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ATAKSİYA-TELANGİEKTAZİYA: OFTALMOLOJİ TƏZAHÜRLƏRLƏ MÜŞAYİƏT OLUNAN NADİR İRSİ SİNDROMU (KLİNİK HAL)

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XÜLASƏ

Ataksiya-telangiektaziya (AT), həmçinin Lui-Bar sindromu kimi tanınan bu xəstəlik, ATM genindəki mutasiyalar nəticəsində yaranan nadir autosom-resessiv patologiyadır. Xəstəlik proqressiv serebellar ataksiya, immunçatışmazlıq, dəridə və konyunktivada telangiektaziya ilə xarakterizə olunan klassik triada ilə səciyyələnir. Oftalmoloji təzahürlər, xüsusən də konyunktival telangiektaziya, xəstəliyin xaricdən görünən ilk əlamətləri ola bilər və erkən diaqnoz üçün əhəmiyyət kəsb edir.

Məqsəd – AT ilə bir uşaqda oftalmoloji əlamətləri təqdim etməklə nadir irsi xəstəliyin erkən aşkar edilməsində və multidisiplinar idarə olunmasında oftalmoloqun rolunu qiymətləndirmək.
10 yaşlı AT diaqnozu ilə qız uşağı qız uşağı görmə itiliyin zəifləməsi və dövri konyunktivanın qızartısı şikayətləri ilə müraciət etmişdir. Oftalmoloji müayinədə ikitərəfli göz almasının konyunktivasının teleangioektaziyaları, həmçinin optik koherens tomoqrafiyada qanglion hüceyrə qatında orta dərəcəli incəlmə aşkar edilmişdir ki, bu da tor qişanın erkən neyrodegenerativ dəyişikliklərinə işarə edir. Genetik müayinə patogen ATM mutasiyasını təsdiqləmişdir.
Təqdim olunan klinik hal oftalmoloqların AT-nın erkən aşkarlanmasında həlledici rolunu vurğulayır. Konyunktival teleangioektaziyalar nevroloji və immunoloji patoloji klinik əlamətləri ilə birlikdə patoqnomik və asan müşahidə olunan marker kimi çıxış edir. Oftalmoloji əlamətlərin erkən aşkar edilməsi xəstələrin genetik və immunoloji müayinələrə vaxtında yönləndirməni təmin edir və bu patologiyanın müalicə taktikasında çoxşaxəli yanaşmasına təşviq edir.

Açar sözlər: *ataksiya- telangiektaziya, Lui-Bar sindromu, konyunktival teleangioektaziyalar*

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ATAXIA-TELANGIECTASIA: A CLINICAL CASE OF A RARE HEREDITARY SYNDROME WITH OPHTHALMOLOGIC MANIFESTATIONS (CLINICAL CASE)

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Concept and design of investigation: Aghayeva T.M.
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SUMMARY

Ataxia-telangiectasia (AT), also known as Louis-Bar syndrome, is a rare autosomal recessive disorder caused by mutations in the ATM gene. The disease is characterized by a classic triad of progressive cerebellar ataxia, immunodeficiency, and telangiectasias of the skin and conjunctiva. Ophthalmologic manifestations, particularly conjunctival telangiectasias, may be the first externally visible signs of the disease and provide an important clue for early diagnosis.

Purpose – to demonstrate ophthalmologic presentations of ataxia-telangiectasia in a child and to emphasize the diagnostic role of the ophthalmologist in the early detection and interdisciplinary management of rare inherited diseases.

We report the case of a 10-year-old girl with AT presenting with decreased vision and intermittent conjunctival redness. Examination revealed symmetrical bulbar conjunctival telangiectasias, and moderate thinning of the ganglion cell layer on optical coherence tomography (OCT), suggesting early retinal neurodegenerative changes. Genetic analysis confirmed a pathogenic ATM mutation.

This case highlights the critical role of ophthalmologists in the early recognition of AT. Conjunctival telangiectasias, when combined with neurological and immunological findings, serve as a pathognomonic and visually accessible marker. Early ophthalmologic recognition facilitates timely referral for genetic and immunologic evaluation, contributing to multidisciplinary management and improved quality of life for affected patients.

Key words: *Ataxia-telangiectasia, Louis-Bar syndrome, conjunctival telangiectasias*

Ataxia-telangiectasia (AT), or Louis-Bar syndrome, was first described in 1941 [1]. The disease occurs at an incidence of 1:40,000-1:100,000 newborns [cc2] and is caused by mutations in the ATM gene located on the long arm of chromosome 11 (11q22-23) [2, 3].

