RETINITIS PIGMENTOSA

Pranesh

Abiotrophy of Neuro.Epithelium = Photoreceptors.

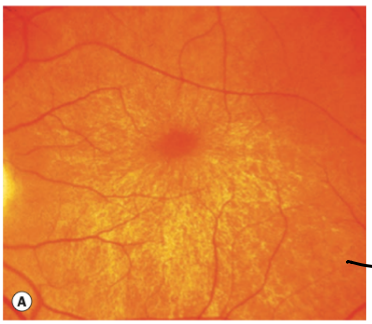
- Most common Hereditary Fundus Dystrophy. / M.C. Inherited disease of Retina.
- Diverse group of Inherited Disorders

↓

Progressive Diffuse Retinal degenerative diseases affecting 1st - Rods --> then, Cones

- Modes of Inheritance

- Sporadic = Simplex / Isolated RP ↓ Most common (50%)
- Inherited
  - AD ↓ Best Prognosis (25%)
  - AR (25%)
  - XLR - (10% ↓ least comm, most severe, Worst prognosis



♀ carriers of XLR ⇓ TAPETAL REFLEX of Macula (Shiny reflection on macula)

∴ Pathophysio : B/L Disease

Genetic mutations of
Proteins involved in Photo-transduction cascade

eg. Rhodopsin gene mutation - 1st major RP gene.

↓

Rod cell Death by Apoptosis

1st Hallmark symptom → Defective Night vision [= Nyctalopia] & Defective Dark Adaptation

2nd Hallmark symptom → Mid-peripheral Visual field Loss ↓ Progressing to Tunnel Vision / Ring Scotoma.

Now, Rod cell Dysfunction / Death

↓

RPE dysfunction - Eventual Cone cell death

↓

Melanin Pigment release into retinal layers.

↓

① Jet Black spots resembling 'Bone spicules'

∴ RPE Discoloration - RPE Atrophy

↓

Choroidal vessel show = Tessellated Fundus.

- Seen in mid-periphery
- Sheathing veins > arteries

↓

Eventual Weakening of Vessels

↓

② Arteriolar Attenuation

Ganglion Cell Death

↓

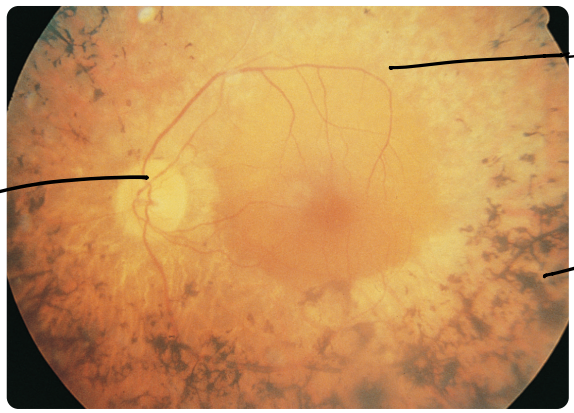
Consecutive Optic Atrophy

↓

③ Waxy pale Disc

c Gliosis.

TRIAD of RP



I) Complications

- Macula
  - Atrophy
  - Epiretinal membrane
  - Cystoid Macular Edema
- Lens
  - PSCC
  - Cortical Cataract

II) RP Associations:

- Ocular
  - Myopia
  - Microphthalmos
  - Keratoconus
  - POAG
  - PVD
  - OD Drusen
- Systemic = Syndromic RP
  - Laurence-Moon-Bardet Biedl Syndrome — Polydactyly, MR + Bull's eye Macula
  - Cockayne Synd
  - Refsum's Disease - Phytanic Acid accumul'
  - Usher's Synd — Hearing Loss
    - [M.C. Type of Syndromic RP]
  - Hallgren's
  - Abetalipoproteinemia / Bassen-kornzweig. Synd
    - Acanthocytes in blood film.
  - Kearns-Sayre Synd = Mitochondrial Inheritance + Chronic Progressive External Ophthalmoplegia

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III) Atypical RP

- Syndromic RP
- Cone-rod dystrophy
- RP sine pigmento
- Retinitis Punctata Albescens
- Leber Congenital Amaurosis

↓ Cones involved 1st → ↓ Central Vision, ↓ Color Vision

No pigments

↓ White dots on fundus (instead of black)

↓ M.C. Genetic Cause of DV in children

[Rlo Pseudo-RP conditions — eg. Syphilis, Rubella]

