

# ***Retinal Dysplasia***

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***What is meant by retinal  
Dysplasia?***

*It is the presence of primitive  
retinal structure*

***“Pseudo-retina”***

## Definitions

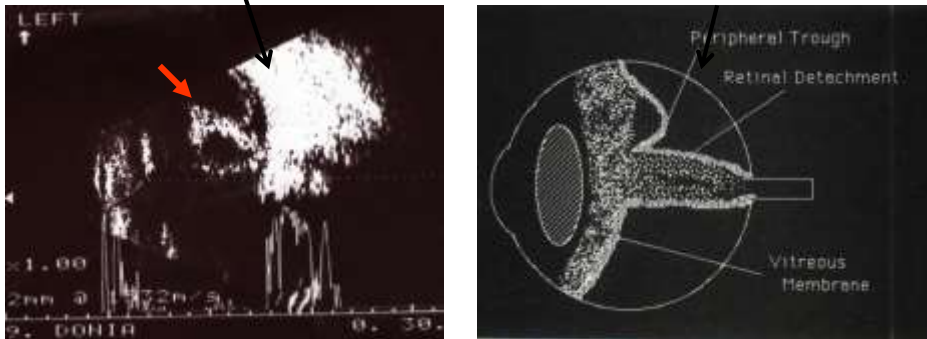
- Dysplasia is irregular *arrangement* and loss of *maturation* of cells in an epithelium
- Dysplasia is abnormal growth and *differentiation* of embryonic retina
- Structural deviations of the retina is related to lack of normal histogenesis of the *pigment epithelium*
- Impaired *genetic* mechanisms contribute

## Retinal Dysplasia

Early described in Norrie's disease

- Norrie's disease:
  - X-linked recessive syndrome characterized by congenital oculo-acoustic-cerebral degeneration
  - Bilateral white retrolental vascularized masses are noted after birth.
  - The pathologic event occurs in the retina in 3rd gestational month
  - Clinically Retinal Dysplasia could be described as retrolental fibrous vascular mass incorporating primitive retinal structure

## Typical US findings as described in Norrie's disease



- High amplitude echoes vitreous floaters with  
**Total closed funnel-like RD**
- Peripheral retinal loop (**Trough**)

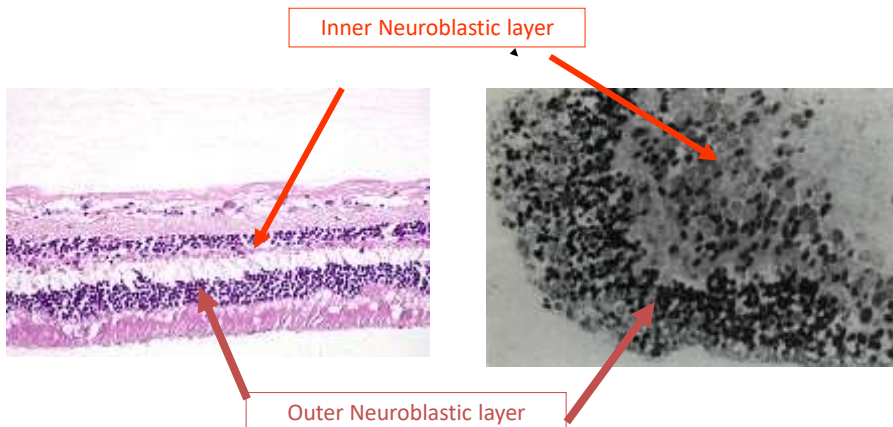
## Ultrastructural Description

- The **Retrolental membrane** is composed of layered collagenous tissue with structures resembling blood vessels
- **Retinal tissue** shows avascular inner & outer neuroblastic layers
- An **epiretinal mass** composed of portions of retina & cortical vitreous with primitive vascular structures

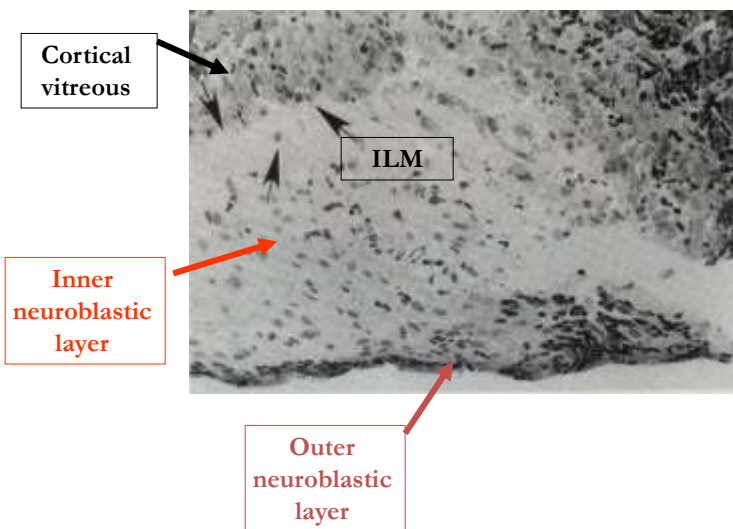
# Histopathology

Histology of Normal retina

Histology of retinal dysplasia



## Histology of Preretinal Vascular Mass and Underlying Retina



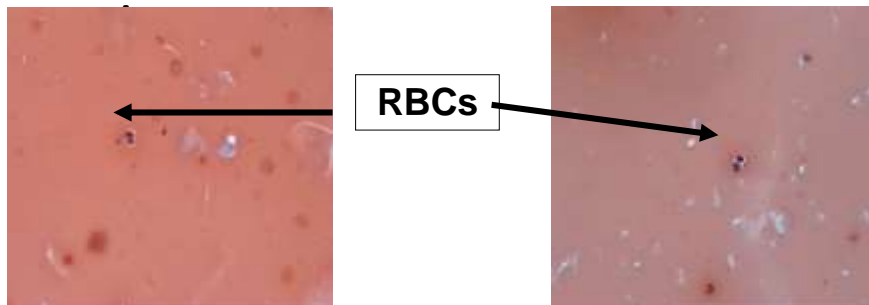
## Clinical Presentations

- Newborn
- Blind
- Anterior segment Dysgenesis
- Leukocoria



## Findings during Surgery

- Retina could not be identified
- Clumped connective tissue in the vitreous
- Blood tinged subretinal fluid & vitreous



## Associated Ocular Malformations

- Microphthalmia
- Congenital glaucoma
- Anterior segment dysgenesis :
  - **Corneal opacity &/or Iris hypoplasia**
- Fellow eye in unilateral cases may show :
  - **Optic nerve hypoplasia**
  - **Salt & pepper fundus**

## Ultrasonographic Features



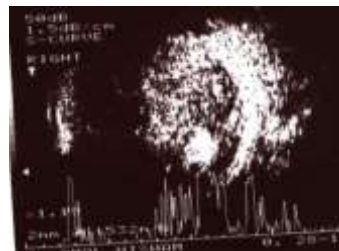
- Posterior vitreous opacities of indeterminate configuration
- Highly echogenic Retrolental membrane
- Anterior retinal loops
- Closed funnel-like RD
- Small areas of calcification may be detected but not as extensive as in retinoblastoma

## Differential Diagnosis

**PHPV**  
(Microphthalmia)



**Retinoblastoma**  
(Calcification)



***CT & MRI may not be conclusive in differentiation***

## In Conclusion

- Retinal Dysplasia can be confused with **PHPV** and **Retinoblastoma**
- Missing the diagnosis is usually due to wrong interpretation of **Ultrasonography**
- There is **NO** treatment for Retinal Dysplasia

# Thank You

