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Hereditary Pigmentary Degeneration

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Abstract: The review article examines the genetic aspects of the development of retinitis pigmentosa, a hereditary retinal pathology that can have different types of inheritance and can cause disability and reduce the quality of life of patients. The genetic basis for the development of this group of degenerative conditions can be a number of mutations. The pathogenesis of retinitis pigmentosa is quite complex and requires detailed study, taking into account the morphology and functioning of photoreceptor cells - rods.

Keywords: ophthalmology, retinitis pigmentosa, rhodopsin, heredity.

INTRODUCTION

Retinitis pigmentosa (RP) – retinal pigmentary abiotrophy, taperetinal retinal degeneration, retinal pigmentary degeneration – is represented by a whole galaxy of heterogeneous hereditary dystrophic changes in the retina with initial damage to rod photoreceptors [2].

MATERIALS AND METHODS

Frequency RP – 1:3000 [1]. This pathology ranks first in the structure of all hereditary lesions of the retina. RP was described by A. Graefe in 1958 [3]. RP is manifested by symptoms such as nyctalopia (night blindness), photophobia, the formation of “tunnel” vision with subsequent impairment of central vision. Late stages of the disease may even be accompanied by the development of complete blindness [3]. Impaired twilight vision in this pathology cannot be treated with vitamin A [5]. The age of disease manifestation and the severity of symptoms depend on the form of RP. The onset of RP usually occurs during childhood. Damage to photoreceptors manifests itself in the form of their destruction, vacuolization of the outer sections of cells; swelling, desquamation and movement of the pigment epithelium into the inner layers of the retina occurs; The appearance of fibrosis and hyalinosis of blood vessels and glial proliferation are characteristic [6].

RESULTS AND DISCUSSION

By the age of 20, when performing ophthalmoscopy, you can most often see the full typical picture [2]: the presence of pigment deposits in the retina in the form of so-called “bone bodies”, pale waxy discs of the optic nerves, a reduction in the number and narrowing of the lumen of blood vessels [3]. Sometimes drusen are visible in the fundus - pinpoint deposits of hyaline on the vitreous plate of the choroid [4].

Changes in the macula in RP are quite common - up to 74% [5], manifesting themselves in the form of cystic degeneration with localized hyperfluorescence; dystrophy with a fenestrated bull's eye defect during fluorescein angiography. In some cases, swelling of the macular area is detected, which can be fraught with the development of central retinal breaks. The pathogenesis of macular edema is interpreted differently: on the one hand, it is explained by the reaction of the perimacular network of capillaries to toxins released during degeneration in RP, on the other

hand, the causes may be an increase in the osmotic pressure of the vitreous body and hypoxia of the retina [5]. Cystoid macular edema is found in 10-22% of patients with RP, accompanied by disruption of the blood-retinal barrier and accumulation of serous transudate in the retina. Edema and death of Müller cells can contribute to increased degenerative changes and potentiate the formation of large cysts [4].

The pathogenetic basis of RP is the loss of rods in the photoreceptor layer of the retina [4]. This process occurs from the periphery to the central region, which explains the phenomenon of the appearance of "tunnel" vision. Subsequently, damage to other photoreceptor cells—cones—occurs, and the pigment epithelium, inner nuclear layer, and ganglion cells may also be involved [5]. Damage to the cones leads to loss of central vision [2]. The reasons for the death of cones are still being considered. It is assumed that this is facilitated by the loss of their connection with the pigment epithelium, the release of endotoxins, and metabolic overload [3]. It has been shown in animals that oxidative stress and the action of reactive oxygen species play an important role in triggering cone apoptosis. Also, the death of cones is promoted by the local release of cytotoxic products after the death of rods by activated Müller cells [4]. Electroretinogram data confirms the presence of a degenerative process in the retina - a decrease in the overall retinogram or its absence is characteristic [5]. With RP, visual field impairment is manifested by the appearance of a ring-shaped scotoma, which gradually increases towards the periphery and towards the center. Central vision, especially after 30 years, can be sharply impaired due to damage to the cones [6].

The types of inheritance characteristic of RP are autosomal dominant, autosomal recessive, X-linked, and RP can also be caused by a violation of the sequences of the genetic apparatus of mitochondria. Currently, a fairly large number of mutations are known that constitute the substrate for the development of RP. For example, in the rhodopsin protein gene RHO alone, about 100 different mutations have been identified that cause autosomal dominant RP, therefore RP is characterized by high genotypic and phenotypic heterogeneity [2]. Synthesis of pathological protein when these mutations occur triggers cell apoptosis [2]. Rhodopsin, a chromoprotein, the visual pigment of rods, consists of the opsin protein and the chromophore 11-cis-retinal, located in the outer segments of the rods [3]. The gene responsible for the synthesis of rhodopsin is located on the long arm of chromosome 3 (3q21) and contains 5 exons [4]. Part of rhodopsin is immersed in the lipid bilayer of the membrane and is formed almost entirely by hydrophobic amino acids, and the other part emerges from the membrane and is located in the cytoplasm of the outer segments of the rods; it is rich in hydrophilic amino acids. According to Retina International, mutations that disrupt the structure of the hybrophobic part of rhodopsin or the region of attachment of oligosaccharides disrupt the structure and function of rhodopsin with its subsequent accumulation in the internal segments of photoreceptor cells. Mutations that damage part of the gene responsible for the synthesis of the C-terminal region of the molecule reduce transduction activity [5]. Greenberg E.R. et al (2007) discovered one of the polymorphisms in exon 4 of the RHO gene. This was a single nucleotide polymorphism G/C at position 755 (corresponding to Arg/Pro at position 252 of rhodopsin). As the authors suggest, a change in the amino acid sequence in this area can affect the addition of transducin and the launch of the enzyme cascade [3]. There are also reports that mutations in the nucleotide sequence of the human inosine 5′monophosphate dehydrogenase-1 gene, encoding a subdomain (Bateman domain), lead to the development of hereditary diseases, including RP.

Newsom D.A. (1988) proposed using three main criteria for classifying RP: by genetic type (according to the nature of inheritance), by anatomical type (typical form and a number of atypical forms) and by functional disorders: (weakly progressive; progressive, rapidly progressive). Among the forms of RP, a number of atypical variants of the disease are distinguished. Such an atypical form is, for example, pigmentary degeneration of the retina without pigment, in which there is no deposition of pigment on the fundus [2], sectoral horseshoe-shaped retinitis pigmentosa [5]. The literature contains descriptions of cases of unilateral RP. For example, the presence of unilateral PR combined with heart disease (atrial

septal defect, patent foramen ovale with shunt) and right-sided nephroptosis has been described [4]. The question of the possibility of developing unilateral RP is considered controversial, since some authors believe that in the second eye, degenerative changes in a specific period of time can occur in an unexpressed, subclinical form. To diagnose unilateral RP, it is recommended to use the criterion of the absence of typical clinical and functional changes in the other eye for at least five years. Spot white retinal degeneration is characterized by the presence in the fundus of the eye, with the exception of the macula area, of many small or larger lesions with clear boundaries. Central pigmentary degeneration of the retina is characterized by the formation of central and paracentral scotomas with accompanying dystrophic changes in the macula and the deposition of small lumps of pigment in this area.

CONCLUSION

Thus, at present, quite extensive material has been accumulated on the etiology and pathogenesis of retinal pigmentary degeneration, which is widely covered in the scientific literature. This problem is considered from the standpoint of molecular genetics, morphology and physiology of the organ of vision. However, the scientific search for new polymorphisms that may cause the development of retinitis pigmentosa continues.

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