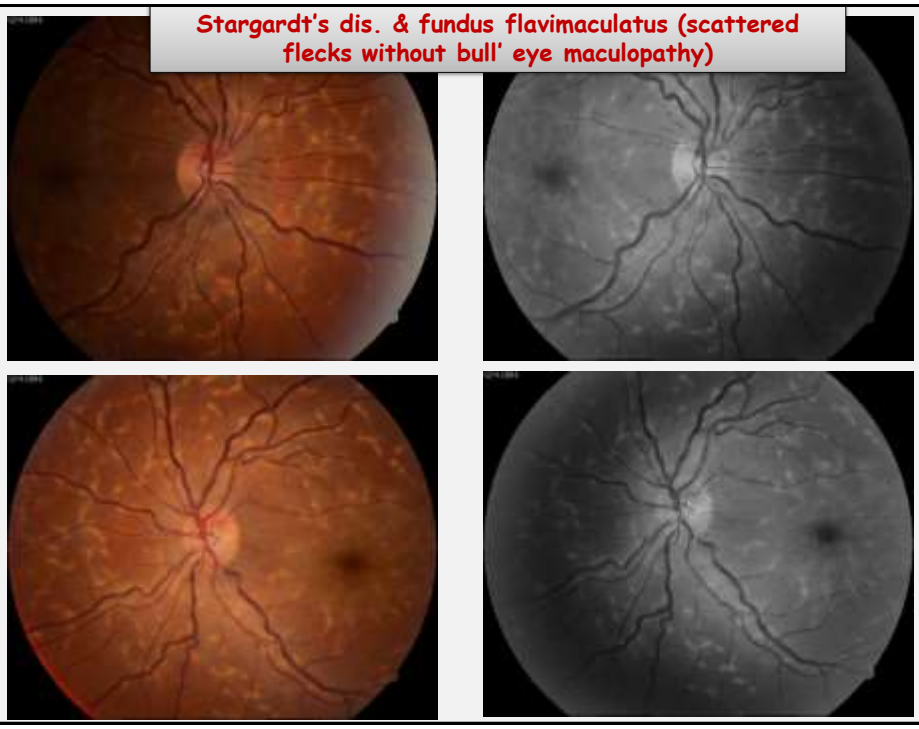
Stargardt's dis. & fundus flavimaculatus (macular lesion and scattered retinal flecks)



Stargardt's dis. & fundus flavimaculatus with geographic atrophy of the macula



Stargardt's dis. & fundus flavimaculatus (scattered flecks without bull' eye maculopathy)



Pigmented paravenous chorioretinal atrophy

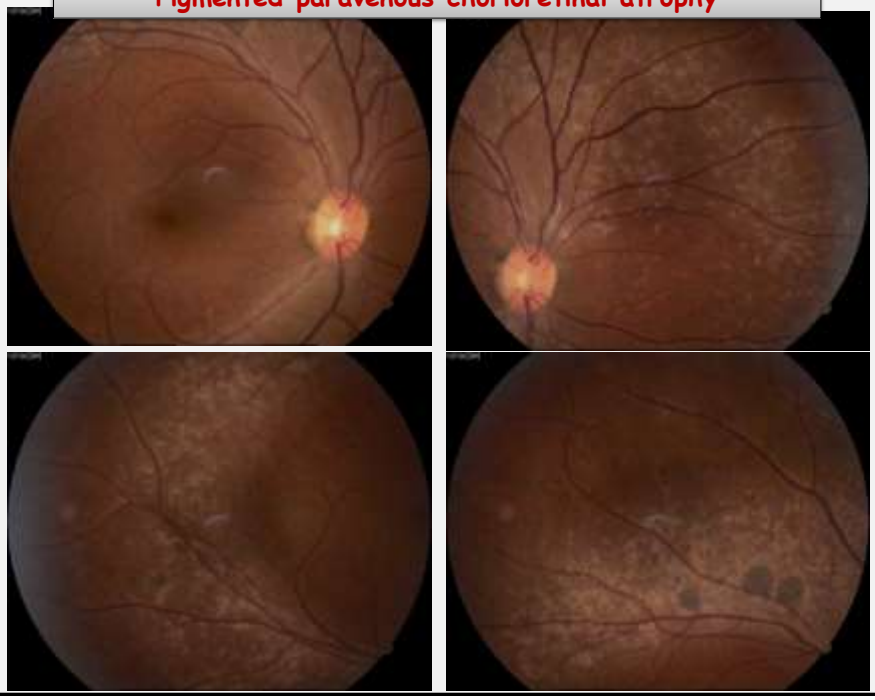
Presentation: usually innocuous, asymptomatic, stationary and accidentally discovered.

Signs:
- Sharply outlined atrophic patches along the major choroidal veins.
- Bone spicule like pigment deposits.
- Normal retinal blood vessels and optic disc.

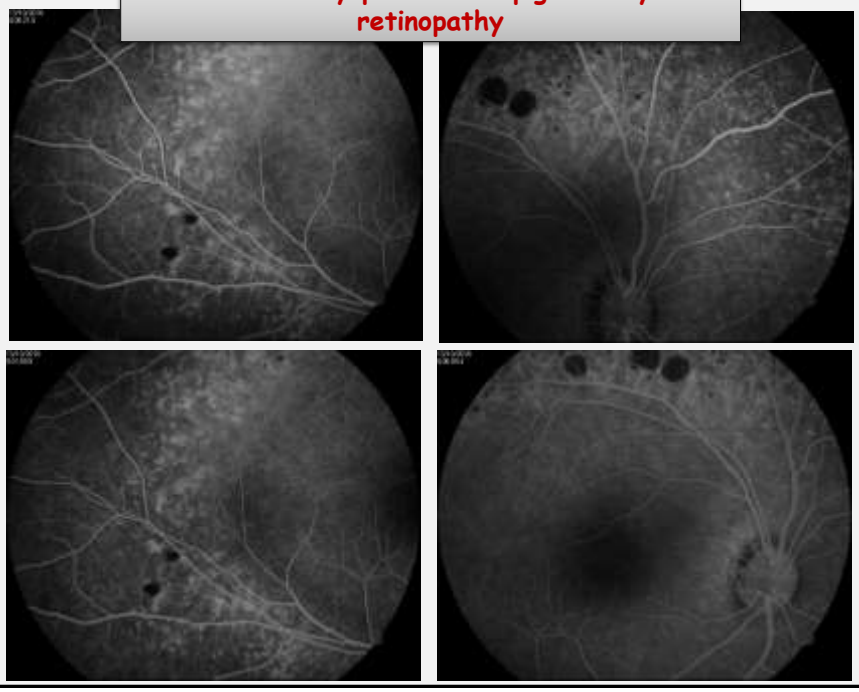
Investigations:

- ERG is normal.

Pigmented paravenous chorioretinal atrophy



Hereditary paravenous pigmentary retinopathy



Bietti crystalline corneo-retinal dystrophy

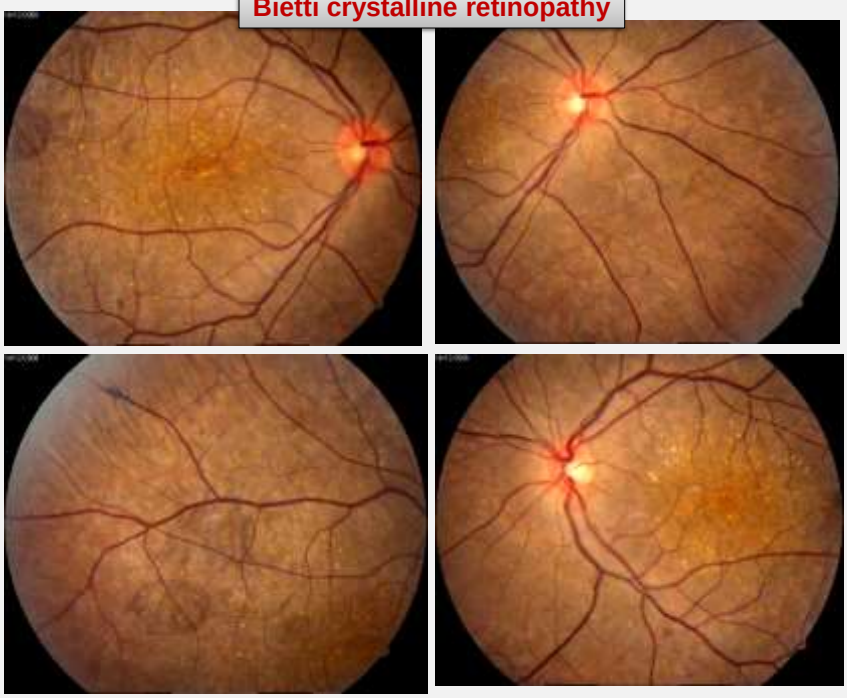
Presentation: third to fourth decade with slowly progressive vision loss

Signs:
- Variable coloured, crystals at all retinal layers level.
- Patches of atrophy of RPE and choriocapillares which extend from the macula to the periphery with diffuse affection and gradual disappearance of crystals.

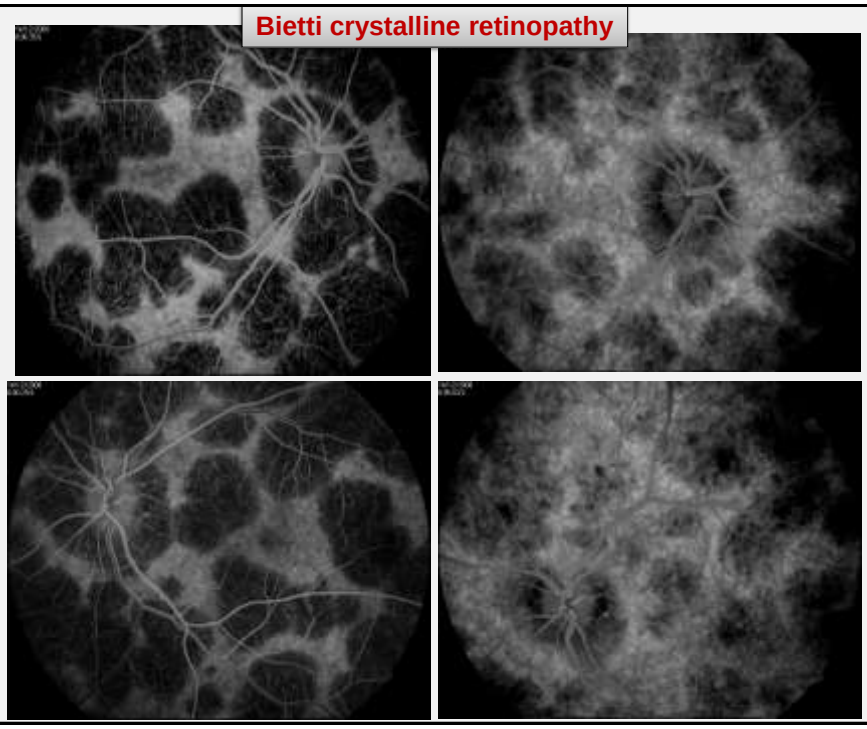
Investigations:

- ERG & FFA

Bietti crystalline retinopathy



Bietti crystalline retinopathy



Macular Dystrophies

They include the following:

- Juvenile Best macular dystrophy.
- Multifocal vitelliform lesions.
- Pattern dystrophy.
- North Carolina macular dystrophy.
- Familial dominant drusen.
- Sorsby pseudo-inflammatory dystrophy.
- Benign concentric annular macular dystrophy.
- Central areolar choroidal dystrophy.
- Domiant cystoid macular oedema.
- Sjogren-Larsson syndrome.
- Familial internal limiting membrane dystrophy.

