Translation from Romanian

Retinitis Pigmentosa

DEFINITION:
→ **Retinitis pigmentosa (RP)** is an ***inherited retinal dystrophy*** that leads to slow progressive loss of ***photoreceptors and retinal pigment epithelium***; it can cause a gradual decrease in vision over several decades.

PREVALENCE
→ RP has a **prevalence of 1 in 3000-7000 individuals.**
→ No ethnic specificity

CLINICAL ASPECTS
→ **Retinitis pigmentosa** brings together a group of genetic diseases, clinically and genetically heterogeneous, caused by progressive, bilateral and symmetrical degeneration of the retina (by abnormalities of the receptors or retinal pigment epithelium), which lead to the progressive loss of vision.
→ ***Symptomatology:*** Clinical manifestations begin in adolescence, the patient having difficulty adapting to night vision (nyctalopia), followed by progressive narrowing of the visual field
→ In syndromic forms (8 forms), RP is associated with other clinical manifestations:
- intellectual disability, metabolic disorders → **Bardet-Biedl Syndrome;**
- deafness → **Usher Syndrome** (the most frequent)

→ **The ophthalmological examination** may reveal the following:
- posterior subcapsular cataract
- changes in color perception
- cystoid macular edema
- pigment deposits in the form of spicules or Chinese letters
- attenuated retinal vascularization
- retinal atrophy
- pale, waxy appearance of the optic nerve papilla
- changes of the electroretinographic route
- OCT: loss of photoreceptors

GENETICAL ASPECTS
→ Molecular **genetic testing** using the single-gene technique, multi-gene RP panel or exon sequencing allows classification into a genetic subtype. In cases with X-linked transmission, testing of female carriers can be performed.
→ More than **3000 mutations** have been described **in more than 50 genes** involved in the production of isolated, non-syndromic RPs that may have transmission:
- **autosomal dominance (RHO, PRPH2 / RDS, PRPF31 and RP1) - 23 forms: 30-40% of RP**
- **autosomal recessive (RPE65, PDE6A, PDE6B and RP25) - 45 forms; 50-60% of RP**
- **X-linked recessive (RPGR, RP2) - 7 forms; 5-15% of RP**
- **Y-related, mitochondrial or digenic forms**

→ **The prenatal diagnosis** for high-risk pregnancies is possible by DNA analysis obtained from amniocentesis or chorionic villus sampling.

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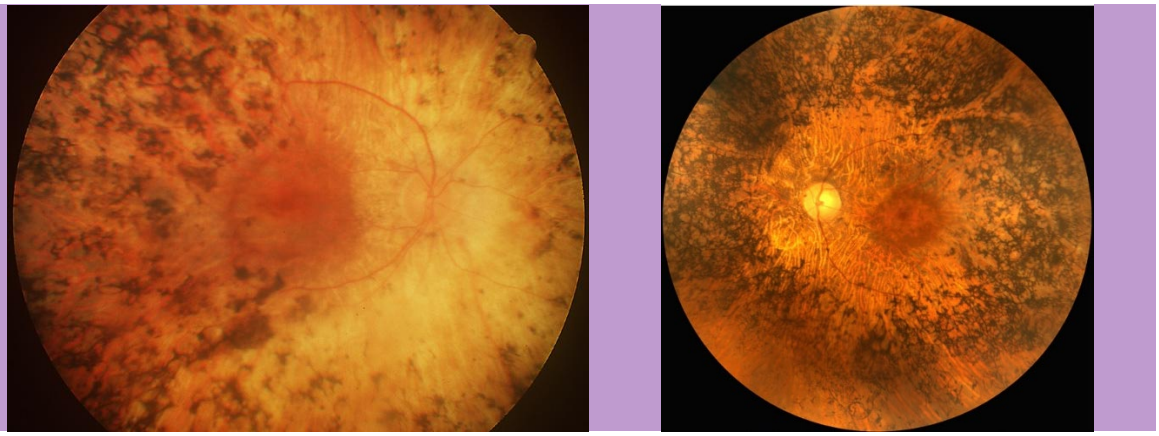
THE TREATMENT - its main purpose is to slow the progression of the disease.

- **Vitamin A palmitate and lutein-DHA** can be administered for retinal protective purposes as antioxidants.
- Oral **acetazolamide** or topical **dorzolamide** are used to reduce cystoid macular edema.
- Lens extraction is required when visual acuity is reduced due to cataracts.
- Sunglasses improve visual performance.
- Optical magnifiers can be recommended - in the more advanced stages.