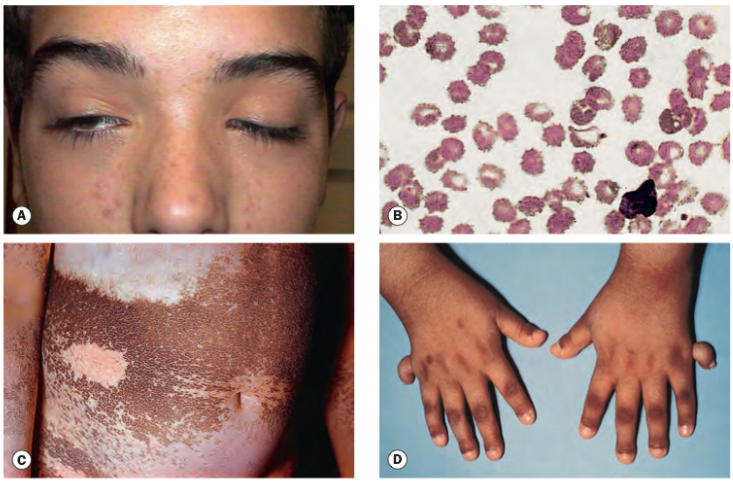
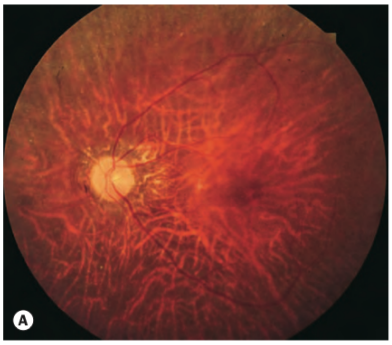
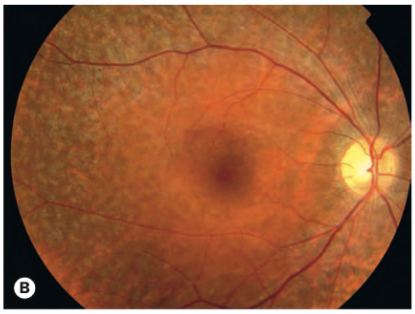


Fig. 15.13 Selected systemic associations of retinitis pigmentosa. (A) Ptosis in Kearns–Sayre syndrome; (B) acanthocytosis in Bassen–Kornzweig syndrome; (C) ichthyosis in adult Refsum disease; (D) polydactyly in Bardet–Biedl syndrome



RP sine pigmento
↳ No pigments



Refinitis Punctata Albescens
↳ White dots.

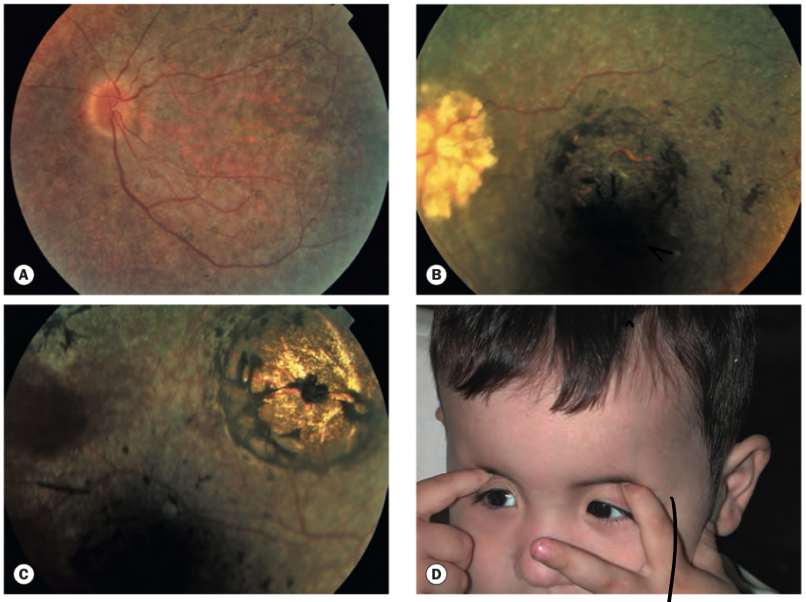


Fig. 15.15 Leber congenital amaurosis. **(A)** Mild pigmentary retinopathy; **(B)** macular pigmentation and optic disc drusen; **(C)** coloboma-like macular atrophy; **(D)** oculodigital syndrome