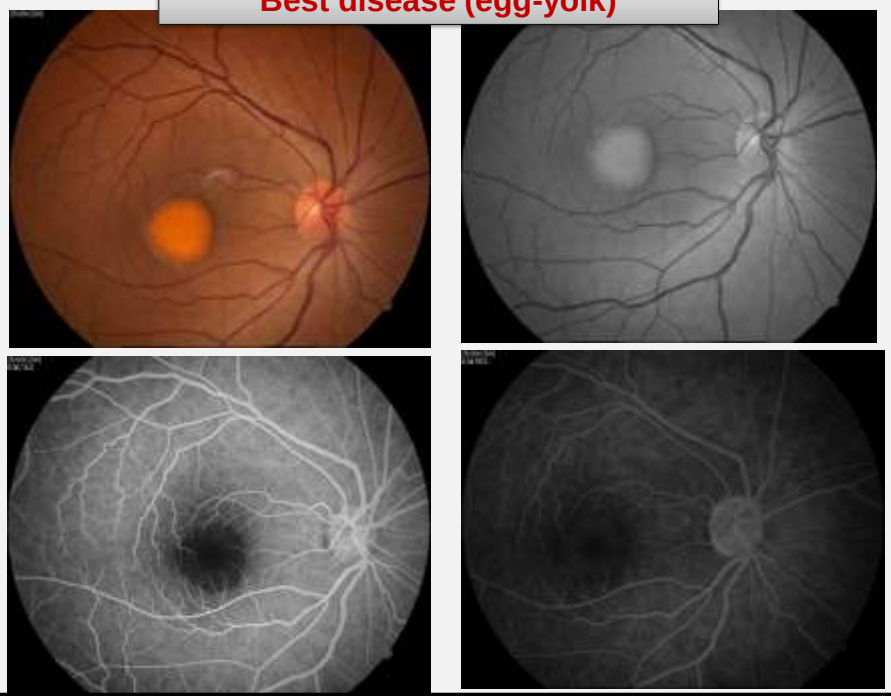
Juvenile Best macular dystrophy

It evolves in the following stages:

- Pre-vitelliform stage: subnormal EOG with normal fundus in asymptomatic child.
- Vitelliform stage: rounded sharply delineated (sunny, side-up, egg yolk) macular lesion about half to one disc diameter.
- Pesudohypopyon stage: appears when part of the lesion is absorbed.
- Vitelliruptive sstage: when egg yolk, breaks up (scramble).
- Atrophic stage: when allpigment disappears leaving a patch of RPE atrophy.

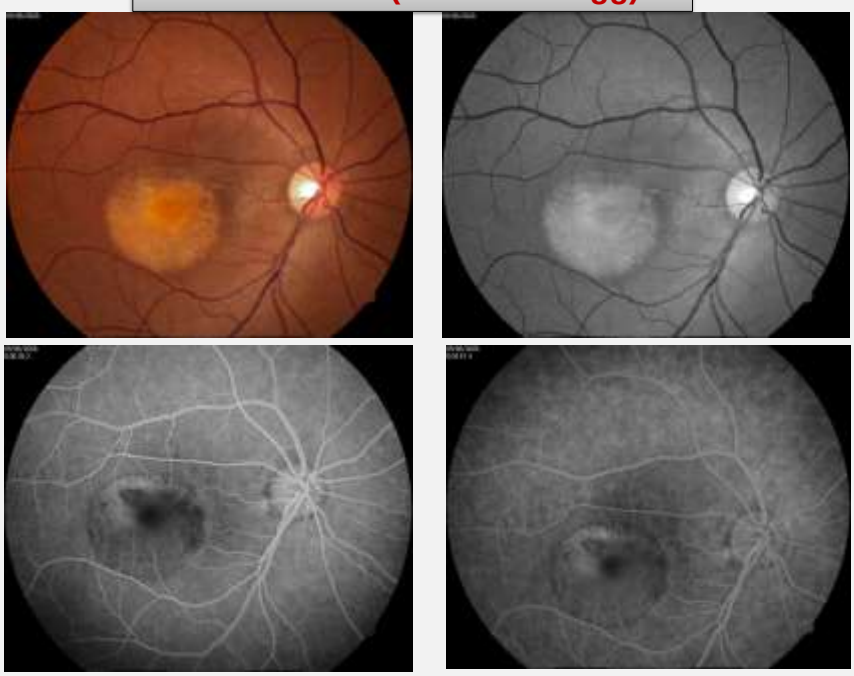
Investigations: EOG, FFA & OCT.

Best disease (egg-yolk)

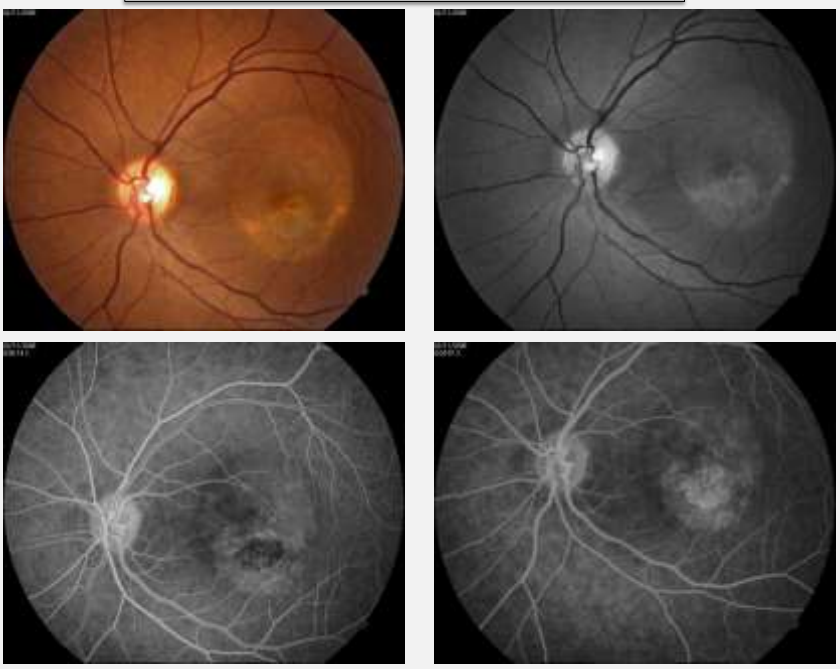


Best disease (pseudo-hypopyon)

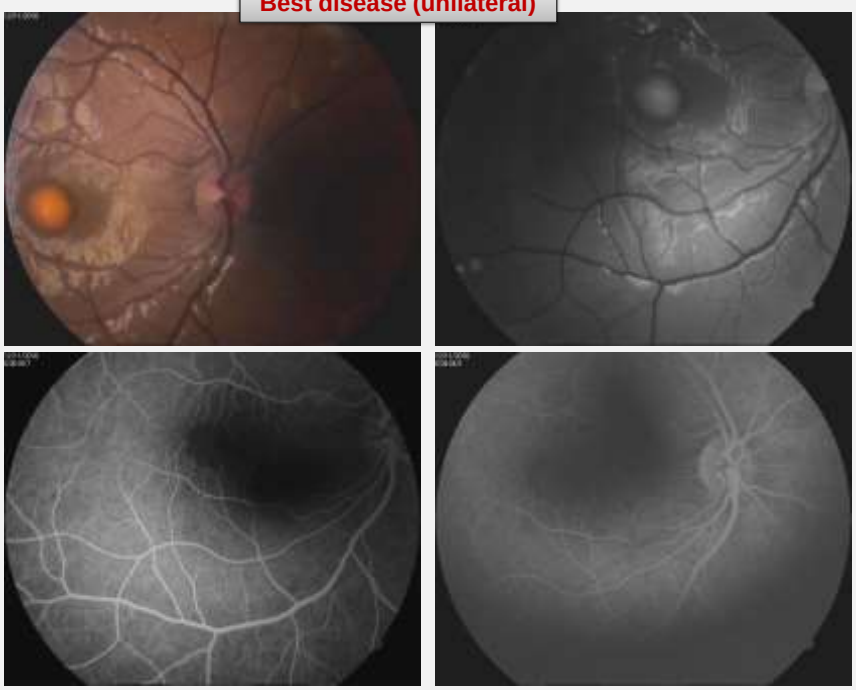
Best disease (scrambled-egg)

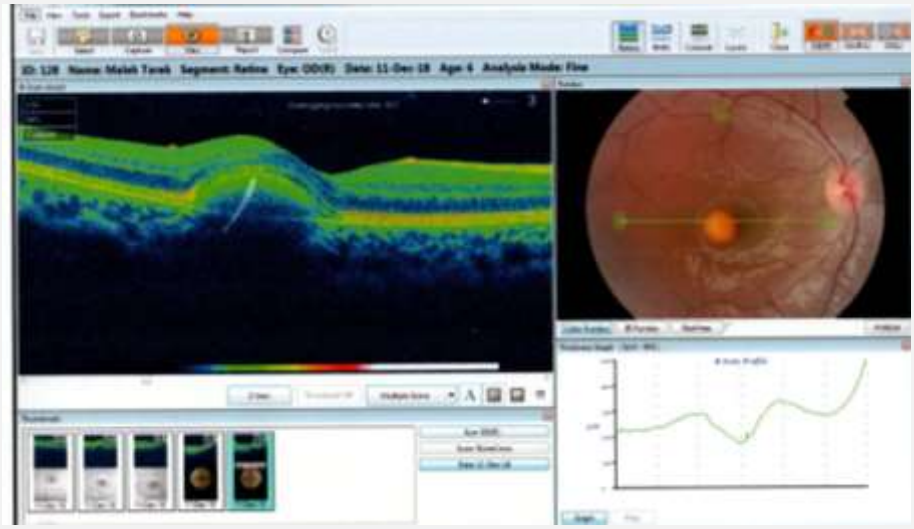


Best disease (atrophic stage)