PROGNOSIS: Severity is partially correlated with the mode of transmission:

- cases with X-linked transmission → the most severe prognosis,
- autosomal recessive and spontaneous cases → intermediate severity
- autosomal dominant cases → the most favorable prognosis.



Img. 1 Ophthalmoscopic lesions characteristic of RP (pigment deposits in the form of spicules or Chinese letters)

Source: https://emedicine.medscape.com/ ; http://retina.umh.es/webvision/imageswv/DONFig12b.jpg

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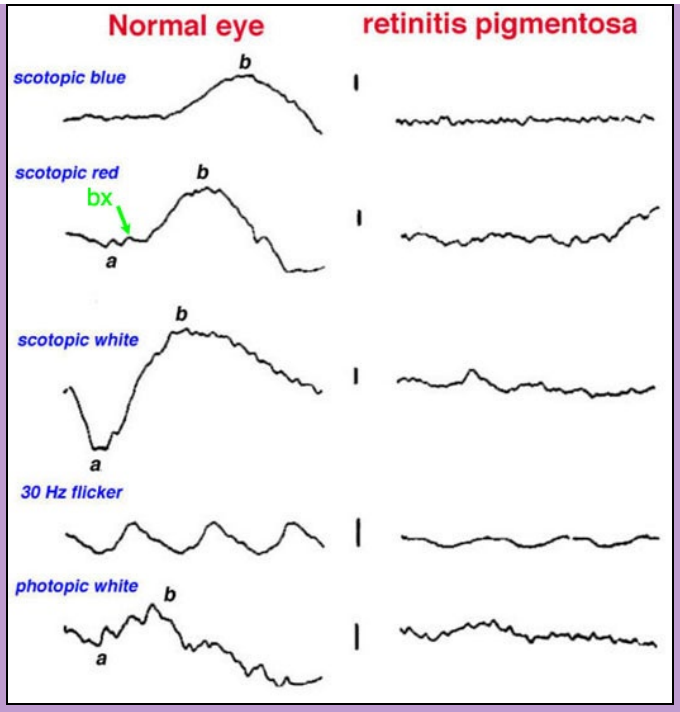
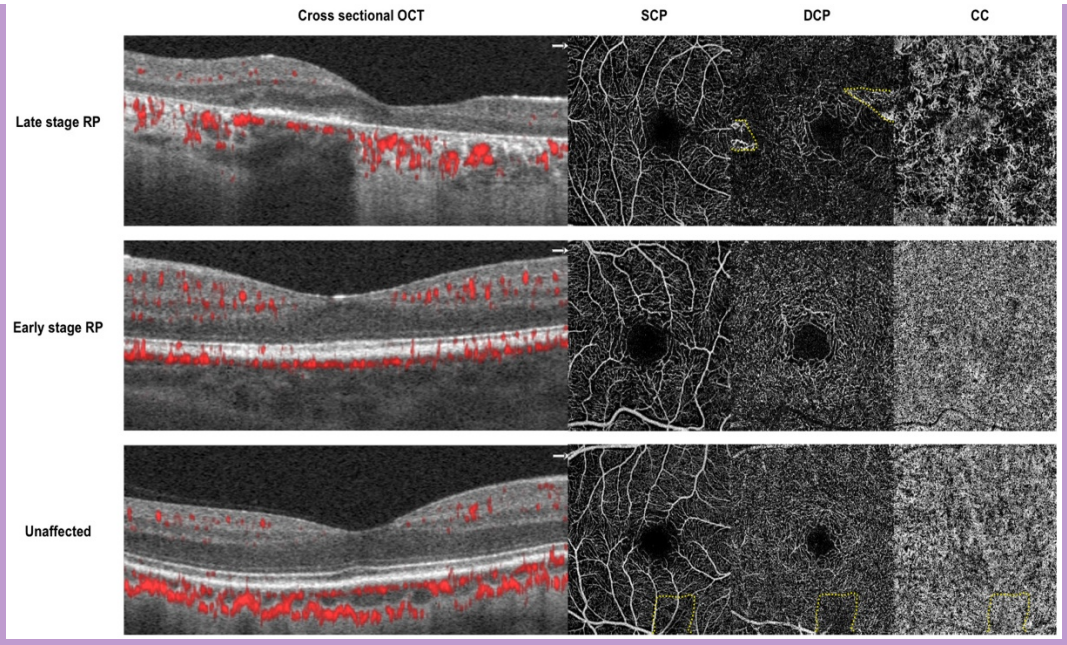


👁 Img. 2: Comparative ERG pathway between a normal patient and a patient diagnosed with retinitis pigmentosa

