Comprehensive ophthalmological assessment in children with down syndrome: a cross-sectional study from Uzbekistan

Umida Khamraeva, Muyassar Karimova

Republican Specialized Scientific and Practical Medical Center of Eye Microsurgery, Tashkent (Uzbekistan)

Комплексне офтальмологічне обстеження дітей із синдромом Дауна: перехресне дослідження з Узбекистану

Уміда Хамраєва, Муяссар Карімова

Республіканський спеціалізований науково-практичний медичний центр мікрохірургії ока, Ташкент (Узбекистан)

Abstract

Objective. *To determine the prevalence and spectrum of ocular abnormalities in children with Down syndrome in Uzbekistan using an adapted, comprehensive clinical and functional ophthalmological assessment protocol.*

Materials and Methods. *A descriptive observational clinical study was carried out at the Republican Specialized Scientific-Practical Medical Center for Eye Microsurgery and included 61 children (122 eyes) aged from 4 months to 15 years with genetically confirmed Down syndrome. All participants underwent an expanded ophthalmological examination that included adapted visometry, slit-lamp biomicroscopy, ophthalmoscopy, objective refraction assessment and autorefractometry, tonometry, B-scan ultrasonography, corneal Scheimpflug tomography (Pentacam and WaveLight Oculyzer), and OCT. Examination techniques were modified according to the patients' age, cognitive characteristics, and degree of cooperation. Quantitative parameters were compared with published age-specific normative values.*

Results*. Multiple concurrent ocular findings were present in the majority of children; only 6.6% had no detectable ocular pathology. The most common findings were epicanthus (91.8%), refractive error (73.8%), Brushfield spots (72.1%), strabismus (27.9%), lacrimal drainage pathology (18.0%), amblyopia (13.1%), keratoconus (4.9%), and optic nerve anomalies (4.9%). Mean cycloplegic spherical equivalent was +0.20 ± 2.76 D, mean intraocular pressure 14.4 ± 2.9 mmHg, and mean axial length 22.83 ± 1.49 mm. Comprehensive tomography additionally detected suspected subclinical corneal ectasia in two children in whom routine examination had found no keratoconus.*

Conclusion. *A structured and multidisciplinary ophthalmological examination algorithm substantially improves the detection of ocular pathology in patients with Down syndrome, including subclinical abnormalities. The integration of adapted comprehensive assessment protocols into routine ophthalmological follow-up may contribute to improved visual outcomes, enhanced social adaptation, and reduced long-term healthcare burden.*

Keywords: *Down syndrome, Down's Syndrome Medical Interest Group, multiple ophthalmological disorders, eyelid and ocular adnexa anomalies, oculomotor system disorders, ocular surface disorders, refractive Errors, retinal and optic nerve pathology.*

Резюме

Мета. *Визначити поширеність та спектр очних аномалій у дітей із синдромом Дауна в Узбекистані за допомогою адаптованого, комплексного клінічного та функціонального офтальмологічного протоколу оцінки.*

Матеріали та методи. *У Республіканському спеціалізованому науково-практичному медичному центрі мікро-*

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Corresponding author: Khamraeva Umida, PhD student; Republican Specialized Scientific and Practical Medical Center of Eye Microsurgery; Tashkent, Uzbekistan. E-mail: umidakhamraeva21@gmail.com

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хірургії ока було проведено описове обсерваційне клінічне дослідження, до якого залучено 61 дитину (122 очі) віком від 4 місяців до 15 років з генетично підтвердженим синдромом Дауна. Усім учасникам було проведено розширене офтальмологічне обстеження, яке включало адаптовану візометрію, біомікроскопію зі щілинною лампою, офтальмоскопію, об'єктивну оцінку рефракції та авторефрактометрію, тонометрію, УЗ В-сканування, Шаймпфлюг томографію рогівки (Pentacam та WaveLight Oculyzer) та оптичну когерентну томографію (ОКТ). Методи обстеження були модифіковані відповідно до віку пацієнтів, когнітивних характеристик та ступеня співпраці. Кількісні параметри порівнювали з опублікованими віково-специфічними нормативними значеннями.

Результати. *У більшості дітей спостерігалися численні одночасні очні зміни; лише у 6,6% не було виявленої очної патології. Найчастішими знахідками були епікантус (91,8%), помилка рефракції (73,8%), плями Брашфілда (72,1%), косоокість (27,9%), патологія слізного дренажу (18,0%), амбліопія (13,1%), кератоконус (4,9%) та аномалії зорового нерва (4,9%). Середній циклоплегічний сферичний еквівалент становив +0,20 ± 2,76 D, середній внутрішньоочний тиск 14,4 ± 2,9 мм рт. ст., а середня осьова довжина 22,83 ± 1,49 мм. Комплексна томографія додатково виявила підозру на субклінічну ектазію рогівки у двох дітей, у яких під час планового обстеження не було виявлено кератоконусу.*