The ATM gene encodes a serine/threonine protein kinase that plays a key role in repairing double-stranded DNA breaks and maintaining genomic stability, a defect in this enzyme leads to tissue radiosensitivity, immunodeficiency, cerebellar degeneration and the development of telangiectasias.

The classic clinical triad of the disease includes cerebellar ataxia (onset at the age of 1-4 years and progression), telangiectasias of the skin and conjunctiva typically appearing between 3 and 6 years of age and immunodeficiency due to defective maturation of B- and T-lymphocytes.

Conjunctival telangiectasias, usually painless and symmetrical, may be the first external symptom noticed by the physician and parents, long before the onset of significant abnormalities. Telangiectasias are dilations of small vessels and more commonly appear on the conjunctiva, facial skin, and auricles, but can occur on other areas of the body, so the patient should be carefully examined. Literature shows that they usually manifest not from the very early age, but after 5-6 years of age. In some cases, they may not develop at all, and their absence does not exclude the diagnosis of AT [4]. It should be noted that these are persistent vascular changes, unlike, conjunctivitis, and telangiectasias do not tend to bleed [5, 6]. In the eye, telangiectatic changes predominantly affect the bulbar conjunctiva, particularly near the limbus. Fragility of the conjunctival and episcleral capillaries results from structural weakening of the vascular wall, impaired endothelial repair, and altered perivascular connective tissue. Chronic oxidative stress and immune dysregulation further promote endothelial permeability and irregular vessel remodeling [7, 8].

Clinically, these changes manifest as tortuous, dilated conjunctival vessels without signs of inflammation, representing one of the most characteristic ophthalmic signs of the disease.Thus, the ophthalmologist is often the first specialist able to suspect a rare systemic syndrome.

Purpose - to demonstrate ophthalmologic presentations of ataxia-telangiectasia in a child and to emphasize the diagnostic role of the ophthalmologist in the early detection and interdisciplinary management of rare inherited diseases..

Material and methods

Patient A.M., born in 2015, was delivered prematurely with a birth weight of 2950 g. Her early neonatal period was unremarkable, and her physical and psychomotor development up to one year of age corresponded to age norms. From 18 months of age, the parents noticed instability while walking, unsteady gait, and frequent falls, which gradually progressed over time. From the age of 3 years, she experienced frequent episodes of respiratory infections, including bronchitis, otitis media, and pneumonia, which required repeated courses of antibiotic therapy.According to the medical history, the patient previously exhibited areas of telangiectasia on the skin of the lower leg and foot. A photograph documenting these lesions was provided by the patient's mother **(Figure 1).**

The patient was followed at the Children's Regional Clinical Hospital of Krasnodar. Immunologic evaluation revealed decreased IgG levels and moderate hypo-IgA-globulinemia. The level of α-fetoprotein was significantly increased - more than 300 times compared to the age norm. Genetic study revealed a pathogenic ATM c.5932G>T mutation, confirming the diagnosis of ataxia-telangiectasia. Neurological examination revealed marked cerebellar ataxia, intention tremor, and impaired coordination on motor accuracy tests. Brain MRI showed diffuse cerebellar atrophy with enlargement of the cerebellar sulci.

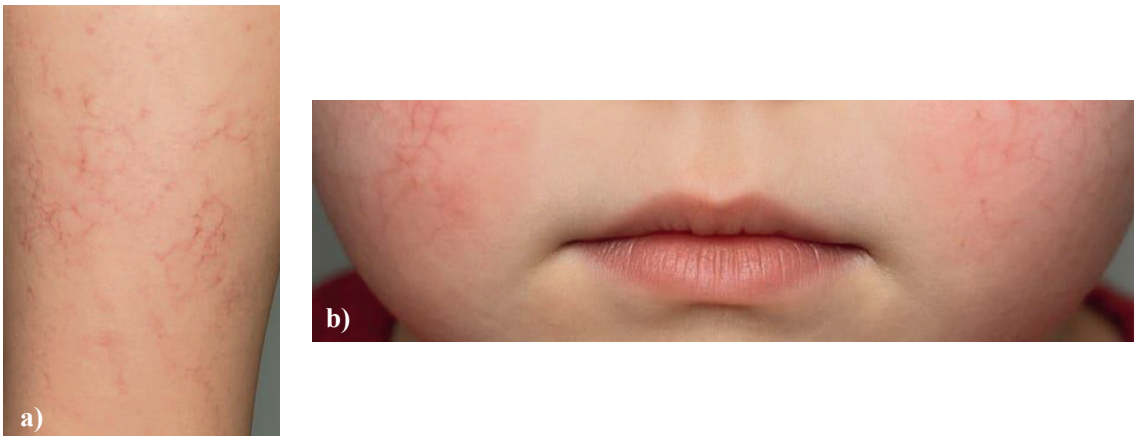

Figure 1. *Local skin manifestations of telangiectasias on the patient's arm(a) and cheek(b).*

