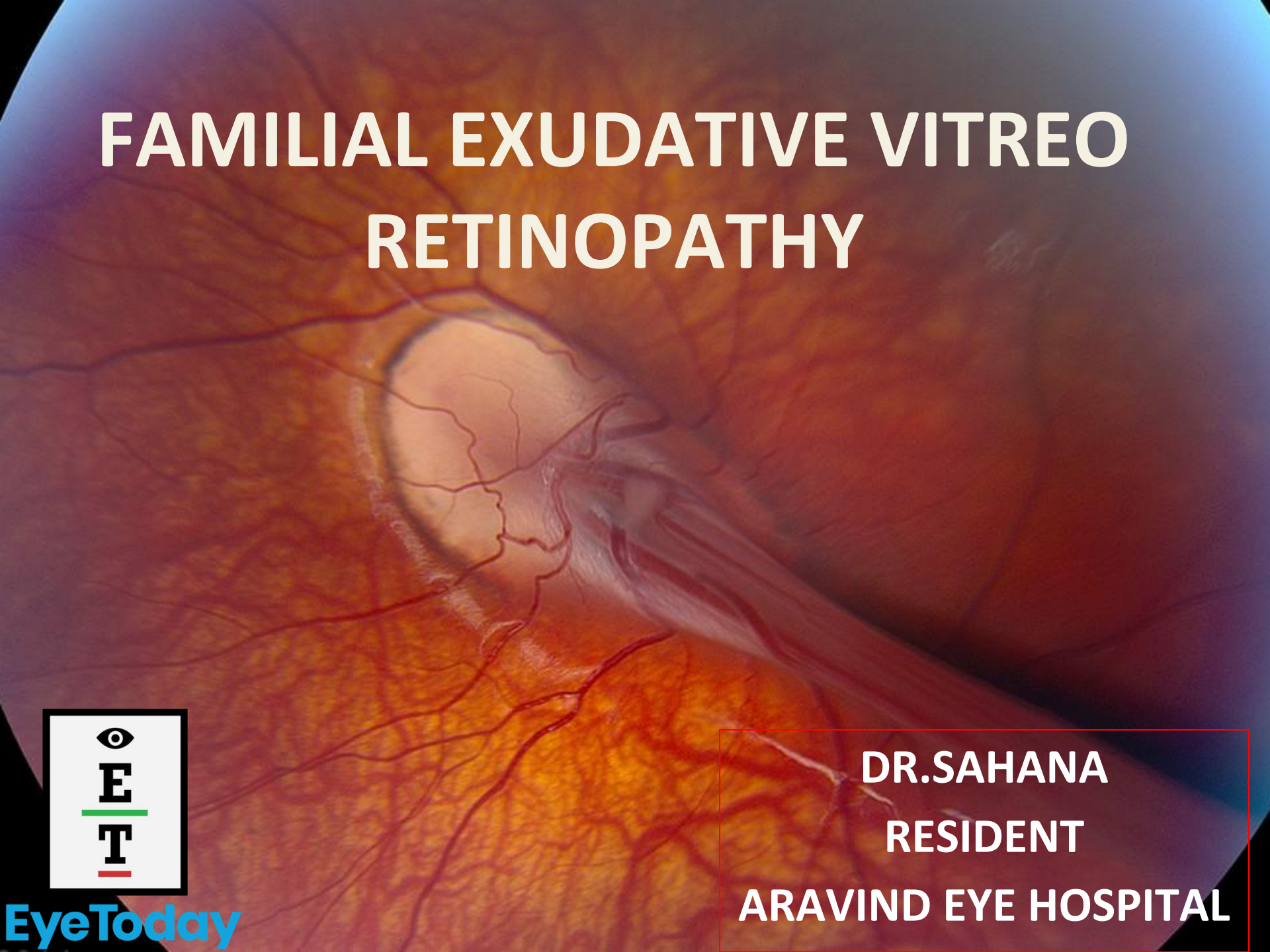
FAMILIAL EXUDATIVE VITREO RETINOPATHY



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Familial exudative vitreoretinopathy (FEVR) is a hereditary retinal vascular disorder first described by **Criswick** and **Schepens** in 1969.

INHERITANCE:

- Bilateral , mostly autosomal dominant, rarely autosomal recessive/ X-linked recessive with high penetrance and variable expressivity.