Best disease (unilateral)

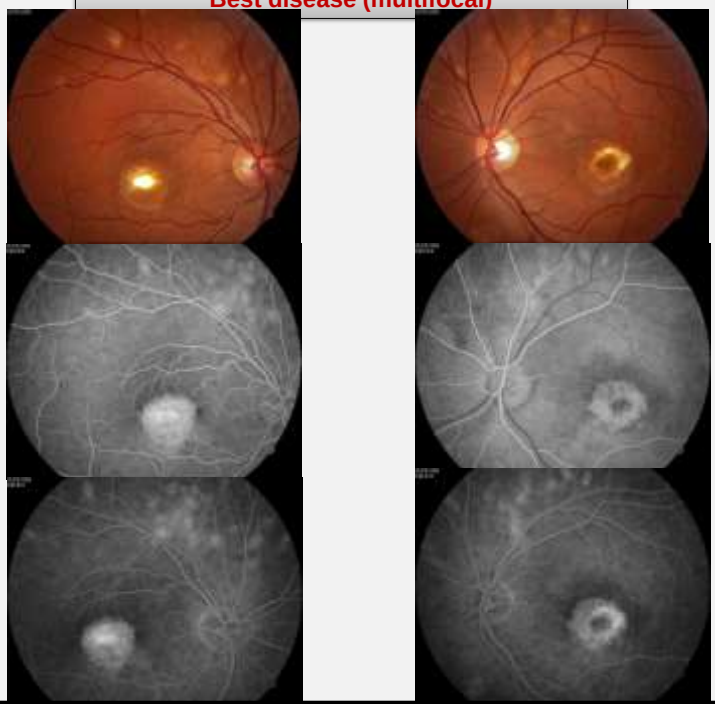




Multifocal vitelliform lesions

- Presentation is with multifocal vitelliform lesions (lipofuscin pigment deposition).
- Some lesions undergo atrophy while other fresh lesions appear.
- May or may not be associated with macular lesion.

Best disease (multifocal)



Pattern dystrophy

It includes a variety of retinal dystrophies characterized by yellow, orange or grey macular lesion of different morphologies, with lipofuscin pigment deposition at the level of RPE.

The common characters are:

- Bilateral, symmetrical affection.
- Relatively benign course.
- Normal ERG & occasional abnormal EOG.

It includes:

- Adult-onset, vitelliform macular dystrophy.
- Butterfly-shaped, macular dystrophy.
- Multifocal pattern dystrophy simulating fundus flavimaculatus .
- Mo[illegible] pattern dystrophy

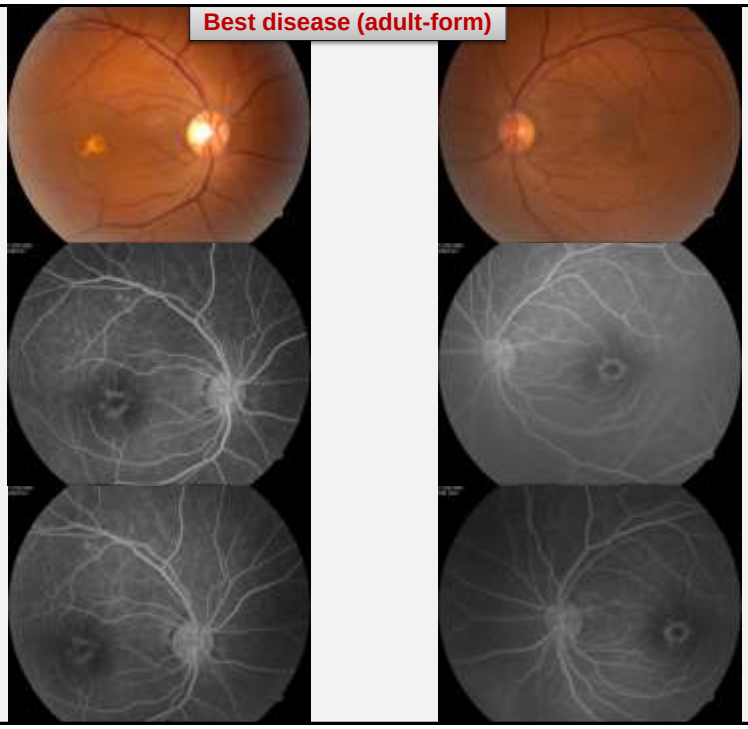
Adult-onset vitelliform macular dystrophy

Presentation: usually in the fourth to six decades of life. By mild to moderate drop of vision and sometimes metamorphopsia, however, in most cases, the condition is discovered by chance.

Signs: bilateral, symmetrical, rounded or oval, slightly elevated, yellow subfoveal deposit, about third disc diameter, often with hhyperpigemnted center.

Investigations: FFA often shows hypofluorescnt center surrounded with hyperfluorescent small irregular ring.

Best disease (adult-form)



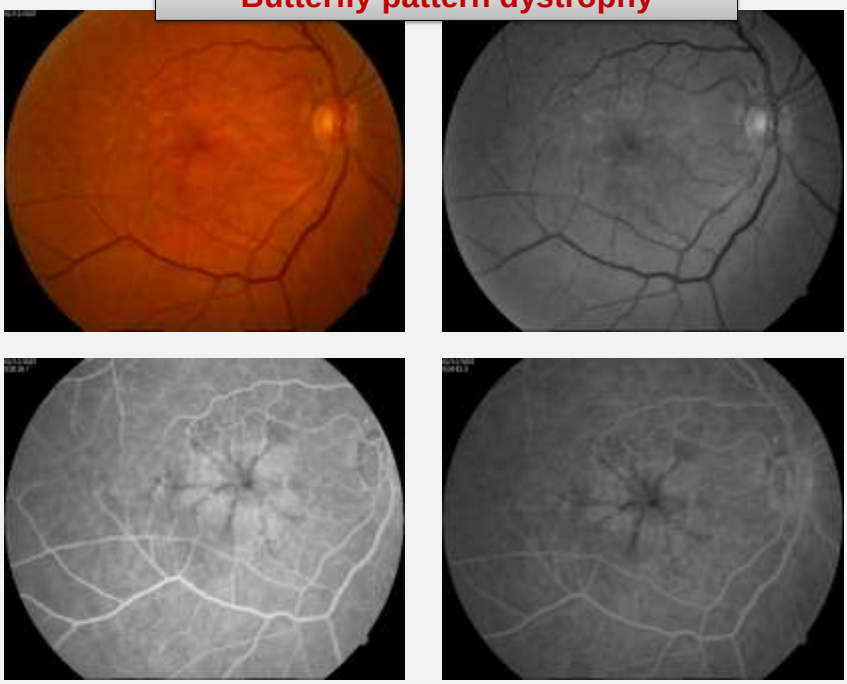
Butterfly-shaped, pattern dytsrophy

Presentation: usually in the second to third decade by mild impairment of vision, but in most cases, it is asymptomatic.

Signs: yellow pigment at the fovea, arranged in radiate manner.
atrophic maculopathy is rare.

Investigations: FFA.

Butterfly pattern dystrophy



Sorsby pseudo-inflammatory macular dystrophy.

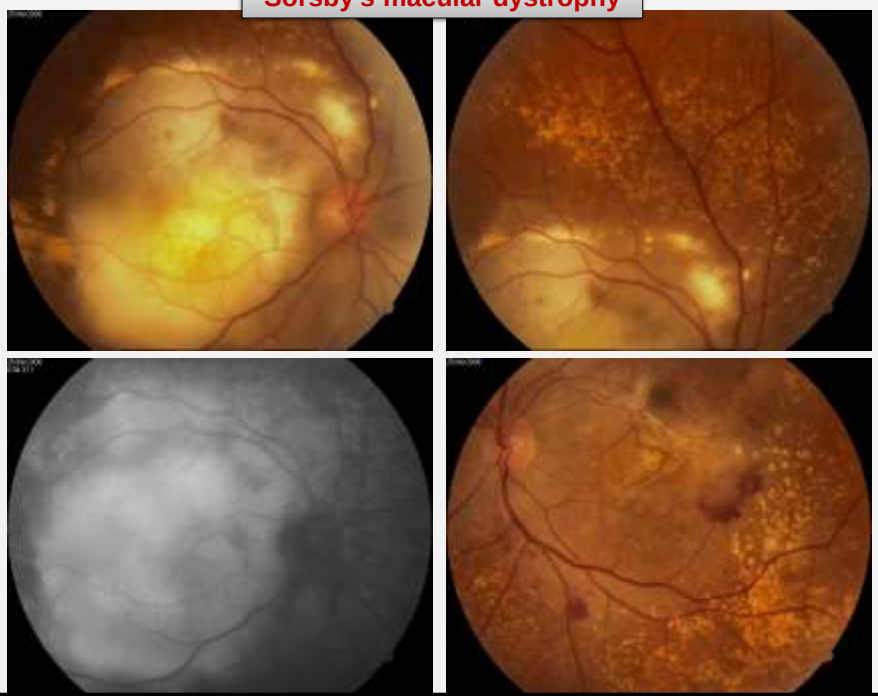
- Also called, haemorrhagic macular dystrophy.
- Very rare, leads to bilateral vision loss in the fifth decade of life.

Presentation: with nyctalopia in the third decade or sudden vision loss due to exudative maculopathy in the fifth decade.

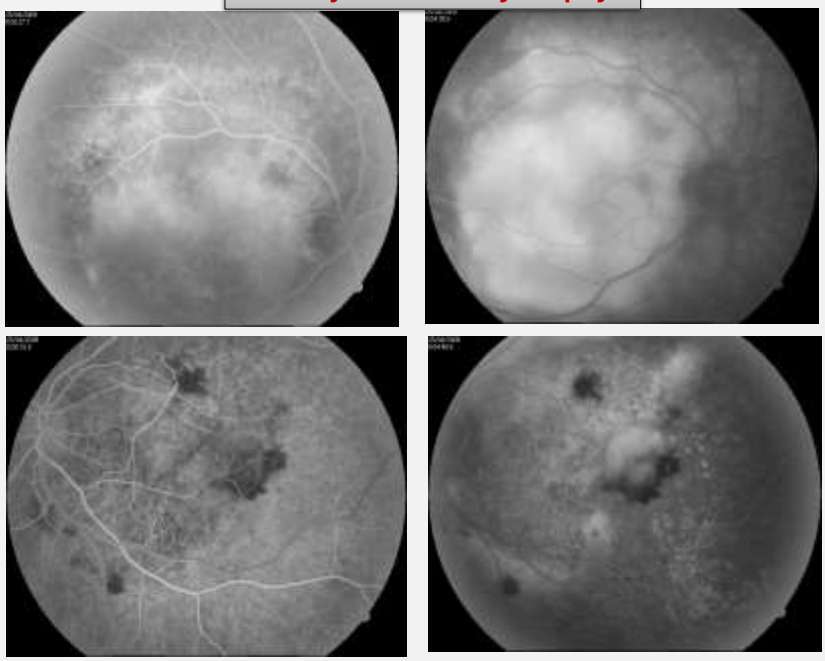
Signs (chronologically):

- Yellow white, confluent, drusen-like deposits along the arcades, nasal to the optic disc and mid-periphery.
- CNV with exudative maculopathy.
- Subretinal scarring.
- peripheral chorio-retinal atrophy may occur in the seventh decade.