Oculodigital sign in Leber's Congenital Amaurosis ⇒ [Constant rubbing of eyes ↓ Orbital Fat Atrophy → Enophthalmos]

Investigations

- Full field ERG
  - 1st – ↓ed scotopic rod & combined response
  - 2nd – ↓ed photopic response
  - Finally – Extinguished ERG
- Visual field
  - 1st – mid peripheral scotoma
  - Finally – Ring scotoma / Tunnel Vision
- OCT
  - CME Edema

[RP → ↓ed a wave
CSNB → ↓ed b wave]

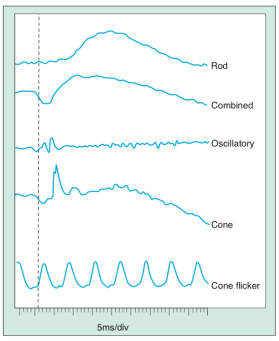
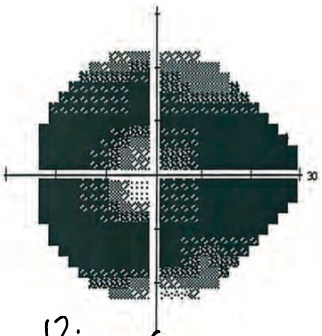


Fig. 15.11 ERG in early retinitis pigmentosa shows reduced scotopic rod and combined responses



Ring Scotoma.

Rx

① Conservative
- Low Vision Aids
- Avoid Retinotoxic Drugs
- Wear sunglasses
  ↙
  To avoid phototoxicity

② Medical
1) High dose Vit. A
- Risk of Lung Ca
- Hepatotoxic.

2) Lutein + Zeaxanthin

3) CME in RP
↓
DOC: Topical & oral CAI

③ Surgical
Cataract Extraction
↑ed risk of post-op inflammation

Recent Advances in Rx of RP

1. Gene Therapy
2. Cell Therapy = Tissue Transplantation of Retinal Progenitor cells
3. Artificial Retina. = Bionic Eye

Recent Update:

ARTIFICIAL RETINA

- epiretinal
- subretinal
- suprachoroidal.

Epi-retinal:

- ARGUS II → [2nd generation epiretinal micro-electrode array retinal prosthesis] → 1st FDA Approved Rx for RP
- EPIRET 3
- Intelligent Retinal Implant System (IRIS)

Sub retinal:

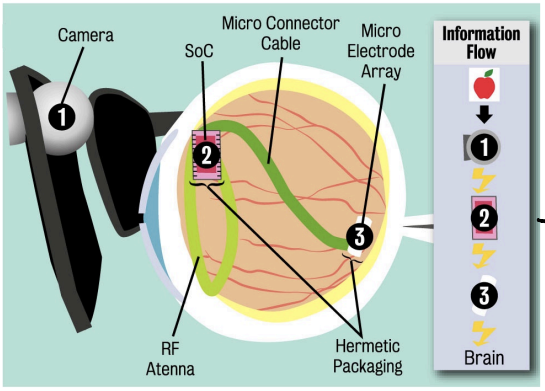
- Retina Implant Alpha-Ims
- Boston Retinal Implant

Suprachoroidal:

- Bionic Vision Australia (BVA) Retinal Prosthesis Implant

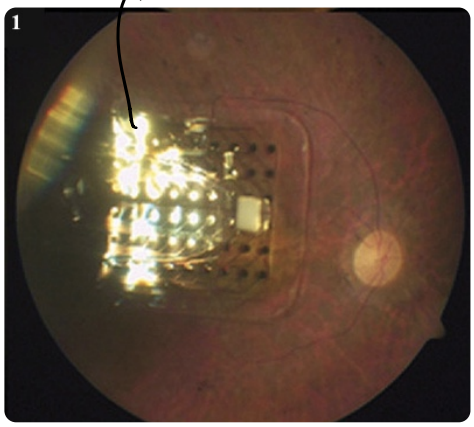
Argus II® Retinal Prosthesis System

A camera mounted on sunglasses transmits images to a chip inside the eye that shares the signals with an array of electrodes. The electrodes convey electric fields to neural impulses, which reach the brain.



SOURCE: Wentai Liu. Graphic reporting by Kanav Saraf, Bruin contributor. Graphic by Samantha Zhang, Bruin contributor.