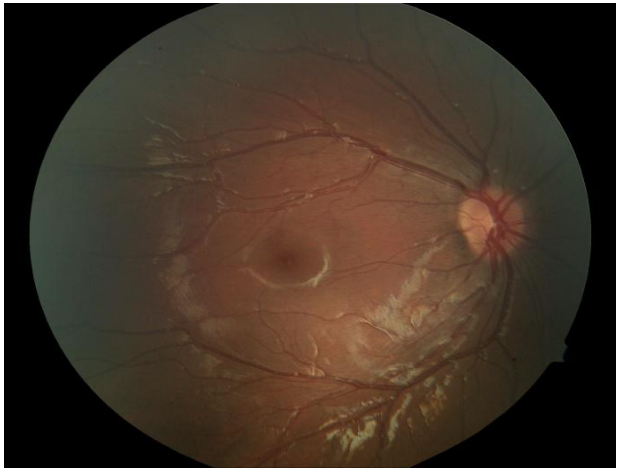
- Six genes so far have been implicated in FEVR: **FZD4, NDP, LRP5, TSPAN12, KIF11and ZN408.**

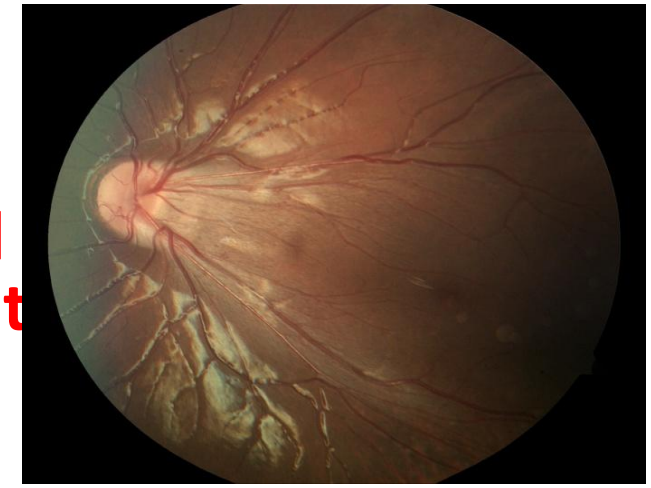
PATHOGENESIS

- Consequence of disturbed development of retinal vasculature in the last months of gestation
- **Failure of the peripheral retina to vascularize**
- The avascular zone in FEVR remains a permanent feature throughout life
- Persistence of the avascular zone maintains the stimulus for the development of peripheral neovascularisation after neonatal period.
- Macular traction or retinal detachment occurs with contraction of mesenchymal elements at the avascular border.
- Fibrous proliferation may be the result of chronic peripheral vascular leakage

CLINICAL FEATURES

- Decreased visual acuity.
- **Fibrovascular mass** with prominent arterial and venous feeders, retinal exudates encompass the ciliary body and peripheral lens capsule.
- **Dragging of the macula, disc** and retinal vessels may occur.
- Tractional forces causes retinal folds extending from temporal quadrant through the macula causing **tractional or rhegmatogeous retinal detachment**
- Pseudoexotropia,myopia,peripheral cystoid degenration, peripheral vitreous snowflakes.





STAGES

STAGE 1: Displays white without pressure associated with vitreous bands, peripheral cystoid degenration and peripheral vascular anomalies

STAGE 2: Demonstrates dilated tortuous vessels,fibrovascular mass in temporal periphery and dragging of disc and macula

STAGE 3: Extensive retinal detachment with vitreous membranes, massive subretinal exudation, cataract,band keratopathy, posterior synechia, glaucoma

In 1996, **Pendergast and Trese** proposed a clinical classification of FEVR based on ophthalmoscopic findings

STAGE	CLINICAL FEATURES
1	Avascular retinal periphery
2	Retinal neovascularization
2A	Without exudate
2B	With exudate
3	Extramacular retinal detachment
3A	Without exudate
3B	With exudate
4	Macula-involving retinaldetachment, subtotal
4A	Without exudate
4B	With exudate
5	Total retinal detachment

CLASSIFICATION

Miyakubo and Hashimoto classified FEVR according to its angiographic appearance

TYPE 1: Avascular zone less than 2DD in width from ora serrata, focal AV shunts, absent neovascularisation.