Sorsby's macular dystrophy



Sorsby's macular dystrophy



Familial internal limiting membrane dystrophy

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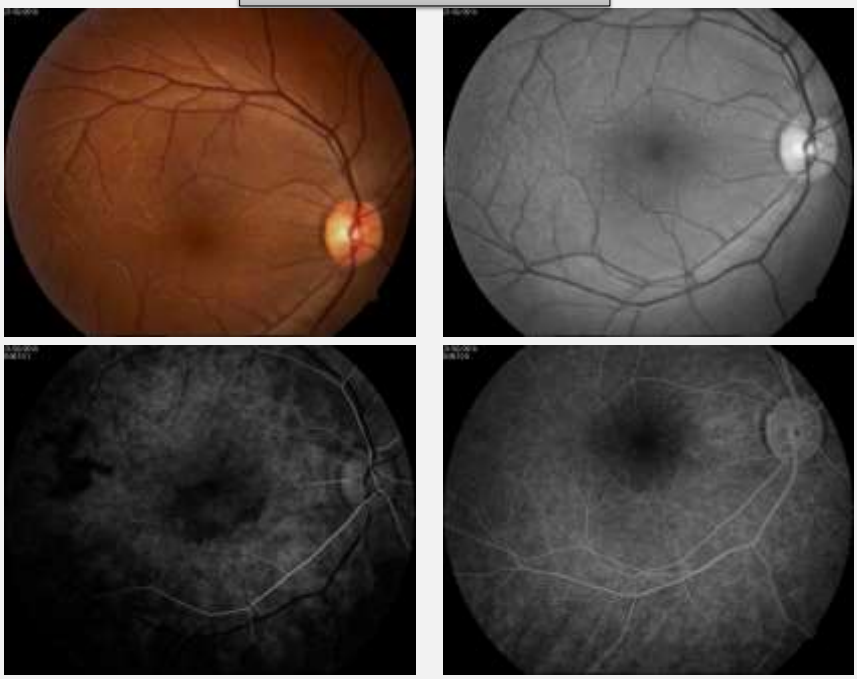
Presentation: in third to fourth decade with vision loss.

Signs:

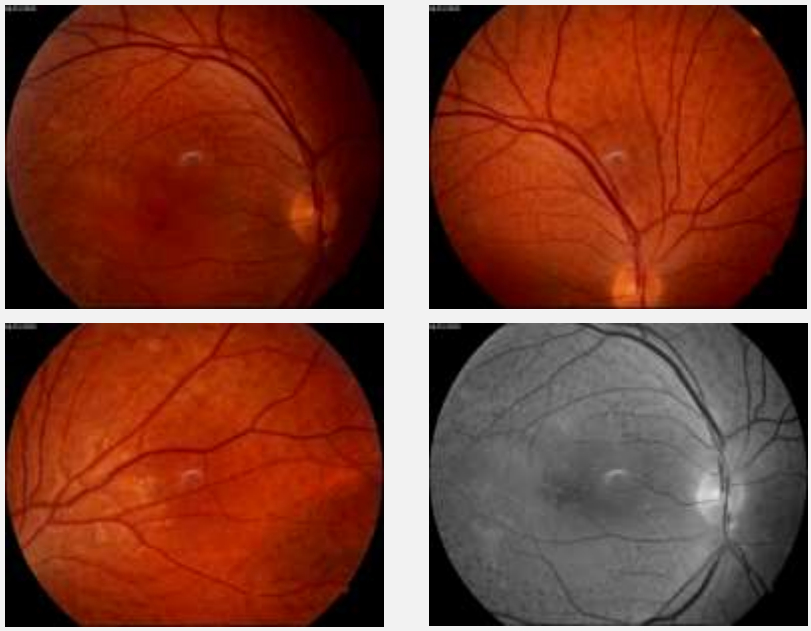
- Glistening inner retinal surface.
- OCT shows epiretinal membranes, schitic cavities in the retina & cystoid macular oedema.

Investigations: ERG (selective diminution of b-wave), FFA (retinal leakage) & OCT.

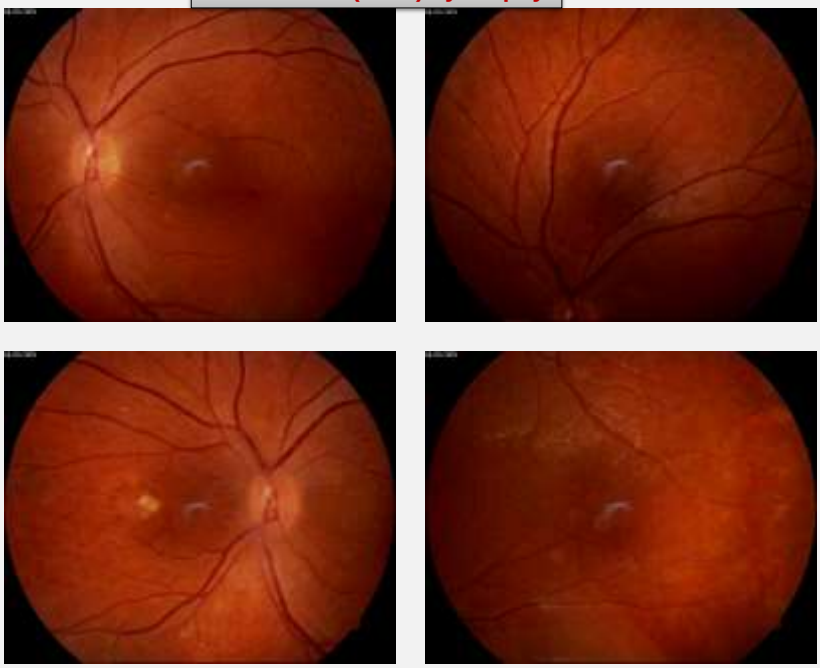
Familial ILM (FILM) dystrophy



Familial ILM (FILM) dystrophy



Familial ILM (FILM) dystrophy



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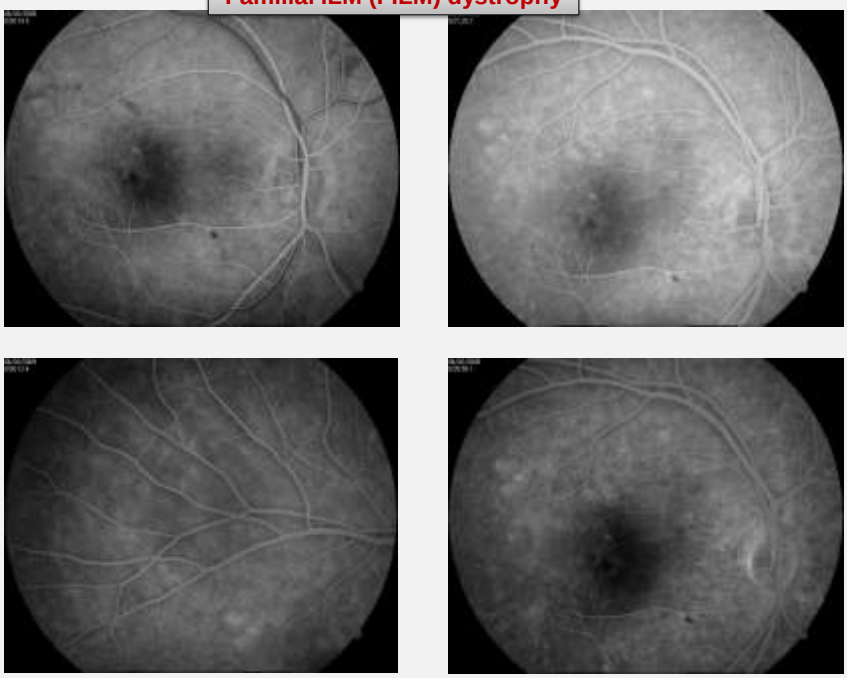
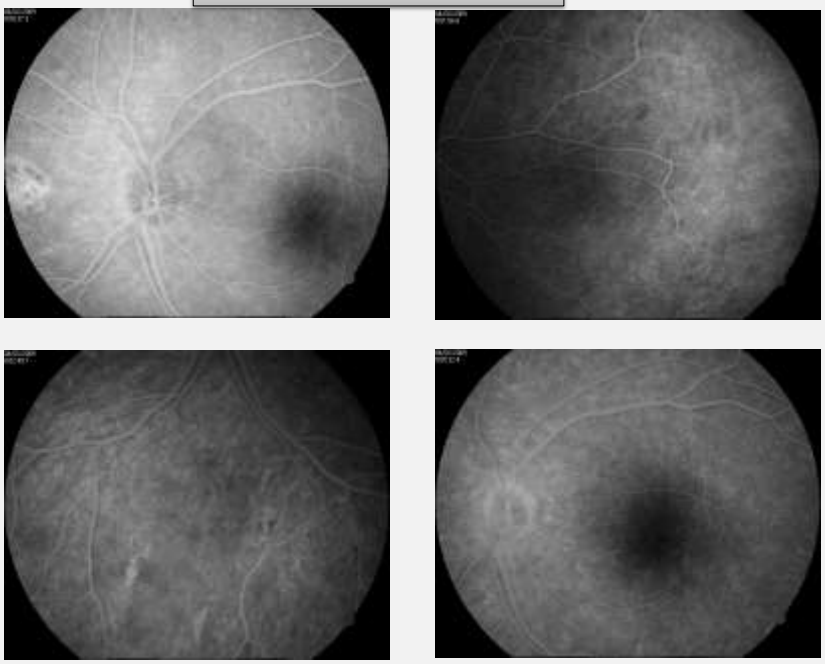