Висновок. *Структурований та міждисциплінарний алгоритм офтальмологічного обстеження суттєво покращує виявлення очної патології у пацієнтів із синдромом Дауна, включаючи субклінічні аномалії. Інтеграція адаптованих протоколів комплексної оцінки в планове офтальмологічне спостереження може сприяти покращенню зорових результатів, покращенню соціальної адаптації та зменшенню довгострокового навантаження на охорону здоров'я..*

Ключові слова: *синдром Дауна, медична група з вивчення синдрому Дауна, множинні офтальмологічні захворювання, аномалії повік та придатків ока, захворювання окорухової системи, захворювання поверхні ока, рефракційні помилки, патологія сітківки та зорового нерва.*

Introduction

Down syndrome (DS) is the most common chromosomal disorder, characterized by complete or partial trisomy of chromosome 21. Beyond its systemic manifestations, DS is associated with a wide range of ophthalmological abnormalities that may affect nearly all structures of the visual system. As a result of improved survival, advances in neonatal care, and increased life expectancy, the global population of individuals with DS continues to grow, and an increasing proportion now survive into adulthood and older age — shifting clinical priorities from predominantly pediatric management toward lifelong ophthalmological surveillance [1,2,3].

Visual impairment is among the most common comorbidities in DS, with reported prevalence of ophthalmological disorders ranging from 60% to 100%, depending on patient age, diagnostic criteria, and study design [4,5,6,7]. The spectrum includes eyelid abnormalities, lacrimal drainage disorders, refractive errors, strabismus, amblyopia, cataract, keratoconus, retinal pathology, and optic nerve anomalies. This involvement is often multifactorial and systemic in nature, reflecting the complex genetic and embryological mechanisms associated with trisomy 21 [5,8]. Consequently, isolated assessment of individual disorders may fail to capture the full extent of visual dysfunction, supporting the need for a systemic, integrated approach to evaluation [7].

The clinical consequences extend beyond ocular health. Impaired vision may adversely affect cognitive development, educational performance, social adaptation, and independent functioning, and adults with intellectual disability and coexisting strabismus experience more pronounced visual deterioration while frequently receiving insufficient ophthalmological care [9]. Given current understanding of neuroplasticity and the role of sensory input in child development, timely identification and correction of ocular abnormalities are considered essential for improving long-term functional prognosis in DS [5,10].

The clinical evidence reinforces both the breadth and the age-dependence of ocular involvement. Strabismus, amblyopia, and anterior segment abnormalities are among the most common findings [2,6,11], while eyelid malpositions, lacrimal pathway abnormalities, and oculomotor dysfunction are also highly prevalent [12]. Refractive errors are particularly frequent and are associated with both systemic and ocular features of the syndrome [11]. Retinal pathology, historically considered uncommon, has more recently been shown to encompass a diverse range of retinal and vascular abnormalities [13]. Clinical focus also shifts with age — from refractive errors, strabismus, and amblyopia in children to cataract, keratoconus, and retinal pathology in adolescents and adults [4,13,14] — and manifestations may vary across populations, highlighting the need for individualized approaches [8].

Although ophthalmological surveillance recommendations exist, including those of the Down's Syndrome Medical Interest Group (DSMIG), significant variability in clinical practice persists, and existing guidelines often do not provide detailed examination algorithms adapted to the clinical and behavioral characteristics of patients with DS [14]. The development of a structured, comprehensive, and adaptable assessment protocol therefore remains an important and clinically relevant challenge [10].

On this basis, we hypothesized that a structured, age- and cognition-adapted comprehensive ophthalmological protocol would reveal a high burden and broad spectrum of ocular abnormalities — including subclinical corneal and

posterior-segment changes — in children with Down syndrome, and that quantitative functional and morphological parameters would deviate from age-specific norms.

Objective of the study: to determine the prevalence and spectrum of ocular abnormalities in children with Down syndrome in Uzbekistan using an adapted, comprehensive clinical and functional ophthalmological assessment protocol.

Materials and methods

This investigation was designed as a clinical descriptive observational study providing a comprehensive clinical and functional evaluation of the visual system in children with Down syndrome. Of 100 children with Down syndrome examined during the study period, the 61 (122 eyes) who underwent the most complete comprehensive assessment were included; selection was based solely on completeness of examination data, independently of clinical findings. The study population included infants, children, and adolescents aged from 4 months to 15 years, with a male-to-female ratio of approximately 3:2.

Inclusion and exclusion criteria

Inclusion criteria were: (1) genetically (karyotype-) confirmed trisomy 21; (2) age 4 months–15 years; (3) written parental or guardian consent; and (4) sufficient cooperation for at least the core non-invasive examination. Exclusion criteria were: (1) absence of genetic confirmation; (2) acute ocular infection or inflammation at examination; (3) prior intraocular surgery or ocular trauma that could confound morphological assessment; (4) systemic condition precluding safe examination; and (5) withdrawal of consent.