In 2025, the patient presented to the National Ophthalmology Centre named after Academician Zarifa Aliyeva with complaints of decreased vision and intermittent conjunctival redness without pain. Ophthalmologic examination revealed visual acuity of 0.5 in both eyes, improving to 1.0 with corrective lenses. Individual optical correction was prescribed (OD -1.5 D sph -0.75 D cyl × 180, OS -1.25 D sph -0.5 D cyl × 175), which provided stable visual acuity, reduced asthenopia, and ensured comfortable binocular vision.

Biomicroscopy revealed pronounced telangiectasias of the bulbar conjunctiva in the interpalpebral area, mainly in the outer quadrants, with tortuous vessels and without associated inflammation or edema. The cornea was completely transparent, with epithelial and stromal layers showing no structural abnormalities.

Fundus examination revealed a normal optic disc with clear borders, retinal vessels of normal caliber, and a macula without signs of degeneration **(Figure 2)**. Tonometry showed intraocular pressures within the normal range (OD 18 mmHg, OS 19 mmHg), indicating no signs of ocular hypertension or glaucoma.

The observed conjunctival telangiectasias exhibited typical localization and morphology, supporting their diagnostic significance in AT, particularly when combined with neurological and immunological abnormalities **(Figure. 3).** As shown in Figure 3, telangiectatic dilatation of conjunctival and episcleral vessels is visible, most pronounced in the bulbar conjunctiva near the limbus. The vessels appear tortuous, irregularly branched, and form a distinct

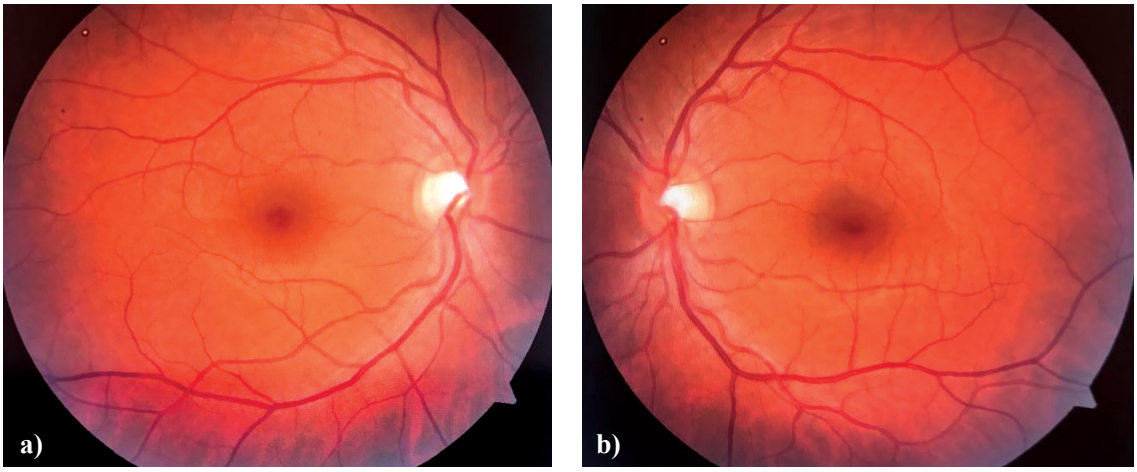

Figure 2. *Ocular fundus of a patient with AT (a-OD, b-OS).*

Figure 3. *The conjunctival telangiectasias of a patient with AT: characteristic ocular vascular changes in AT.*

vascular network. These changes reflect chronic vascular fragility and endothelial dysfunction typical of AT and are often accompanied by mild conjunctival injection without signs of active inflammation.

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OCT was performed to assess the retinal and vascular structures. As shown in **Figure 4**, moderately reduced ganglion cell layer values are visualized, which may correspond to early retinal neurodegenerative changes. Although these changes are not specific to AT alone, in combination with the clinical picture (ataxia, immunosuppression, telangiectasia) they support the diagnosis. These OCT findings may reflect the neurodegenerative progression associated with ATM gene mutations, linking ocular microvascular alterations with systemic disease manifestations.

Discussion

Ophthalmologic examination in AT has a dual diagnostic and monitoring value. Conjunctival telangiectasias often appear 1–2 years before the onset of pronounced neurological disorders and may serve as an early ocular marker of AT [6]. When examining a child with preserved visual acuity, symmetrical vascular changes, and no signs of inflammation, an ophthalmologist should consider a systemic process rather than simple conjunctivitis.