Familial ILM (FILM) dystrophy



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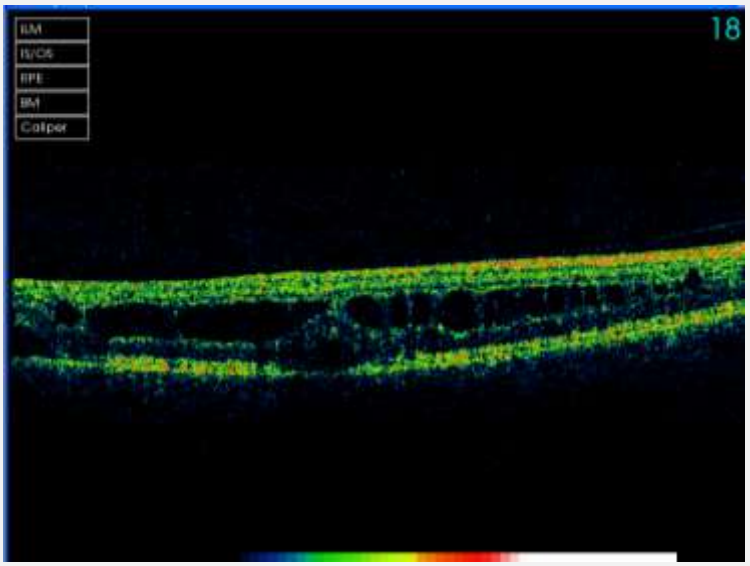
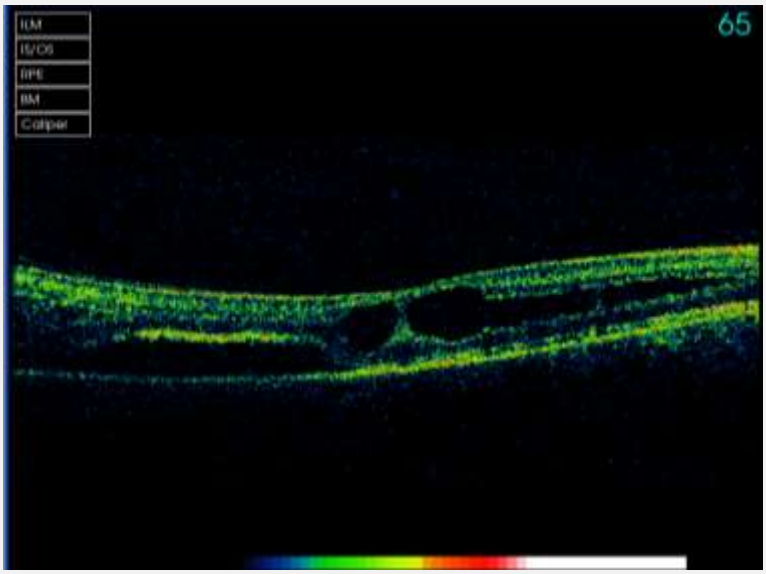
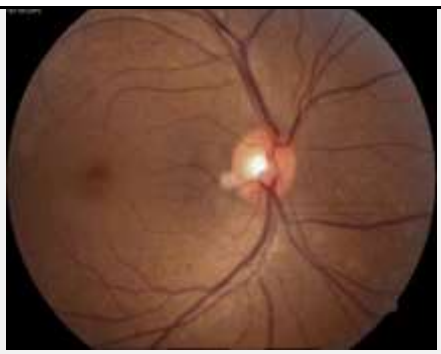
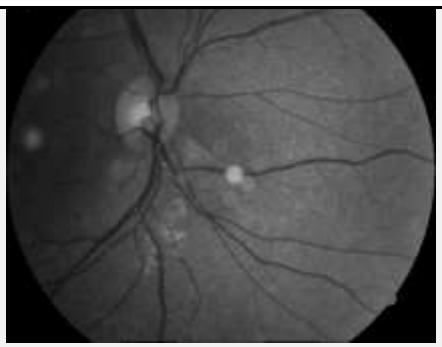
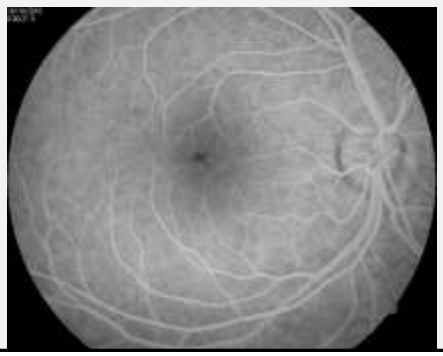


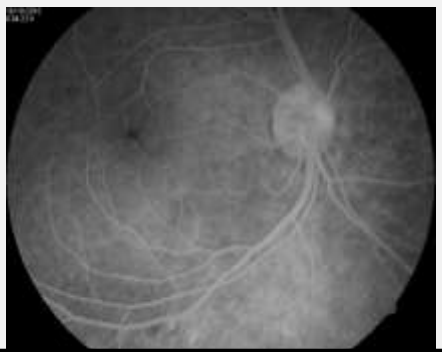
Familial ILM (FILM) dystrophy











Vitreo-retinal dystrophies

They include the following:

- Juvenile, x-linked retinoschisis.
- Stickler syndrome.
- Wagner syndrome.
- Familial exudative vitreo-retinopathy.
- Enhanced s-cone syndrome & Goldmann-Favre syndrome.
- Snowflake vitreoretinal degeneration.
- Dominant inflammatory neovascular vitreoretinopathy.
- Dominant vitreoretinochoroidopathy.
- Kneist dysplasia.

Juvenile, x-linked retinoschisis

Presentation: between 5 to 10 years old with difficult reading due to macular changes & rarely at infancy by squint or nystagmus with severe peripheral schitic changes.

Signs:

- **Foveal schisis:**

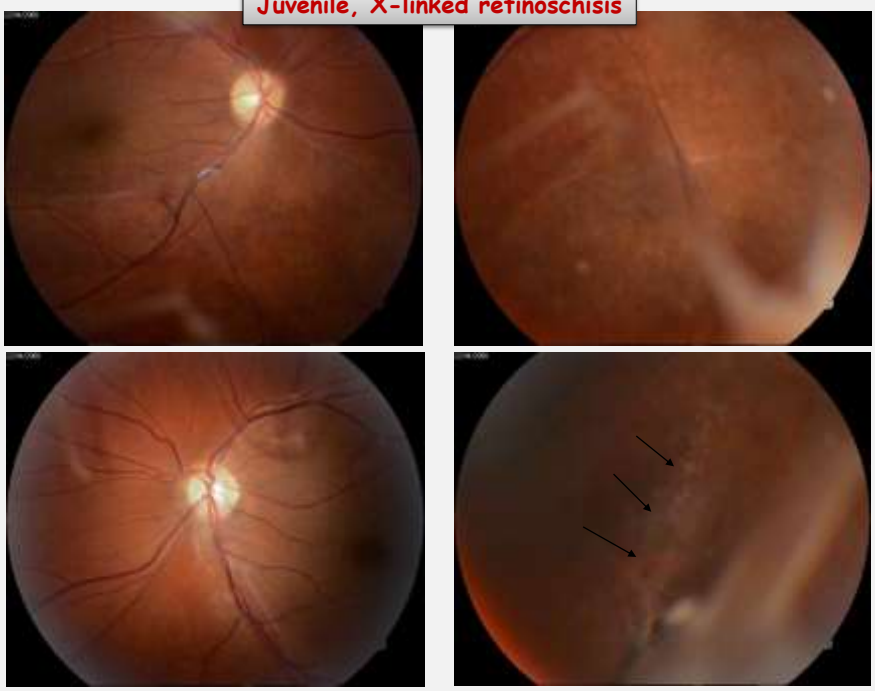
bicycle wheel radial striae with cystoid macular changes.
The striae become less evident with time with blunting of the foveal reflex.

- **Peripheral schisis:** the inner layer consists of the ILM & nerve fiber layer with development of oval holes which may coalesce leaving the retinal blood vessels floating in the vitreous (vitreous veils).

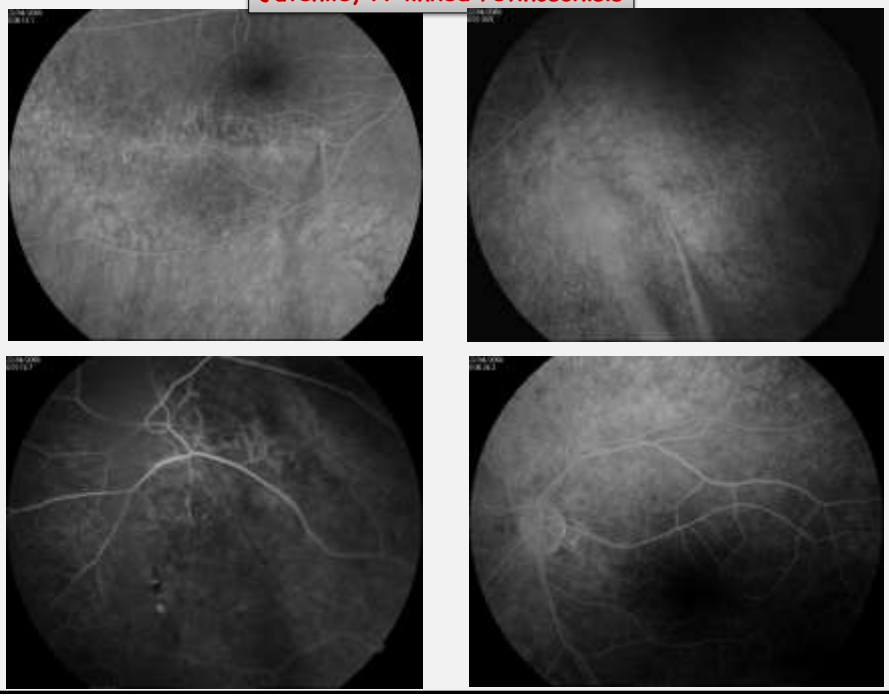
- peripheral silver dendritic figures, vascular sheathing and pigmentary changes are common.

Investigations: OCT, ERG, EOG & FFA

Juvenile, X-linked retinoschisis



Juvenile, X-linked retinoschisis



Familial exudative vitreo-retinopathy

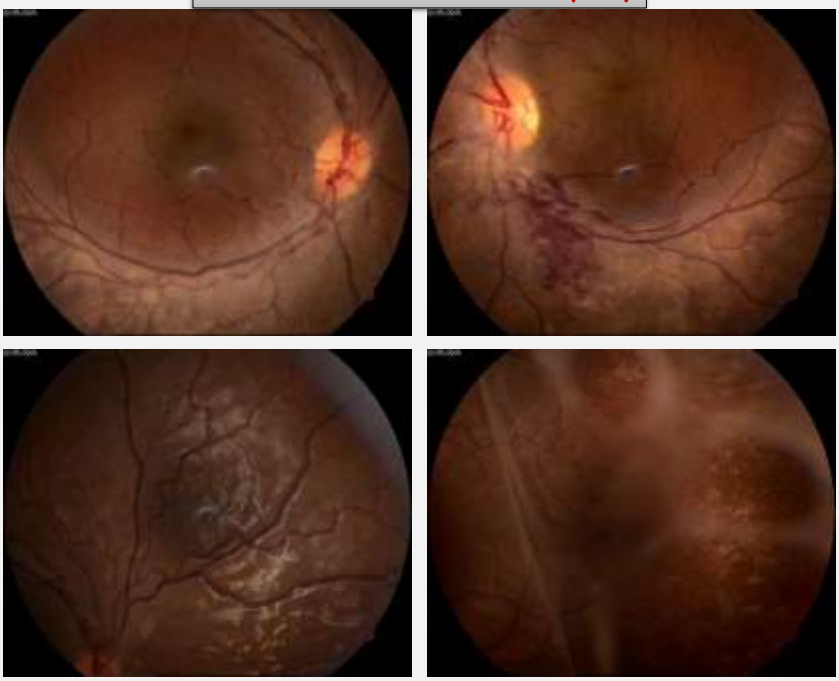
Presentation: in late childhood, due to failure of vascularization of the temporal retinal periphery.