All examinations were performed in accordance with the recommendations of the Down's Syndrome Medical Interest Group (DSMIG). Particular attention was devoted to adapting the diagnostic process according to patient age, cognitive characteristics, behavioral profile, and level of cooperation.

Ophthalmological Examination Methods

The examination block comprised visual acuity assessment, objective refractometry, biomicroscopy, ophthalmoscopy, tonometry, B-ultrasound examination, corneal tomography and optical coherence tomography.

Visual acuity assessment. Visual acuity was evaluated using pediatric and cognitively adapted optotypes, including the Orlova chart and Landolt rings. In young children and those with pronounced cognitive impairment, visual function was assessed using fixation, following, and preferential-looking responses.

Objective refractometry. Cycloplegia was induced using a fixed combination of phenylephrine 5% and tropicamide 0.8% (Midrimax); atropine was not used, in accordance with national paediatric practice, owing to its higher risk of adverse effects in children and the prolonged accommodative recovery it entails. Objective refraction was then assessed by retinoscopy and automated refractokeratometry (HRK-7000A, Huvitz Co., Ltd., Republic of Korea).

Biomicroscopy. Slit-lamp biomicroscopy was used to assess the anterior segment, including eyelid abnormalities, epicanthus, Brushfield spots, conjunctival status, corneal condition, anterior chamber, and lens transparency; a portable handheld slit lamp was used in young or poorly cooperative children (Fig 1 – see cover page 2).

Ophthalmoscopy. Both direct and indirect ophthalmoscopy were performed under mydriasis to evaluate the retina and optic nerve head.

Tonometry. Intraocular pressure was measured by dynamic transpalpebral tonometry without topical anaesthesia using a TVGD-02 portable tonometer (ELAMED, Russia), avoiding corneal contact and anaesthetic instillation.

Ultrasound B-scan. B-scan ultrasonography (E-Z Scan B5500+, Sonomed Escalon, USA) was used to assess the vitreous and posterior-segment coats and to measure axial length, particularly where media opacity or limited cooperation precluded ophthalmoscopy (Fig 2 – see cover page 2).

Corneal tomography. Scheimpflug tomography was performed in a subgroup of cooperative children and children with suspected corneal pathology, using an OCULUS Pentacam (OCULUS Optikgeräte GmbH, Wetzlar, Germany) and a WaveLight Allegro Oculyzer (Alcon/WaveLight, Germany), with analysis via the Belin/Ambrósio Enhanced Ectasia Display (BAD-D). Owing to the small number and partly pathology-driven selection, tomographic findings are presented descriptively per patient and per device and were not used to derive prevalence estimates (Fig 3 – see cover page 3).

Optical coherence tomography. OCT was performed in a subgroup using swept-source and spectral-domain systems (Topcon Maestro2 and Triton plus, Topcon, Japan; REVO, Optopol, Poland), assessing macular thickness and volume and, where available, ganglion-cell parameters. Because the subgroup was examined on different devices with distinct normative databases, OCT values are reported descriptively (Fig 4 – see cover page 3).

Statistical analysis

Absolute and relative frequencies of identified abnormalities were calculated, and continuous variables were expressed as mean ± standard deviation with ranges. As no internal control group was recruited, quantitative parameters were compared against published age-specific normative reference values, each explicitly identified as a literature comparison rather than an internal control. Prevalence of ocular conditions is reported per patient; quantitative ocular parameters (refraction, intraocular pressure, axial length, and visual acuity) are reported per eye. The denominator is specified for each measure.

Adaptation of the examination protocol

Examination of patients with Down syndrome entails several objective difficulties arising from both cognitive features and anatomical-physiological characteristics of this population, and accounting for these was an integral

Table 1. Adaptation of the examination protocol to the characteristics of patients with Down syndrome

Limiting factor	Adaptive measure
Difficulty understanding instructions	Priority on objective methods (retinoscopy, autorefractometry, OCT, B-scan) not requiring a conscious response
Short cooperation window	"Non-contact before contact" sequence; combining several examinations into one short session
Difficulty positioning at devices	Use of portable devices: handheld slit lamp, portable transpalpebral tonometer TVGD-02
Intolerance of contact/anesthesia	Through-the-lid tonometry without anesthetic; non-contact techniques
Heightened anxiety	Presence of a parent/guardian; calm environment; prior familiarization with the room
Inability to assess vision subjectively	Adapted charts, preferential-looking methods, objective methods

part of the study methodology. The principal limiting factors included reduced cognitive function and difficulty understanding instructions; short periods of sustained attention and cooperation; heightened anxiety and negative behavioral reactions in unfamiliar settings; anatomical features (narrow palpebral fissures, epicanthus, nystagmus, strabismus) hindering positioning at stationary devices; and concurrent systemic pathology limiting the use of sedation. To minimize their influence, a set of adaptive measures was implemented (Table 1).