TYPE 2: Avascular zone greater then 2DD and more AV shunts developed

TYPE 3: V shaped notch in avascular zone between superior and inferior temporal arcades

TYPE 4: Incorporates neovascularisation including seafans.

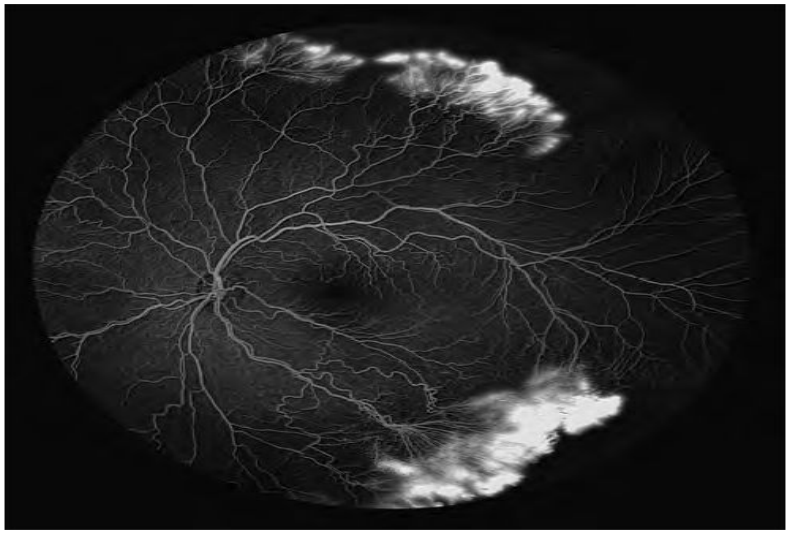
TYPE 5: Denotes cicatricial disease

HISTOPATHOLOGY

- Thickened retina containing **dilated , telangiectatic blood vessels.**
- Thickened blood vessel walls due to perivascular infiltrate
- Cellular and acellular vitreous membranes gets attached to internal limiting membrane throwing retina into folds
- Intraretinal and subretinal inflammation

INVESTIGATION

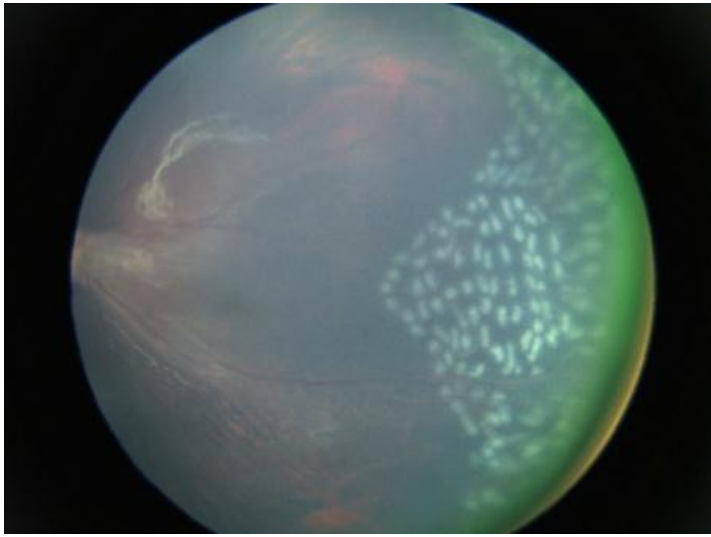
- **Fundus Fluorescein angiography** is the investigation of choice , shows avascular temporal retina , vascular straightening and abrupt termination with leaking capillaries.



- Genetic testing (*FZD4, NDP, TSPAN12, and LRP5*), this confirms the diagnosis in approximately half of patients with FEVR.

TREATMENT

- Ablation of the avascular retina by photocoagulation.
- Anti-VEGF has been tried in cases of FEVR with neovascularisation.
- Vitrectomy in case of retinal detachment .
- Lifelong screening is mandatory



PROGNOSIS

- Majority of carriers do not suffer from visual impairment.
- Patients who were diagnosed before the age of 3 have a more severe course with a very poor visual prognosis
- Visual loss after second or third decade is rare and related to the development of rhegmatogenous retinal detachment.