Translation from Romanian

CRD – Cone Rod Dystrophy

DEFINITION:

→ Rare, isolated genetic disorder of the retina, characterized by ***primary degeneration of the cones,*** with significant secondary damage to the rods, with variable ophthalmoscopic appearance. → Typical presentation includes decreased visual acuity, central scotoma, photophobia, altered color vision, followed by night blindness, and loss of peripheral visual field.

PREVALENCE is estimated at **1 /40.000** in Europe.

CLINICAL ASPECTS :

→ Clinical description: CRD is characterized by primary damage to the cones or, occasionally, by the concomitant loss of both cones and rods, which explains the predominant symptoms of the disease: decreased visual acuity, visual impairment, photoaversion and decreased sensitivity in the central visual field, followed by progressive loss of peripheral vision and night blindness.

→ The ophthalmoscopic appearance varies from normal in the early stages, with a discrete pallor of the optic nerve in the temporal sector, migration and atrophy of the macular pigment or a **"bull's eye" maculopathy**, to atrophy of the peripheral retinal pigment epithelium, intraretinal pigment migration, arteriolar attenuation and pallor of the optic disc as the disease progresses

→ They are most commonly non-syndromic, however they may also be part of several syndromes, such as ***Alström syndrome, Bardet-Biedl syndrome and spinocerebellar ataxia type 7.***

GENETICAL ASPECTS:

→ Genetics: Non-syndromic forms are genetically heterogeneous (28 genes have been identified). **Transmission patterns** depend on the gene involved and can be predominantly autosomal, autosomal recessive, or X-linked recessive. The four most common mutant genes are:

- **ABCA4 (1p22.1)** responsible for 30 - 60% of autosomal recessive CRD,
- **CRX (19q13.33)** and **GUCY2D (17p13.1)** responsible for many cases reported by autosomal dominant CRD and
- **RPGR (Xp11.4)** responsible for X-linked CRD.

DIAGNOSTIC METHODS:

→ The diagnosis of CRD is based on the clinical history, ophthalmoscopic examination, autofluorescence imaging, optical coherence tomography and complete electroretinogram.

→ Molecular diagnosis can be made for some genes.

→ The fundus examination of the eye may be normal in the early stages, with only a subtle pallor of the temporal optic disc, or may present with macular pigmentation migrations and atrophies or maculopathy with the appearance of a “bull’s eye”.

→ Late stage findings include atrophy of the peripheral retinal pigment epithelium, migration of intraretinal pigment, arteriolar attenuation, and pallor of the optic disc.

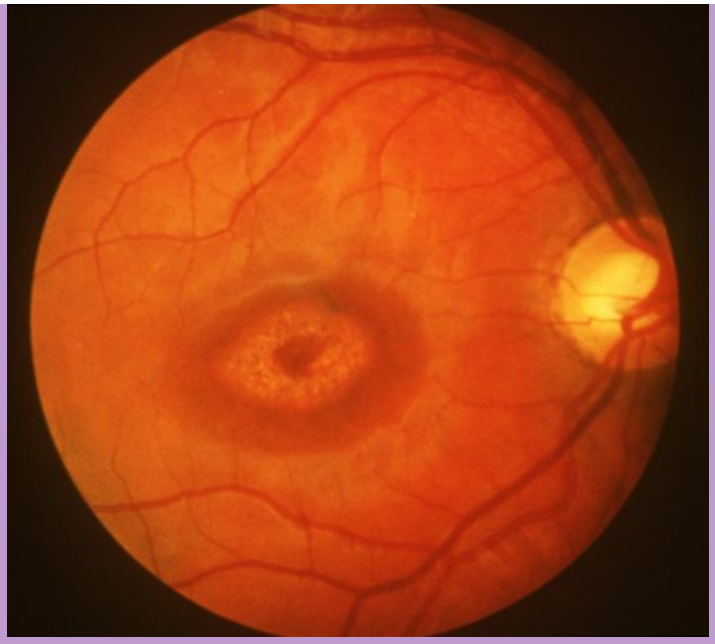
MANAGEMENT AND TREATMENT

→ Currently, there is no therapy to stop the progression of the disease or to restore the vision.

→ Management aims to slow down the degenerative process, treat complications, visual rehabilitation and help the patients to cope with the social and psychological impact of profoundly decreased visual acuity.

→ Genetic counseling is always recommended.

VISUAL FORECASTING is variable, with early central vision loss and progressive visual dysfunction that can lead to blindness before the age of 40, in most cases.