This protocol adaptation establishes a reproducible, patient-capability-oriented scheme of ophthalmological examination in Down syndrome that yields the fullest possible diagnostic information with minimal patient burden.

Ethical Considerations

The study was conducted in full accordance with the ethical principles of the Declaration of Helsinki (2013 revision) and established standards of biomedical ethics. At the time the study was conducted, the Republican Specialized Scientific-Practical Medical Center for Eye Microsurgery did not maintain a separate standalone ethics committee; institutional ethical review of research is performed by the Scientific Council, whose membership includes a representative of the National Ethical Committee of Uzbekistan (NECU). The study was reviewed and approved by the Scientific Council (Extract from Minutes No. 2, 27 February 2025). Particular consideration was given to the ethical aspects of research in children with Down syndrome, who are regarded as a vulnerable population; all procedures were non-invasive and corresponded to routine ophthalmological practice, with invasive procedures avoided on both ethical grounds and the absence of clinical indications. Written informed consent was obtained from the parents or legal guardians of all participating children prior to inclusion.

Results

Study population. Sixty-one children with genetically confirmed trisomy 21 were included. Mean age was 6.1 ± 4.5 years (median 4.5; range 4 months–15 years); 36 (59.0%) were male and 25 (41.0%) female. Twenty-two children (36.1%) were under 3 years, 15 (24.6%) were 3–6 years, and 24 (39.3%) were 7 years or older (Table 2A).

Visual acuity. Quantitative visual acuity was obtainable in 14 of 61 children (23.0%; 27 eyes), all aged 7 years or older; uncorrected acuity averaged 0.68 ± 0.37 logMAR and best-corrected acuity 0.51 ± 0.30 logMAR (8 children, 16 eyes). In the remaining 47 children (77.0%), subjective testing was not feasible: 44 (72.1%) fixated and followed, 2 (3.3%) did neither, and 1 (1.6%) was uncooperative. This distribution reflects the age and cognitive profile of the cohort rather than a limitation of the protocol (Table 2A).

Intraocular pressure and axial length. Transpalpebral tonometry succeeded in all 61 children (122 eyes), mean 14.4 ± 2.9 mmHg (range 9–21); no eye exceeded 21 mmHg and no glaucoma was identified. Axial length, measured in 18 children (36 eyes), averaged 22.83 ± 1.49 mm (range 18.2–24.5) (Table 2A).

Refraction. Cycloplegic refraction was obtained in 55 children (90.2%; 110 eyes) and was not obtainable in 6 (9.8%) owing to media opacity or non-cooperation. Mean spherical equivalent was +0.20 ± 2.76 D (range −8.62 to +7.25). Hyperopia (SE ≥ +0.50 D) was present in 57 eyes (51.8%), myopia (≤ −0.50 D) in 30 (27.3%), and emmetropia in 23 (20.9%); astigmatism ≥ 1.00 D was recorded in 25 eyes (22.7%) (Table 2B).

B-scan ultrasonography. Performed in all 61 children, B-scan showed vitreous changes in 39 (63.9%): slight opacities in 21 (34.4%), moderate in 9 (14.8%), filamentary in 5 (8.2%), and marked destruction in 4 (6.6%) (Table 2B).

Corneal tomography

Scheimpflug tomography was performed in 4 children on two devices (Table 3). Keratoconus was confirmed in 3 of 61 children (4.9%; 5 of 122 eyes, 4.1%), all adolescents: bilateral advanced disease in one 15-year-old (left eye K-max 72.1 D, thinnest pachymetry 399 μm, BAD-D 18.40; the right eye returned values at the limit of measurability, consistent with end-stage ectasia and probable scarring); bilateral stage 2–3 disease following corneal crosslinking in one 14-year-old (BAD-D 2.22 right, 2.05 left); and unilateral early keratoconus in one child (left eye BAD-D 4.45, pachymetry 468 μm) with suspected subclinical

Table 2 A. Cohort characteristics and quantitative ophthalmological parameters (n = 61)

Parameter	n assessed	Statistic	Value
Age, years	61	Mean ± SD (median; range)	6.1 ± 4.5 (median 4.5; 4 mo–15 y)
Sex: male	61	n (%)	36 (59.0%)
Sex: female	61	n (%)	25 (41.0%)
Age < 3 years	61	n (%)	22 (36.1%)
Age 3–6 years	61	n (%)	15 (24.6%)
Age ≥ 7 years	61	n (%)	24 (39.3%)
Uncorrected VA, decimal	14 (27 eyes)	Mean ± SD	0.27 ± 0.16
Uncorrected VA, logMAR	14 (27 eyes)	Mean ± SD (range)	0.68 ± 0.37 (0.22–1.70)
Best-corrected VA, decimal	8 (16 eyes)	Mean ± SD	0.37 ± 0.18
Best-corrected VA, logMAR	8 (16 eyes)	Mean ± SD	0.51 ± 0.30
Fixates / follows only	–	n (%)	44 (72.1%)
Neither fixates nor follows	–	n (%)	2 (3.3%)
Non-cooperative for VA	–	n (%)	1 (1.6%)
Cycloplegic spherical equivalent, D	55 (110 eyes)	Mean ± SD (range)	+0.20 ± 2.76 (−8.62 to +7.25)
Intraocular pressure, mmHg	61 (122 eyes)	Mean ± SD (range)	14.4 ± 2.9 (9–21)
Axial length, mm	18 (36 eyes)	Mean ± SD (range)	22.83 ± 1.49 (18.2–24.5)