Signs:

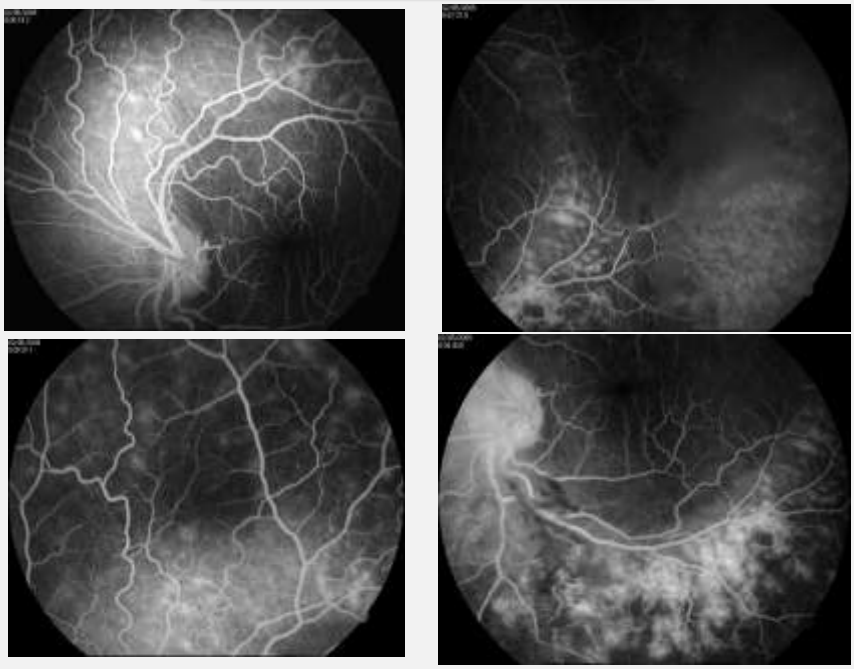
- Vitreous degeneration, peripheral vitreoretinal attachment &areas of white without pressure.
- Abrupt cessation of retinal blood vessels at temporal equator.
- Peripheral vascular tortuousity, telangiectasia and neovascularization.
- Fibrovascular proliferation & vitreo-retinal traction.
- Vascular straightening and temporal dragging on disc and macula.
- Yellowish retinal deposits and vascular leakage.

Investigations: FFA

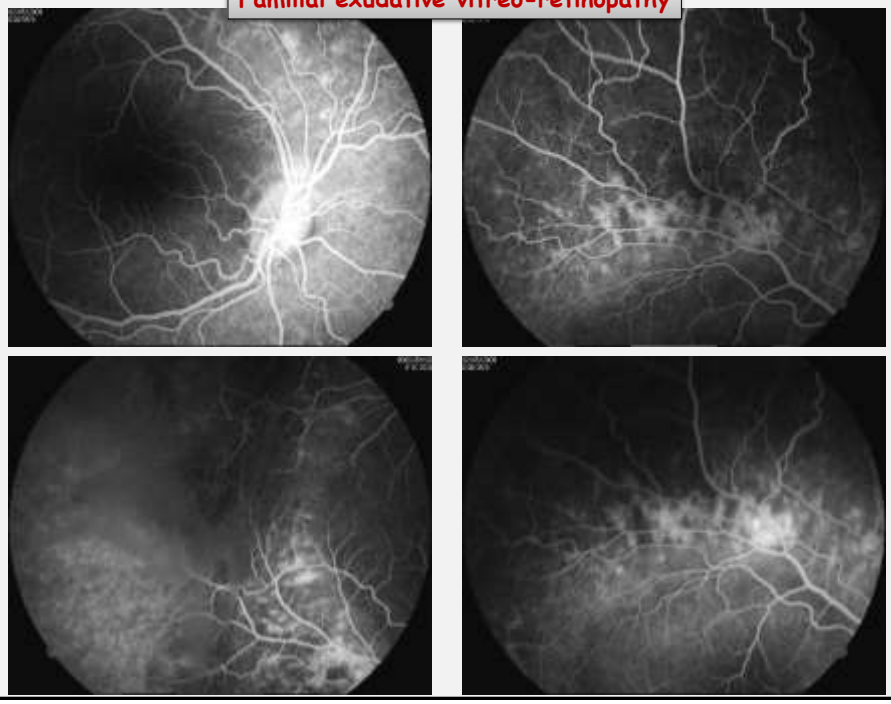
Familial exudative vitreo-retinopathy



Familial exudative vitreo-retinopathy



Familial exudative vitreo-retinopathy



Wagner syndrome

•**Presentation:** early life with pseudostrabismus due to congenital temporal displacement of the fovea & nyctalopia.

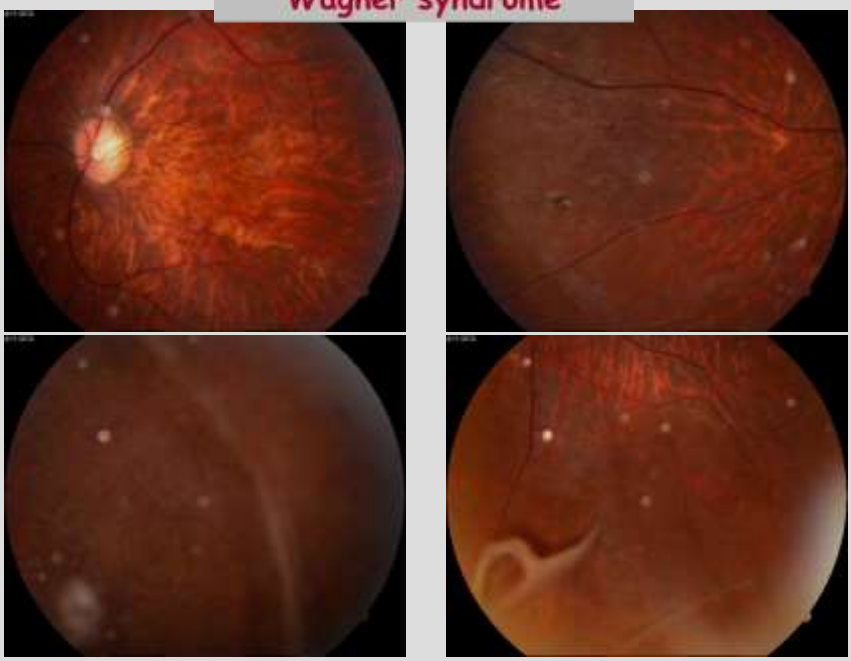
•**Signs:**

- Low myopia.
- Vitreous degeneration with completely absent normal scaffolding.
- Avascular greyish white pre-retinal membranes extending from the posterior pole to retinal periphery.
- Progressive chorioretinal atrophy.

•**Investigations**: FFA & ERG

Wagner syndrome

Wagner syndrome



Generalized choroidal dystrophies

They include the following:

- **Choroideremia.**
- **Gyrate atrophy.**
- **Generalized choroidal dystrophy.**
- **Progressive bifocal chorioretinal atrophy.**

Choroideremia

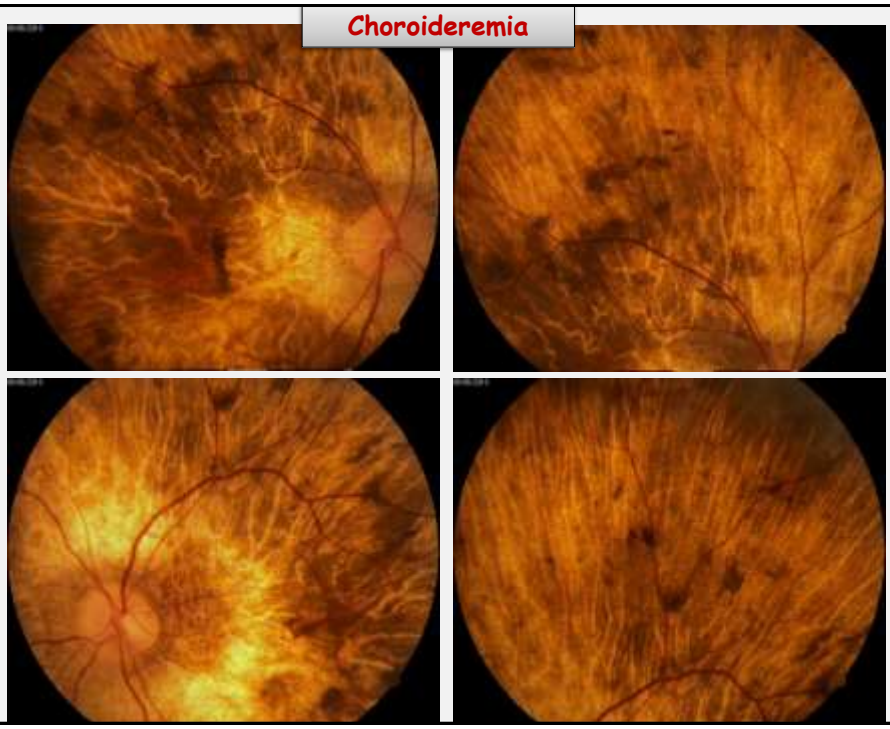
Presentation: in second to third decades with nyctalopia due to progressive diffuse atrophy of choroid, RPE & photoreceptors, followed some years later by loss of peripheral vision.

