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PIGMENTARY RETINOSIS: A REVIEW

Raiana Borges de Sousa

<http://lattes.cnpq.br/2200145100528619>

Benedito Antônio de Sousa

<http://lattes.cnpq.br/3433630625294888>

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Abstract: A review of the scientific literature was carried out with the descriptors “retinitis pigmentosa”. It is a set of retinal disorders with genetic factors involved and progressive evolution, whose effective treatment is still under study. **Objective:** The objective of this work is to carry out a review of the scientific literature in order to present the main clinical characteristics and epidemiological of pigmentary retinitis. **Methodology and data sources:** A search for scientific articles was carried out in the CAPES Portal journals using the descriptors “retinitis pigmentosa”, published in the last five years. 19 articles were found, of which 6 were selected for this work because they present content relevant to the proposed theme. **Discussion:** RIOS et al (2013) states that retinitis pigmentosa is a set of retinal disorders characterized by heredity, bilaterality, progressive dysfunction of photoreceptors, cell loss and retinal atrophy. QUEIROZ et al (2013) states that the genetic component can be of autosomal recessive inheritance - the most common form -, autosomal dominant or X-linked recessive. Initially there is a dysfunction of the rods and, later, there are degenerative alterations of the neurons of the inner retina and of the blood vessels, reaching the macula and the optic nerve head (BUCHAIM et al, 2013). Thus, the clinical picture of the pathology evolves according to the changes that have already occurred, with night blindness - commonly in the first two decades of life - being the initial phase, followed by progressive loss of peripheral vision and reduction of visual acuity (JOSE et al, 2015). The incidence in the world population is 1:3000 to 5000 individuals (WANG et al, 2014). This author also states that it is a disease often associated with other clinical conditions, such as Usher syndrome and Leber’s congenital amaurosis. ZHAO et al (2015) mentions that molecular diagnosis is essential for the characterization

of the disease and definition of a clinical prognosis. To date, there is no effective treatment, however, gene therapies have recently been initiated as clinical trials in humans (QUEIROZ et al, 2013). **Conclusion:** Retinitis pigmentosa corresponds to genetic eye disorders, which follow the progressive dysfunction of photoreceptors, often associated with other syndromes. Its genetic and clinical variety make molecular diagnosis important. Studies are still being carried out in search of a better understanding of the disease and its effective treatment.

REFERENCES

BUCHAIM, Guilherme; REZENDE, Marcussi Palata; MAIA, Maurício. Implante intravítreo de liberação crônica de dexametasona (Ozurdex®) para o tratamento de edema macular por retinose pigmentar: relato de caso. *Arquivos Brasileiros de Oftalmologia*, [s.l.], v. 76, n. 6, p.377-379, dez. 2013. FapUNIFESP (SciELO).

PEDROSO, José Luiz et al. Retinitis pigmentosa in pantothenate kinase-associated neurodegeneration. *Arquivos de Neuro-psiquiatria*, [s.l.], v. 72, n. 10, p.816-817, out. 2014. FapUNIFESP (SciELO).

QUEIROZ, Ana Cristina Cotta de et al. Estudo clínico e padrão de herança em pacientes com retinose pigmentar. *Revista Brasileira de Oftalmologia*, [s.l.], v. 72, n. 1, p.26-28, fev. 2013. FapUNIFESP (SciELO).

RIOS, Daniela Fernandes de Carvalho et al. Retinose pigmentar unilateral ou pseudoretinose pigmentar?: relato de caso. *Arquivos Brasileiros de Oftalmologia*, [s.l.], v. 76, n. 6, p.383-385, dez. 2013. FapUNIFESP (SciELO).

WANG, Feng et al. Next generation sequencing-based molecular diagnosis of retinitis pigmentosa: identification of a novel genotype-phenotype correlation and clinical refinements. *Human Genetics*, [s.l.], v. 133, n. 3, p.331-345, 24 out. 2013. Springer Nature.

ZHAO, Li et al. Next-generation sequencing-based molecular diagnosis of 82 retinitis pigmentosa probands from Northern Ireland. *Human Genetics*, [s.l.], v. 134, n. 2, p.217-230, 4 dez. 2014. Springer Nature.