SD = standard deviation; VA = visual acuity. Continuous variables are presented as mean ± standard deviation (with median and/or range where indicated); categorical variables as number (percentage). Denominators differ by parameter because not all instruments could be applied to every child.

Table 2 B. Distribution of refractive error and B-scan findings

Refraction (per eye; 110 eyes assessed)

Refractive category	Eyes (n)	%
Hyperopia (SE ≥ +0.50 D)	57	51.8
Clinically significant (SE ≥ +2.00 D)	26	23.6
Myopia (SE ≤ −0.50 D)	30	27.3
Moderate-to-high (SE ≤ −3.00 D)	10	9.1
Emmetropia	23	20.9
Astigmatism ≥ 1.00 D	25	22.7

B-scan ultrasonography (per patient; 61 assessed)

Vitreous finding	Patients (n)	%
No changes (attached retina)	22	36.1
Slight opacities	21	34.4
Moderate opacities	9	14.8
Filamentary opacities	5	8.2
Marked vitreous destruction	4	6.6
Any vitreous change	39	63.9

SE = spherical equivalent. Refraction reported per eye; B-scan per patient.

Table 3. Corneal Scheimpflug tomography in 4 children (descriptive)

Pt	Age	Device	Eye	K-max (D)	BAD-D	Interpretation
17	12	Pentacam	OD	45.7	2.54	suspected subclinical ectasia
			OS	45.7	0.70	borderline/normal
21	14	Oculyzer	OD	46.7	2.45	suspected subclinical ectasia
			OS	46.5	4.45	early keratoconus
35	15	Oculyzer	OD	104.3*	60.15*	advanced KC (measurement limit)
			OS	72.1	18.40	advanced keratoconus
81	14	Pentacam	OD	51.9	2.22	stage 2–3 KC, post-CXL
			OS	52.2	2.05	stage 2–3 KC, post-CXL

Pt = patient; OD = right eye; OS = left eye; D = dioptres; K-max = maximum keratometry; BAD-D = Belin/Ambrósio Enhanced Ectasia Display final deviation value; KC = keratoconus; CXL = corneal crosslinking. *Right-eye values in patient 35 reflect the limit of tomographic measurability in end-stage ectasia with probable corneal scarring and should be interpreted with caution.

Table 4. Prevalence of ophthalmological findings in 61 children with Down syndrome

Ocular finding	n	% (per patient)
Epicanthus†	56	91.8
Refractive error (any)	45	73.8
Hyperopia	29	47.5
Astigmatism	14	23.0
Myopia	13	21.3
Brushfield spots†	44	72.1
Strabismus (any)	17	27.9
Lacrimal drainage pathology	11	18.0
Amblyopia	8	13.1
Cataract / pseudophakia	7	11.5
Blepharitis	7	11.5
Nystagmus	5	8.2
Retinal pathology	5	8.2
Conjunctivitis	4	6.6
No ocular pathology	4	6.6
Optic nerve anomaly	3	4.9
Dry eye	3	4.9
Keratoconus	3	4.9
Pseudostrabismus	2	3.3
Microphthalmos	1	1.6
Blepharophimosis	1	1.6

†Epicanthus and Brushfield spots were recorded in the clinical diagnostic field rather than systematically graded; these figures may underestimate their true frequency, which approaches universality in trisomy 21.

ectasia in the fellow eye (BAD-D 2.45). In one additional child without keratoconus on routine examination, tomography demonstrated a borderline-abnormal deviation value in the right eye (BAD-D 2.54), interpreted as suspected subclinical ectasia. Thus comprehensive tomography identified early or subclinical corneal ectasia not detected on routine examination in two children.

Optical coherence tomography. OCT was obtained in 3 children across three devices and is reported descriptively. In all three – including the 15-year-old with bilateral advanced keratoconus – macular architecture was structurally normal (central subfield 191–306 μm; macular volume 8.3–8.6 mm³; normal ETDRS sectors), indicating that visual impairment in these children was attributable to corneal and lenticular disease rather than to maculopathy.

Comparison with normative values. Mean cycloplegic spherical equivalent (+0.20 ± 2.76 D) showed a central value comparable to published paediatric norms (+0.71 to +0.81 D) but a markedly wider dispersion (SD 2.76 vs 1.46–2.03 D), indicating greater refractive heterogeneity. Mean IOP (14.4 ± 2.9 mmHg) and axial length (22.83 ± 1.49 mm) fell within published paediatric reference ranges. Best-corrected visual acuity (0.51 ± 0.30 logMAR) was approximately fourfold higher (worse) than published age norms, indicating a substantial functional deficit, although this comparison is limited by the small measurable subgroup.