Signs:

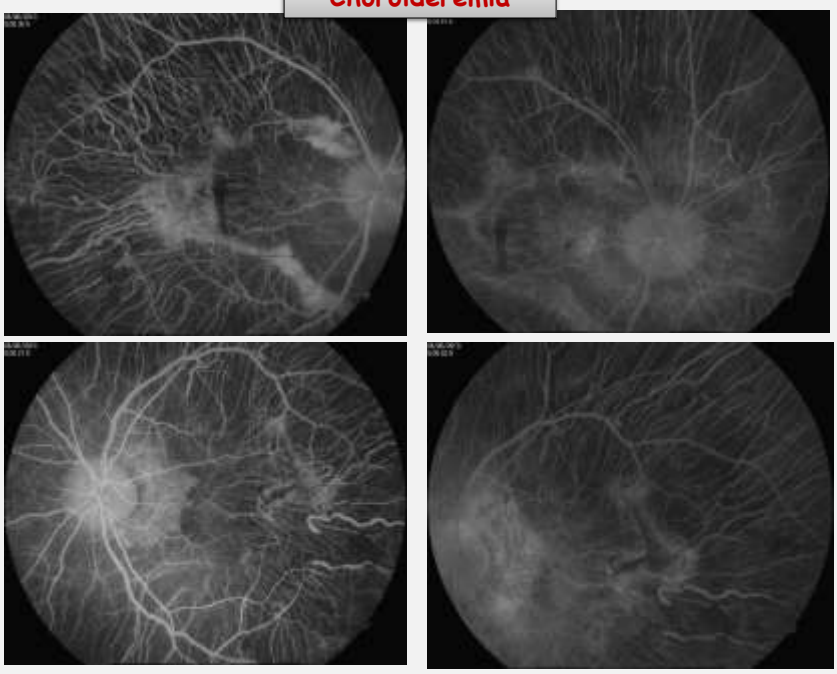
- Mild peripheral RPE abnormalities.
- Atrophy of RPE and choroid spreads peripherally and centrally.
- End-stage disease shows few choroidal vessels coursing over bare white sclera, vascular attenuation, and optic atrophy.
- The fovea is spared until late in the disease.

Investigations: FFA & ERG

Choroideremia



Choroideremia



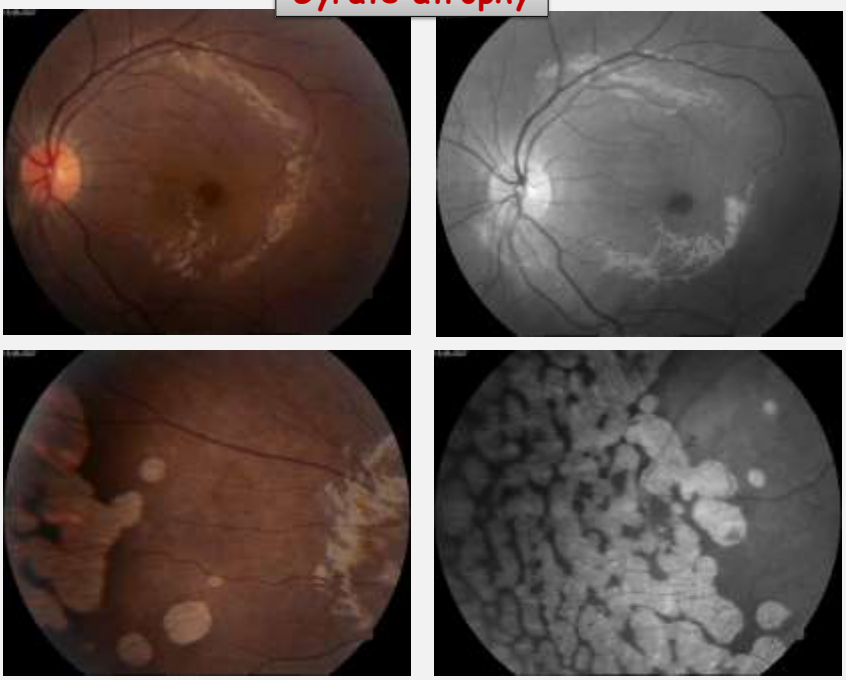
Gyrate atrophy

Presentation: in the first to second decade with myopia & nyctalopia.

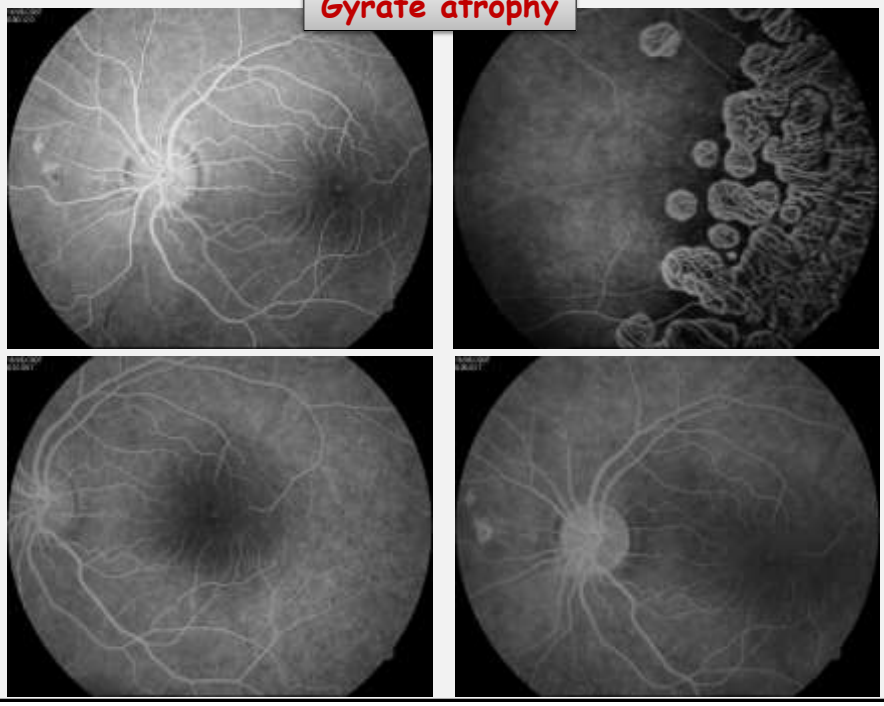
Signs:

- Mid-peripheral depigmented spots associated with pigmentary mottling.
- Sharply-demarcated, oval or rounded areas of chorio-retinal atrophy which may be associated with numerous glistening crystals at the posterior pole.
- Coalescence of the atrophic areas with progressive peripheral and central spread.
- Extreme attenuation of retinal blood vessels.
- The fovea is spared till late

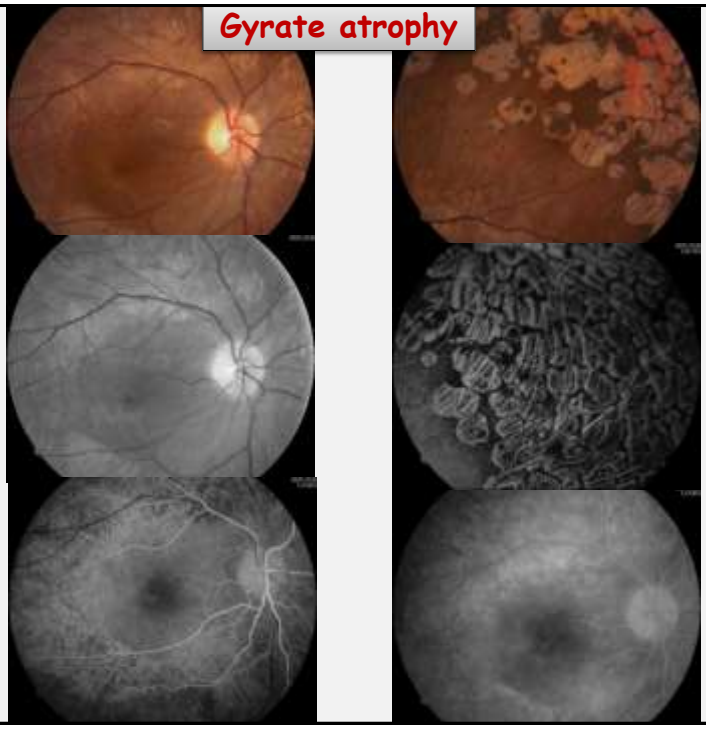
Gyrate atrophy



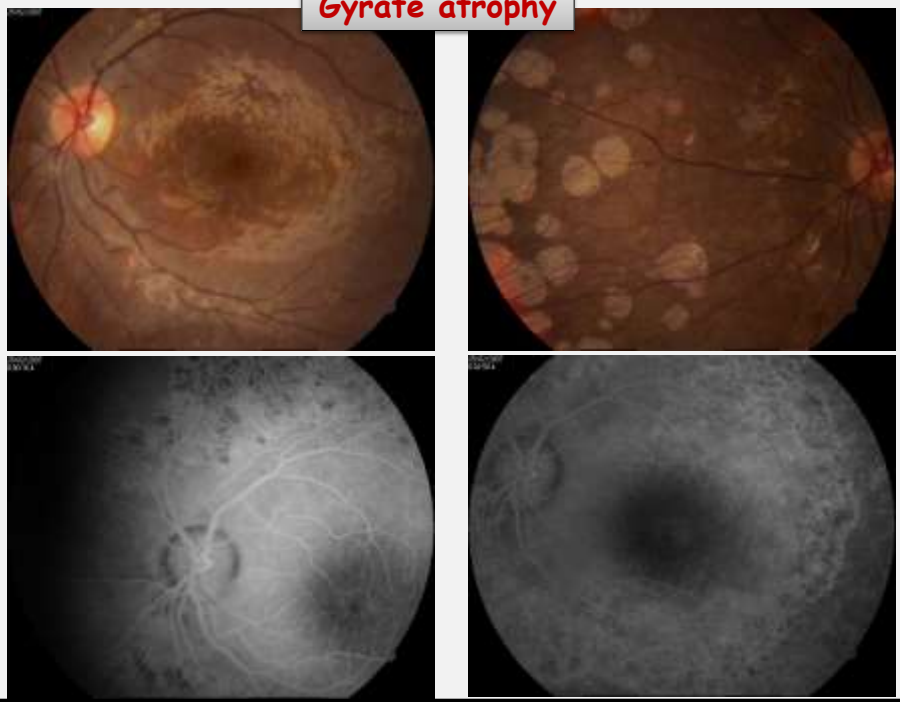
Gyrate atrophy



Gyrate atrophy



Gyrate atrophy



Albinism

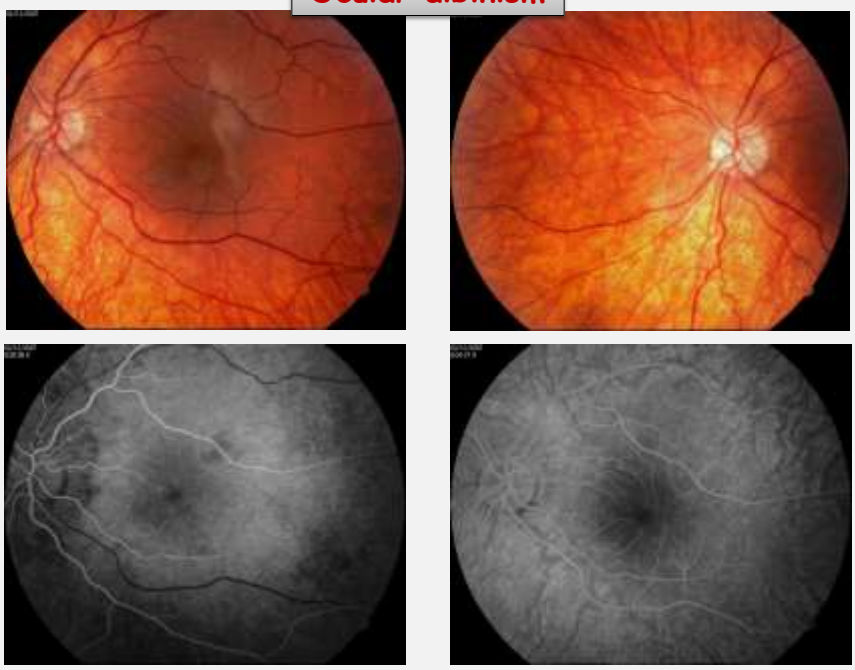
They include the following:

- Tyrosinase-negative oculo-cutanoeus albinism.
- Tyrosinase-positive oculo-cutanoeus albinism.
- Ocular albinism.