Img. 1: "Bull's eye" maculopathy

Source: https://emedicine.medscape.com

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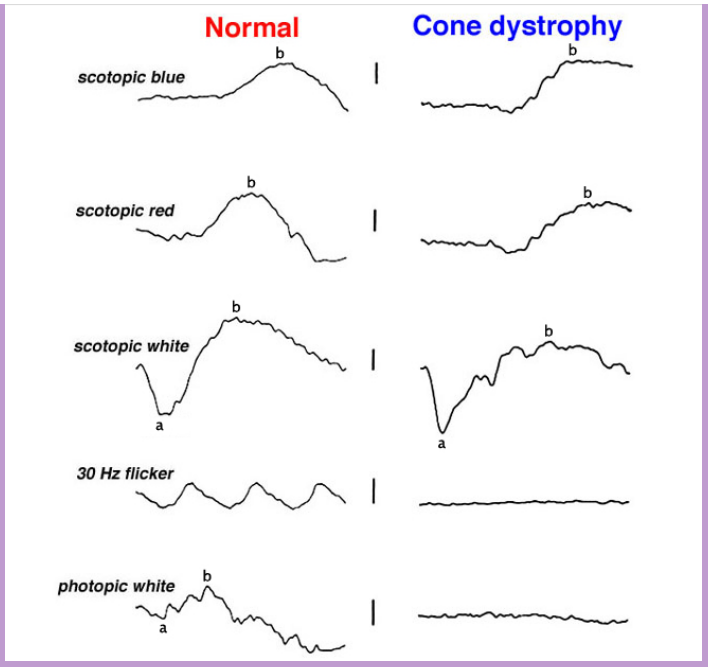
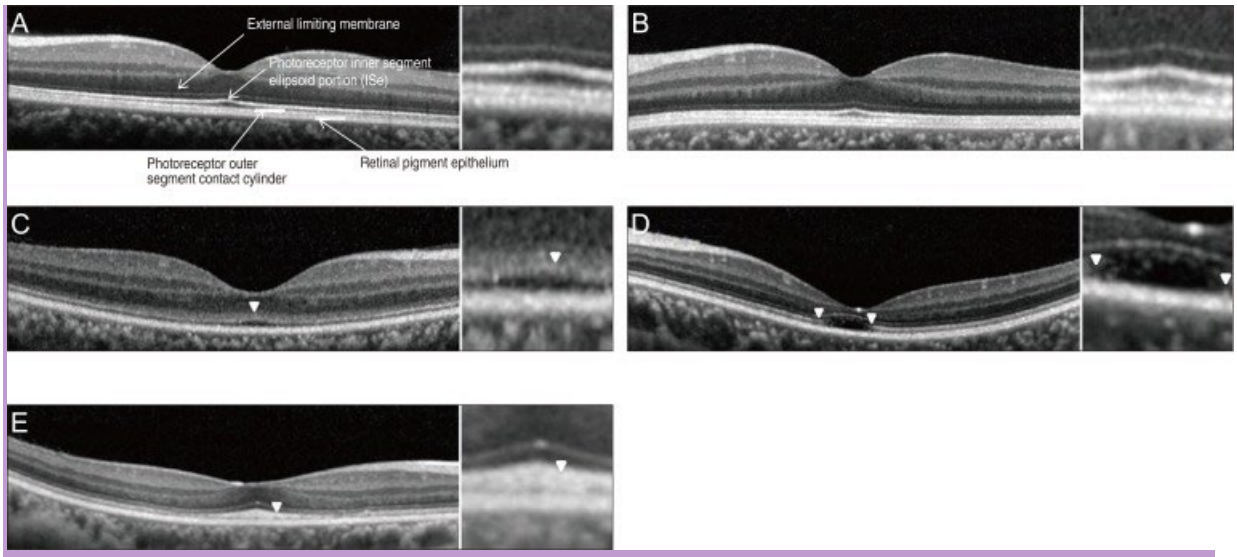


Img.2: Comparative ERG between a normal patient and one diagnosed with cone and rod dystrophy

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



Img.3: OCT aspect in the dystrophy of cones and rods: A, B: Normal aspects; C -at the foveal level, the irregular loss of the ellipsoid portion of the inner segment band (Ise) and the discrete erasure of the boundary between the ISe and the outer limiting membrane are observed. D-thinning of the central retina and segmental loss of the ISe band at the foveal level. E -central thickening of the Ise band at the level of the fovea and irregular perifoveal loss of the Ise band.

Source:
https://www.researchgate.net/publication/235393061_Morphologic_Characteristics_of_the_Outer_Retina_in_Cone_Dystrophy_on_Spectral-domain_Optical_Coherence_Tomography

Translation from Romanian

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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