Spectrum and prevalence of ocular pathology (Table 4). Multiple concurrent findings were present in the majority of children; only 4 (6.6%) had no detectable ocular pathology. The most frequent findings were epicanthus (91.8%), refractive error (73.8%), and Brushfield spots (72.1%), followed by strabismus (27.9%), lacrimal drainage pathology (18.0%), amblyopia (13.1%), cataract or pseudophakia (11.5%), and blepharitis (11.5%).

Retinal pathology (5/61, 8.2%) ranged from peripheral retinal degeneration in three children – all with high myopia – to a single infant with severe congenital retinopathy comprising tractional retinal detachment, macular atrophy, and proliferative vitreoretinopathy. Optic nerve anomalies (3/61, 4.9%) comprised optic disc hypoplasia, partial optic atrophy, and myelinated nerve fibres (one each).

Discussion

This cross-sectional study applied a structured, age- and cognition-adapted comprehensive ophthalmological protocol to children with Down syndrome in Uzbekistan and confirmed a high burden and broad spectrum of ocular abnormality: all but four children had at least one ocular finding, and most had several concurrently. The predominance of refractive error, strabismus, and adnexal and lacrimal pathology is concordant with prior cohorts and systematic reviews, while the quantitative data – refraction, visual acuity, intraocular pressure, axial length, and tomographic and OCT parameters – allow the degree of functional and structural involvement to be characterised rather than merely enumerated.

*Refraction***.** In our cohort the mean cycloplegic spherical equivalent was +0.20 ± 2.76 D (110 eyes). Published normative data in healthy children give broadly comparable central values but markedly lower dispersion: a population-based feasibility study by Singh and coauthors reported a mean cycloplegic spherical equivalent of +0.81 ± 1.46 D (range −5.88 to +5.75 D), and Panke and coauthors' study of 472 healthy participants of European origin aged 3–28 years using cyclopentolate 1% reported a mean cycloplegic SE of 0.71 ± 2.03 D [15,16]. The near-doubling of the standard deviation in our cohort (2.76 D vs 1.46–2.03 D) and the far wider range (−8.62 to +7.25 D) indicate substantially greater refractive heterogeneity in children with Down syndrome, with both high hyperopia and high myopia over-represented relative to typically developing children. Because cycloplegia was achieved with a tropicamide-based agent rather than cyclopentolate, a small residual accommodation cannot be excluded; any resulting effect would underestimate hyperopia, rendering our refractive findings conservative..

Intraocular pressure. Mean IOP in our cohort was 14.4 ± 2.9 mmHg (122 eyes), with no eye exceeding 21 mmHg. This lies within published pediatric reference val-

ues: a study of 215 children aged 4–15 years with normal ophthalmological findings reported a mean IOP of 16.08 ± 3.08 mmHg across 406 eyes, a large Iranian study of 9154 eyes in children aged 9–15 years reported a mean of 15.58 ± 2.83 mmHg, and an Austrian school-children cohort reported means of 15.02 mmHg (SD 2.19) in boys and 14.44 mmHg (SD 2.01) in girls. Normal pediatric IOP is generally cited as ranging between 12 and 22 mmHg [17-20]. Our slightly lower mean is consistent with the younger age structure of our cohort and with the use of transpalpebral rather than applanation tonometry; importantly, no evidence of ocular hypertension or glaucoma was found.

Axial length. Mean axial length was 22.83 ± 1.49 mm (36 eyes). This is closely comparable to published pediatric values: 22.47 ± 0.91 mm (range 20.62–25.82 mm) in a population-based study and 22.87 ± 1.29 mm in a cohort of 187 children with a mean age of 5.6 years [21]. The greater dispersion in our data reflects the inclusion of an infant with bilateral microphthalmos (18.2–18.5 mm) alongside adolescents with high myopia (up to 24.5 mm).

Visual acuity. Best-corrected visual acuity in the subgroup in whom it could be measured (8 children, 16 eyes) averaged 0.51 ± 0.30 logMAR. For comparison, a pediatric cohort with a mean age of 5.6 years reported a mean BCVA of 0.12 ± 0.20 logMAR [21]. The approximately fourfold higher logMAR value in our cohort indicates a substantial deficit in corrected visual function, although this comparison must be interpreted with caution given the small measurable subgroup and the probable selection toward older, more cooperative children.