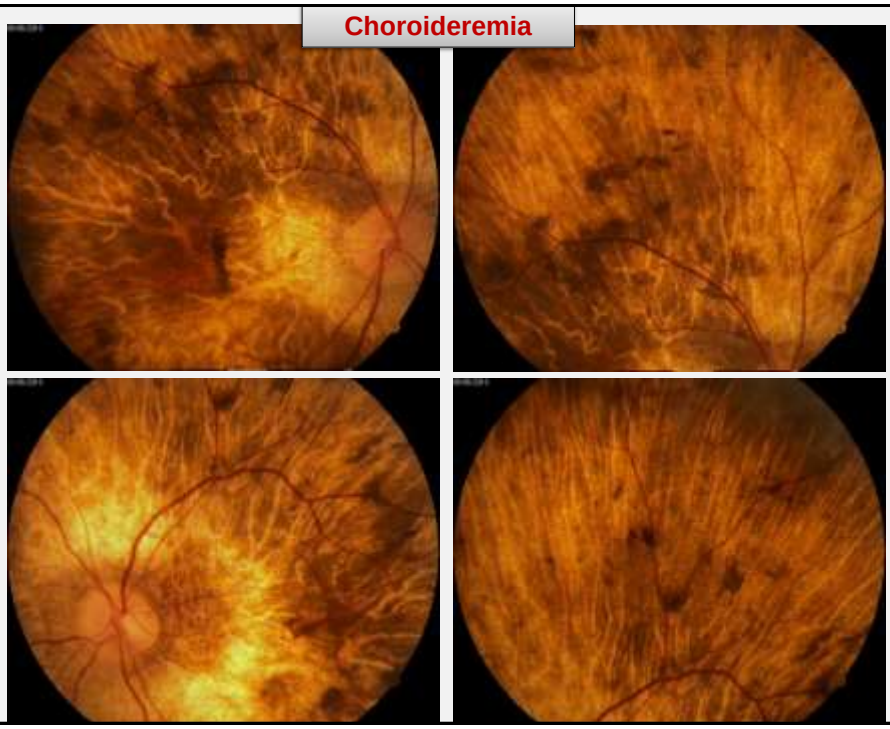
Ocular Albinism

- Presentation is predominantly ocular with normal skin and hair, although in some cases hypopigmented skin macules are evident.
- The fundus appears hypopigmented with enhanced visualization of choroidal veins and sclera.

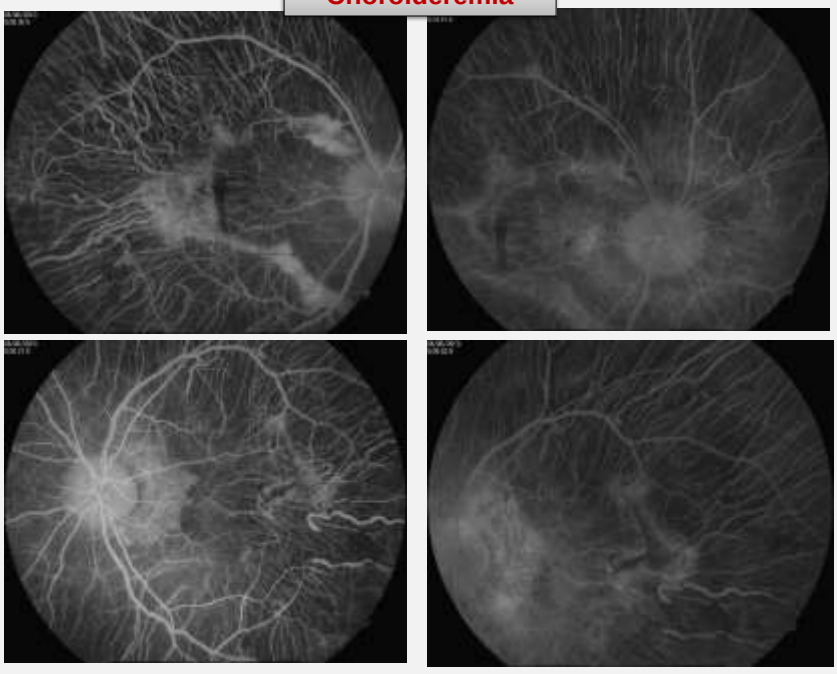
Ocular albinism



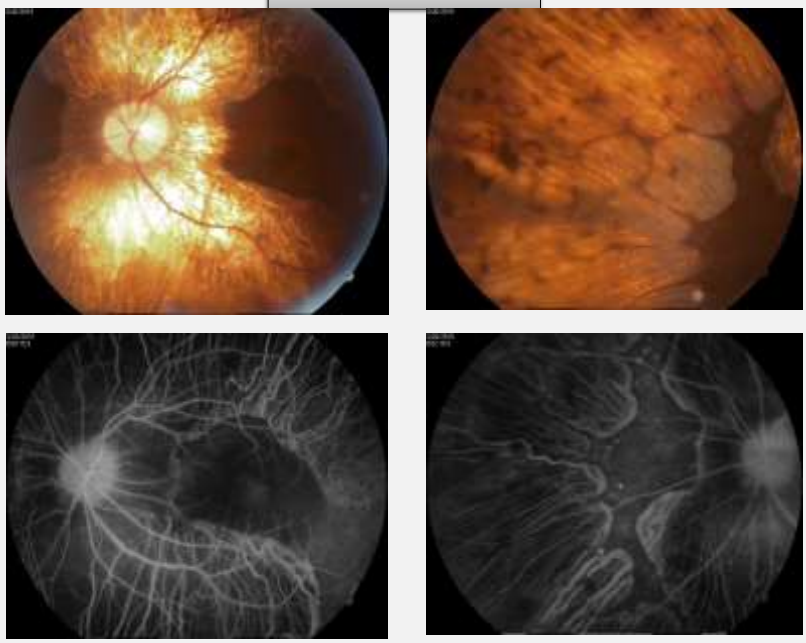
Choroideremia



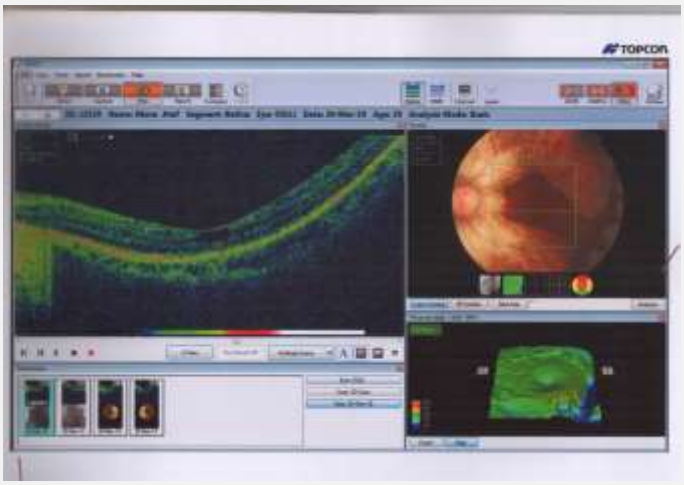
Choroideremia

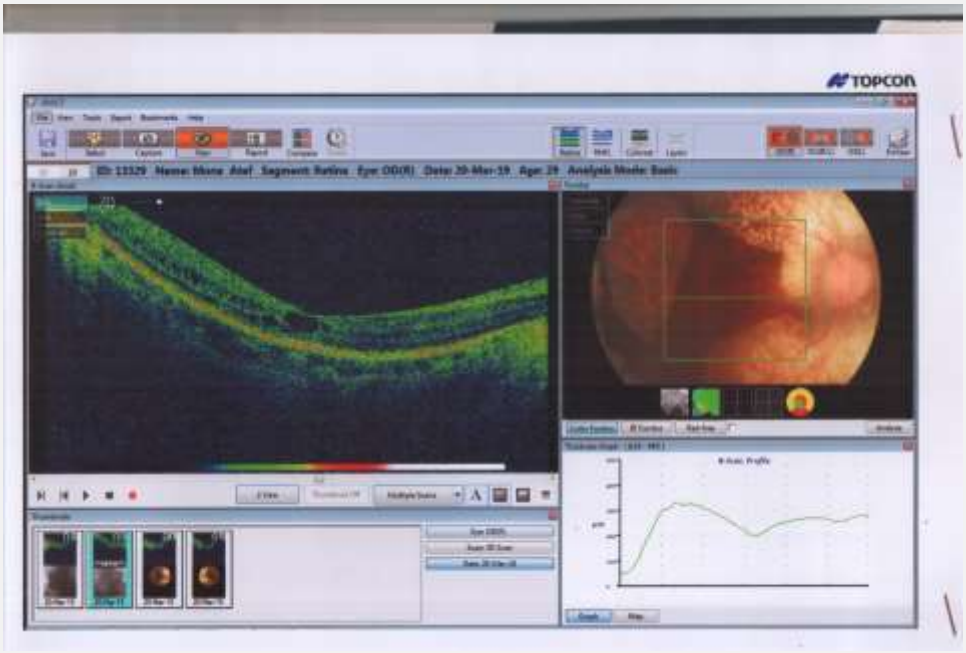


Choroideremia



Choroideremia





Cherry red spot at the macula

They include the following:

- GM1 gangliosidosis.
- Mucolipidosis typee-1.
- GM2 gangliosidosis.
- Niemann-Pick disease.
- Farber disease.

Thank you