The differential diagnosis of conjunctival telangiectasias should include chronic blepharoconjunctivitis, rosacea with vascular changes, radiation- and toxin-induced angiopathies, Osler-Rendu-Weber syndrome (hereditary hemorrhagic telangiectasia), and vascular changes associated with chronic ocular hypertension [7]. In patients with AT, however, vascular changes exhibit characteristic features: they are non-inflammatory, symmetric, persistent under pressure, and follow typical age-related dynamics, often appearing between 3 and 6 years of age. These features distinguish them from normal conjunctival vascular changes [9].

Modern ophthalmologic technologies allow not only to more accurately record vascular changes, but also to objectively assess their progression. Such monitoring is particularly important in AT, as the immunodeficiency characteristic of the disease can be accompanied by frequent

conjunctivitis, keratitis, and herpetic eye lesions [5, 9]. Moreover, ophthalmologic observation in such cases is not limited to vessel visualization. It is also important to monitor the anterior segment (cornea and anterior chamber), the ocular fundus, particularly during prolonged disease courses, and to assess interactions with neurological manifestations, such as oculomotor apraxia, nystagmus, and fixation disorders [10]. Thus, the ophthalmologist acts not only as a "conjunctival diagnostician" but also as part of multidisciplinary team to help with early recognition, correction, and monitoring of AT complications.

Conclusion

Ataxia-telangiectasia is a rare but clinically recognizable condition in which ophthalmic manifestations may be the first clue to the diagnosis. Conjunctival telangiectasia is a pathognomonic and visually accessible symptom, especially when combined with mild neurological abnormalities in the child.

Early ophthalmologic recognition facilitates referral for genetic and immunologic testing, thereby accelerating diagnosis and initiation of therapy. Multidisciplinary follow-up involving an ophthalmologist, neurologist, and immunologist improves quality of life and reduces the risk of complications in patients with AT.

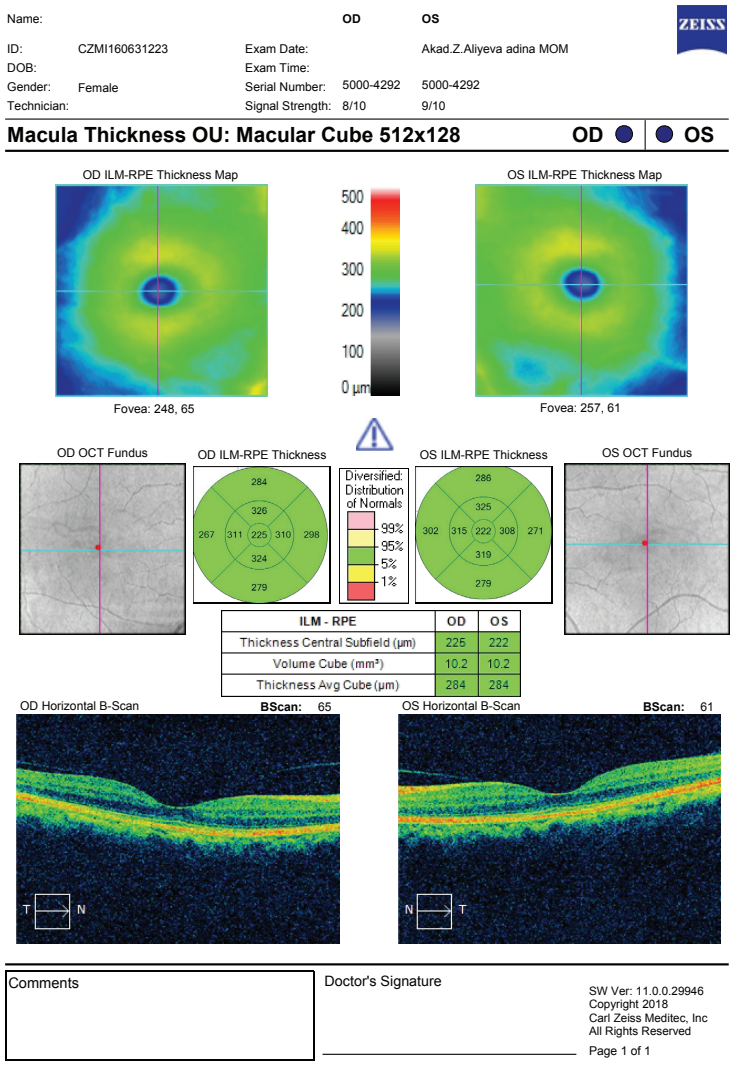

Figure 4. *OCT of a patient with AT: moderately reduced ganglion cell layer values are visualized, which may correspond to early retinal neurodegenerative changes.*

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