Corneal findings. The corneal findings merit particular emphasis. Keratoconus was confirmed in three children (4.9%), all adolescents, consistent with the recognised emergence of ectasia in the second decade; the low figure relative to adult Down syndrome series reflects the young age of this cohort rather than a discordant result. More importantly, comprehensive Scheimpflug tomography detected early or subclinical corneal ectasia in two further children in whom routine examination had found no keratoconus. This directly supports the central rationale of the study: a comprehensive protocol reveals corneal disease at a stage when standard assessment does not, offering the opportunity for earlier monitoring and timely crosslinking – an argument reinforced by the one child already stabilised after crosslinking. OCT, conversely, demonstrated structurally normal maculae even in the child with end-stage keratoconus, localising visual loss to the anterior segment and illustrating the complementary diagnostic value of structural imaging.

Limitations

Ophthalmological examination in children with Down syndrome may be complicated by limited cooperation, attention deficits, and behavioral characteristics, which can affect the precision of certain functional tests – a limitation widely acknowledged in this population. The study was single-centre and descriptive, without an internally recruited control group; quantitative comparisons were therefore made against published age-specific normative values, and findings should be regarded as hypothesis-generating. Quantitative visual acuity was obtainable in under a quarter of children; cycloplegia relied on a tropicamide-based agent, whose comparatively weaker effect may have slightly underestimated hyperopia – an effect that is conservative with respect to our findings – reflecting national paediatric safety practice.

Notwithstanding these challenges, the study contributes valuable data on the clinical and functional characteristics of the visual system in children with Down syndrome and supports the necessity of a comprehensive, individualized, and ethically adapted ophthalmological approach. Also the study demonstrates that a standardised, patient-adapted comprehensive protocol is feasible in a population in whom examination is challenging, and that it yields clinically meaningful quantitative and structural information – most strikingly the detection of subclinical corneal ectasia – that a basic examination would miss.

Conclusion

A structured, comprehensive, and age-adapted ophthalmological assessment protocol substantially improves the detection of ocular pathology in children with Down syndrome, including subclinical corneal ectasia detectable only on tomography. Children with Down syndrome should undergo regular comprehensive ophthalmological evaluation from early childhood, with particular attention to refractive error, strabismus, amblyopia, and keratoconus, and with adapted techniques for those with limited cooperation.

Author Contributions

Kh.U.Sh.: Writing – original draft; review & editing, Formal Analysis, Data collection and analysis, Formal Analysis. K.M.Kh.: Conceptualization, Analysis, Project Administration, Conceptualization, Writing – review & editing. All authors read the and approved the final version of the manuscript.

Conflict of Interest

The authors declare that they have no conflicts of interest that that could influence their opinion on the subject matter or materials described and discussed in this manuscript.

Disclaimers

The opinions expressed in this article are those of the authors and do not reflect the official position of the institution.

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Declaration of Generative AI and AI-assisted technologies in the writing process

During the preparation of this work the authors used an AI-assisted tool (Claude, Paperpal) for language editing and translation of source documents. All data were collected, verified, and interpreted by the authors; all AI-assisted content was reviewed, edited, and verified by the authors, who take full responsibility for the accuracy and integrity of the work.

Data Availability Statement

The datasets generated during and/or analyzed during the current study are available from the corresponding author on reasonable request.

Abbreviations

DS – Down syndrome, DSMIG – Down's Syndrome Medical Interest Group, IOP – intraocular pressure, US – ultrasonography, OCT – Optical Coherence Tomography.

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Рисунок до статті Vahobova M. Sh. et al. «Optimization of therapeutic approaches for meibomian gland dysfunction in women of reproductive age with dry eye disease»



Figure 1. Representative LacryDiag® diagnostic report of a patient from Group I (dry eye disease associated with meibomian gland dysfunction and vitamin D deficiency) before treatment. Abbreviations: DED, MGD, NIBUT, LLT, TMH.

Рисунки до статті Khamraeva U.Sh., Karimova M.Kh. «Comprehensive ophthalmological assessment in children with down syndrome: a cross-sectional study from Uzbekistan»

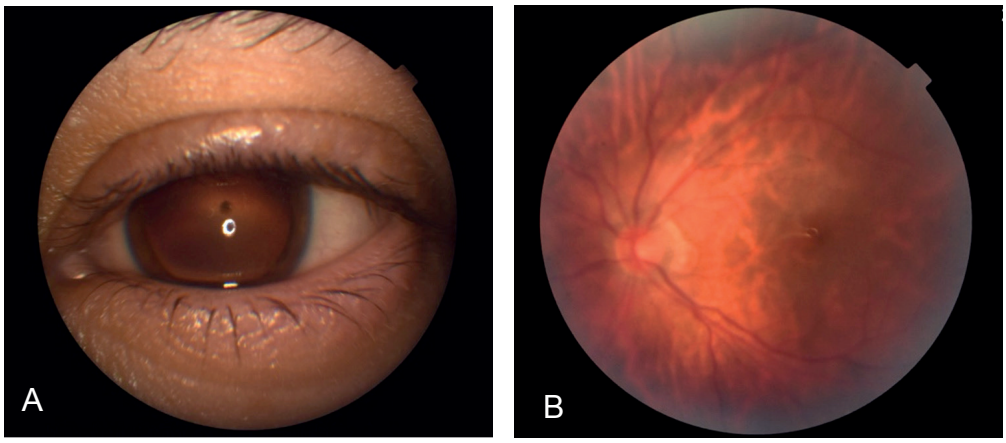


Fig 1. Anterior segment (A) and fundus (B) photo of the patient from the slit-lamp biomicroscopy.