Epiretinal Argus Implant



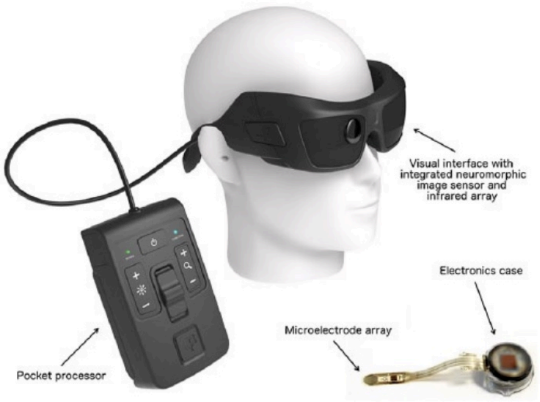


Figure 2. The Intelligent Retinal Implant System (IRIS) II. Source: Adapted from Pixium Vision S.A., Paris, France.

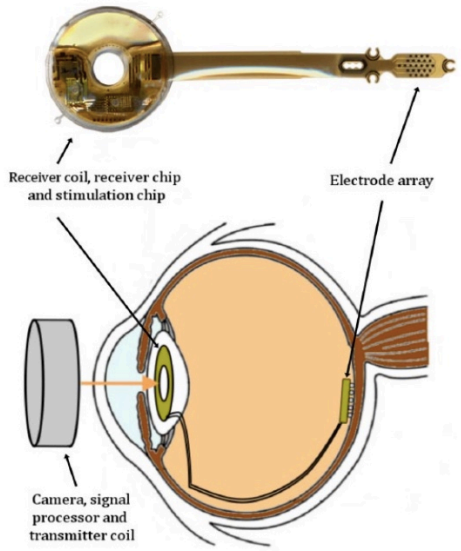


Figure 3. The EPI-RET3 Retinal Prosthesis System. Source: EPI-RET3 Team, RWTH Aachen, Germany.

entirely intraocular

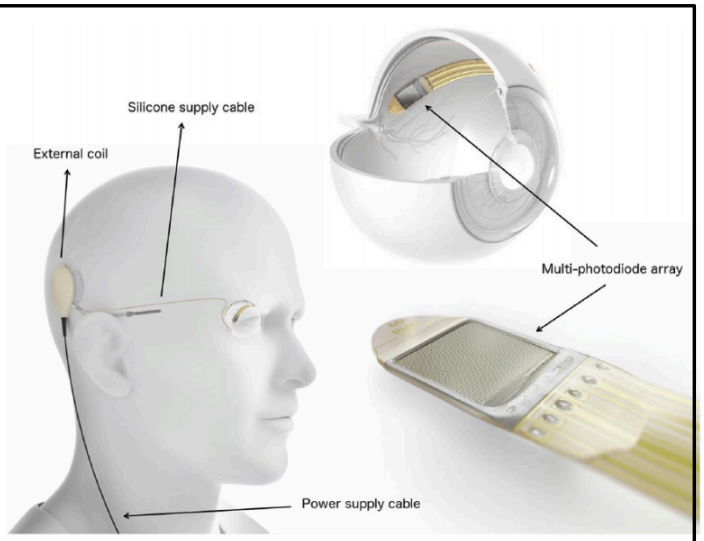
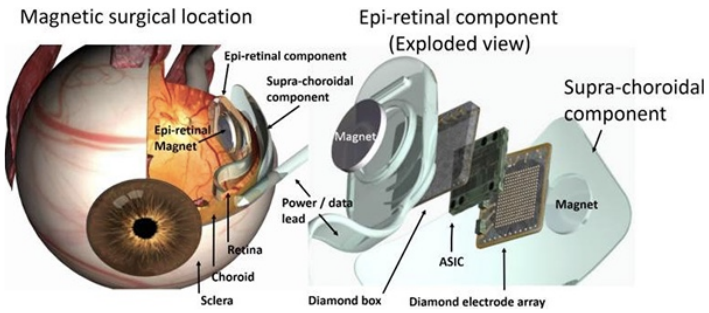


Figure 4. The Retina Implant Alpha AMS System.
Source: Adapted from Retina Implant AG, Reutlingen, Germany.



Bionic Vision Australia (BVA)
Supra. choroidal Retinal Prosthesis Implant