Source: http://retina.umh.es/webvision/imageswv/DONFig13.jpg



👁 Img.3 : OCT and OCT-A comparison between normal retina and RP in the early and late stage

Source: https://www.mdpi.com/2077-0383/8/12/2078/htm

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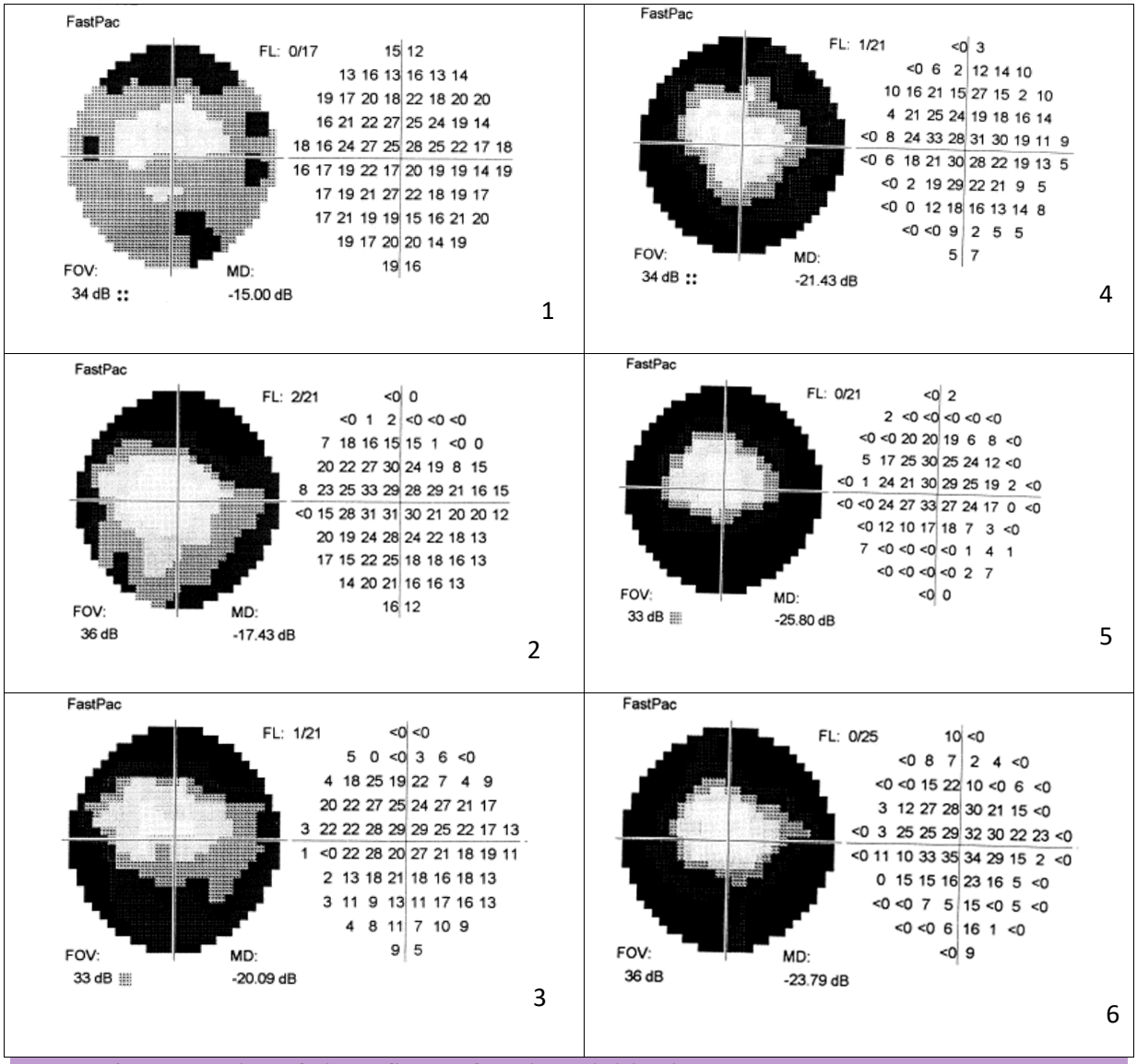

Img. 4: Progression of visual field defects in retinitis pigmentosa

Source: https://www.sciencedirect.com/science/article/pii/S0002939498004085

Text source : ***https://www.orpha.net/consor/cgi-bin/Disease.php?lng=EN***

I, BURA TEODORA - NATALIA, certified interpreter and translator for English, by virtue of the authorization no. 35530/20.03.2013, issued by the Ministry of Justice of Romania, hereby certify the accuracy of the translation from Romanian into English.

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