ANIRIDIA CAUSES, SYMPTOMS AND SOME METHODS OF PREVENTION AND TREATMENT

Kodirov Muhammadumar Shakirovich

Senior teacher of the Department of Ophthalmology

Andijan State Medical Institute

Abstract: In this article, the causes of aniridia, symptoms, and some methods of prevention and treatment

Keywords: Aniridia ,general information. ophthalmoscopy, tonometry, gonioscopy, ultrasonic biomicroscopy

Aniridia is a rare, in most cases, genetically determined eye disease, which is characterized by the complete or partial absence of the iris. Clinically manifested by decreased visual acuity, horizontal nystagmus and photophobia. To diagnose the disease, it is necessary to examine the anterior eyeball, ophthalmoscopy, tonometry, gonioscopy, ultrasonic biomicroscopy, and examine clinical refraction. The etiology of aniridia can be determined by genetic testing. The specific treatment for aniridia is based on the transplantation of an artificial iris.
General information. Aniridia (irideremia) refers to a number of orphan (rare) diseases characterized by hypoplasia of the iris. The frequency of the spread of the disease averages 1:70,000. Aniridia-associated diseases include Gillespie syndrome and WAGR syndrome. Patients with irideremia are at risk of developing cataracts, glaucoma, and corneal clouding. Due to the presence of a genetic mutation in the PAX6 gene, there is a high probability of occurrence of pathologies in other organs and body systems.

Hereditary predisposition is observed in 65% of patients with aniridia, since the disease can be transmitted both in an autosomal dominant and autosomal recessive manner. In 35% of cases, the pathology is sporadic, caused by a mutation that first appeared. The disease is equally common among males and females, has no racial differences and features in geographical distribution.

Causes of aniridia. The etiological factor in the development of congenital aniridia is a mutation of the PAX6 gene, which is localized on chromosome 11. This gene encodes transcription factors that induce the development of the eyeball at the stage of embryogenesis, and also regulate the processes of differentiation of the organs of the central nervous system, nose, and pancreas. In the case of a pathology of PAX6 gene expression, the formation of the anatomical structures of the eyeball is disrupted, which occurs at 12-14 weeks of gestation. The cause of the development of aniridia may be a traumatic injury to the eyes, accompanied by a detachment of the iris at the root.

Symptoms of aniridia. By origin in modern ophthalmology, congenital and acquired (traumatic) forms of aniridia are distinguished. Symptoms of traumatic irideremia depend on the degree of damage to the iris. From a clinical point of view, aniridia is distinguished as complete, partial, and combined with Gillespie and WARG syndrome. The full form of the disease is characterized by the presence of small remnants of the iris root. Clinically, the disease is manifested by a decrease in visual acuity, which is associated with underdevelopment of the structures of the eyeball. The degree of visual acuity reduction directly depends on the history of cataracts, glaucoma and keratopathy. The absence of the iris leads to increased photosensitivity. One of the symptoms of complete aniridia is horizontal nystagmus in combination with strabismus.

For partial aniridia, the same symptoms are characteristic as for the full form of pathology. But due to the fact that the disease is characterized by a mild degree of hypoplasia of the stroma of the iris, the clinical manifestations are mild. WARG syndrome, in addition to aniridia, includes Wilms' tumor (malignant nephroblastoma), diseases of the genitourinary system, and mental retardation. Often this symptom complex includes pancreatitis and chronic renal failure. The appearance of patients may be accompanied by hemihypertrophy (hypertrophy of the muscular apparatus of one of the halves of the body). In the early stages of development, the syndrome may not have pronounced clinical manifestations, which is the reason for underdiagnosis.

Gillespie's syndrome is characterized by a combination of aniridia with a clinical picture of cerebellar ataxia and mental retardation. A number of patients have ptosis, hearing loss, and stenosis of the pulmonary valve.

Diagnostics. The complex of diagnostic measures for aniridia includes visual examination of the anterior parts of the eyes, ophthalmoscopy, tonometry, gonioscopy, clinical refraction study and ultrasound biomicroscopy. Genetic analysis allows you to determine the cause of the development of aniridia. It is carried out only in case of congenital forms of the disease.

• Examination of the eye. When examining the anterior parts of the eyes, it is not possible to visualize the iris. In particular cases, small accumulations of the iris are observed, which is most characteristic of traumatic or partial aniridia. Ophthalmoscopy can reveal hypoplasia of the central part of the retina and optic nerve, which is typical for the full form of aniridia.

• Tonometry. Allows you to measure intraocular pressure (IOP). An increase in IOP above tolerable values is a risk factor for glaucoma. In patients with aniridia, it is recommended to measure blood pressure using the ICare device, which allows manipulation without anesthetics. In order to prevent the development of glaucoma, it is necessary to carry out tonometry and ophthalmoscopy in dynamics. Gonioscopy is required in patients with elevated intraocular pressure for additional examination of the anterior chamber of the eye.

• Study of clinical refraction. It is carried out by skiascopy, direct ophthalmoscopy or refractometry. Depending on the comorbidity, emmetropic, hyperopic and myopic refraction can be detected.

• Ultrasound biomicroscopy. The method of ultrasonic biomicroscopy allows assessing the residual tissue of the iris and examining the fundus of the eye in patients with cataracts or keratopathy.

• Genetic analyzes. To confirm hereditary aniridia, a FISH test and studies to determine the type of defect in the PAX6 gene are performed. A FISH test is necessary to rule out WARG syndrome. Until the genetic analysis is carried out, it

is necessary to conduct an ultrasound scan of the kidneys and pelvic organs once every 3 months.

• Other studies. Optical coherence tomography, visual evoked potentials and corneal topography are used as additional research methods.

Aniridia treatment. Specific treatment for aniridia is based on the implantation of an artificial iris. The prosthesis consists of a special hydrogel with a hole corresponding to the pupil. The color of the artificial iris is selected according to the color of the patient's eyes. Surgical intervention is performed using transscleral surgical access.

Artificial iris implantation is indicated for patients with traumatic aniridia. In congenital forms of the disease, surgery is performed only if the risk of damage to the cornea is minimal. When aniridia is combined with cataract, the tactics of the operation is reduced to the implantation of a prosthesis that replaces both the iris and the lens.

Symptomatic treatment includes the use of cosmetic contact lenses to hide the iris defect. With an increase in intraocular pressure, it is recommended to use drops from the group of carbonic anhydrase inhibitors, prostaglandins and beta-blockers. To prevent the development of keratopathy, it is necessary to use moisturizing drops and gels. All patients must wear sunglasses that provide 100% UV blocking.

Forecast and prevention. Specific measures for the prevention of aniridia have not been developed, since the disease is associated with a genetic mutation. A family in which one of the parents is sick with aniridia should consult a geneticist and an ophthalmologist before planning a pregnancy. Prevention of traumatic aniridia comes down to personal safety, since a strong blow can cause the iris to tear off.

Subject to all recommendations, the prognosis for congenital aniridia is favorable for life. The development of glaucoma or the association of aniridia with Gillespie and WARG syndrome can cause early disability of the patient.

Literature

1. Congenital aniridia. Clinical guidelines. - 2019.

2. Genetic aspects of congenital aniridia / Vasilyeva T.A., Pozdeeva N.A.,

Voskresenskaya A.A. etc.// Practical medicine. - 2015.

3. Clinical features of congenital aniridia in childhood / Voskresenskaya A.A., Pozdeeva N.A., Vasilyeva T.A. and others// Russian pediatric ophthalmology. - 2016.