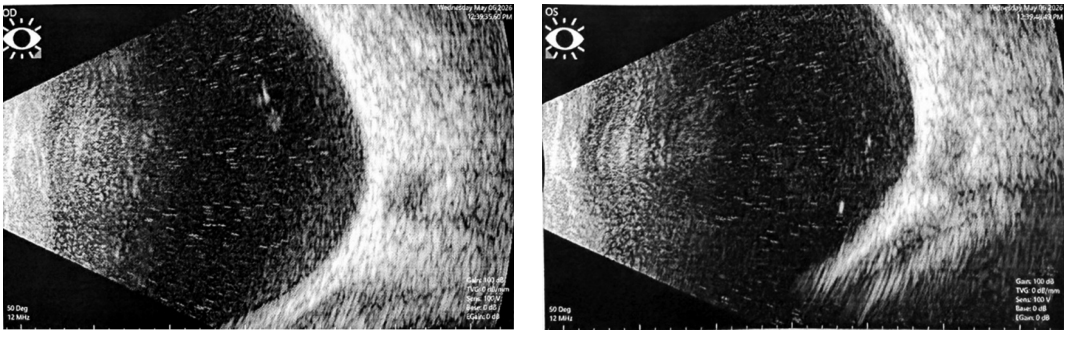


Fig 2. Ultrasonography B-scan of the patient with Down syndrome.

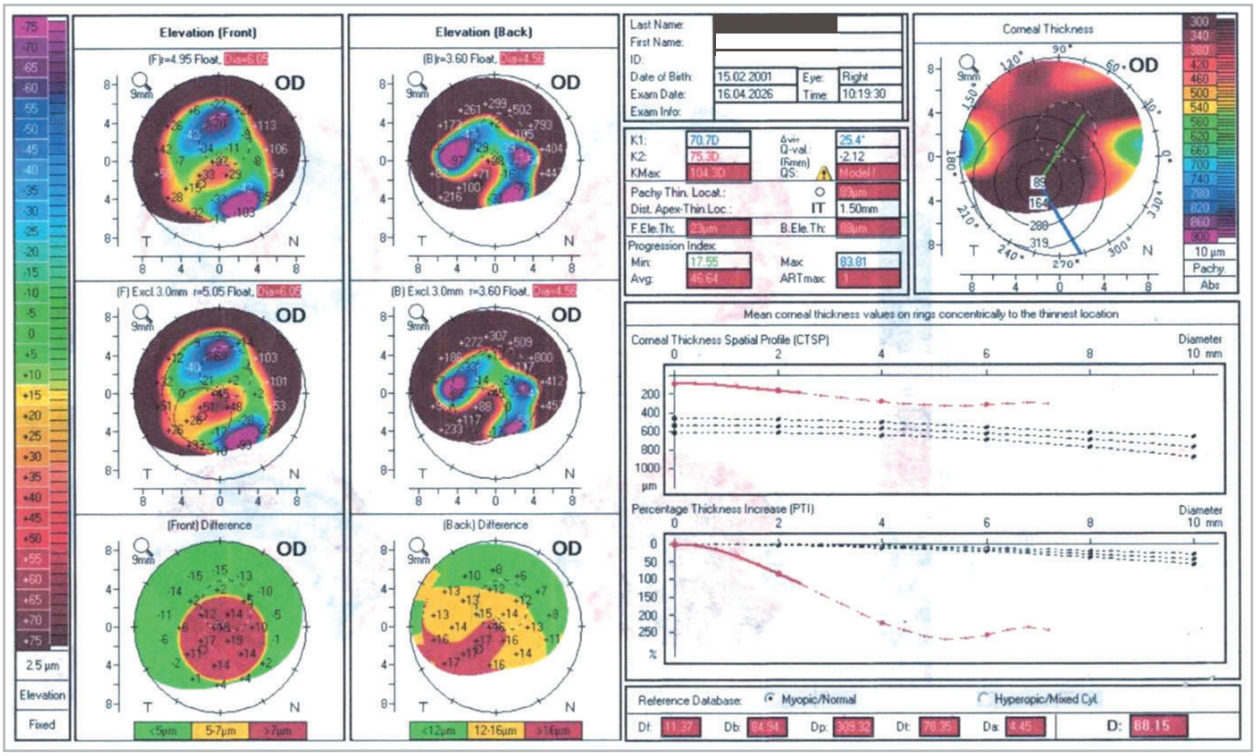


Fig 3. Belin-Ambrosio enhanced ectasia maps of corneal tomography.

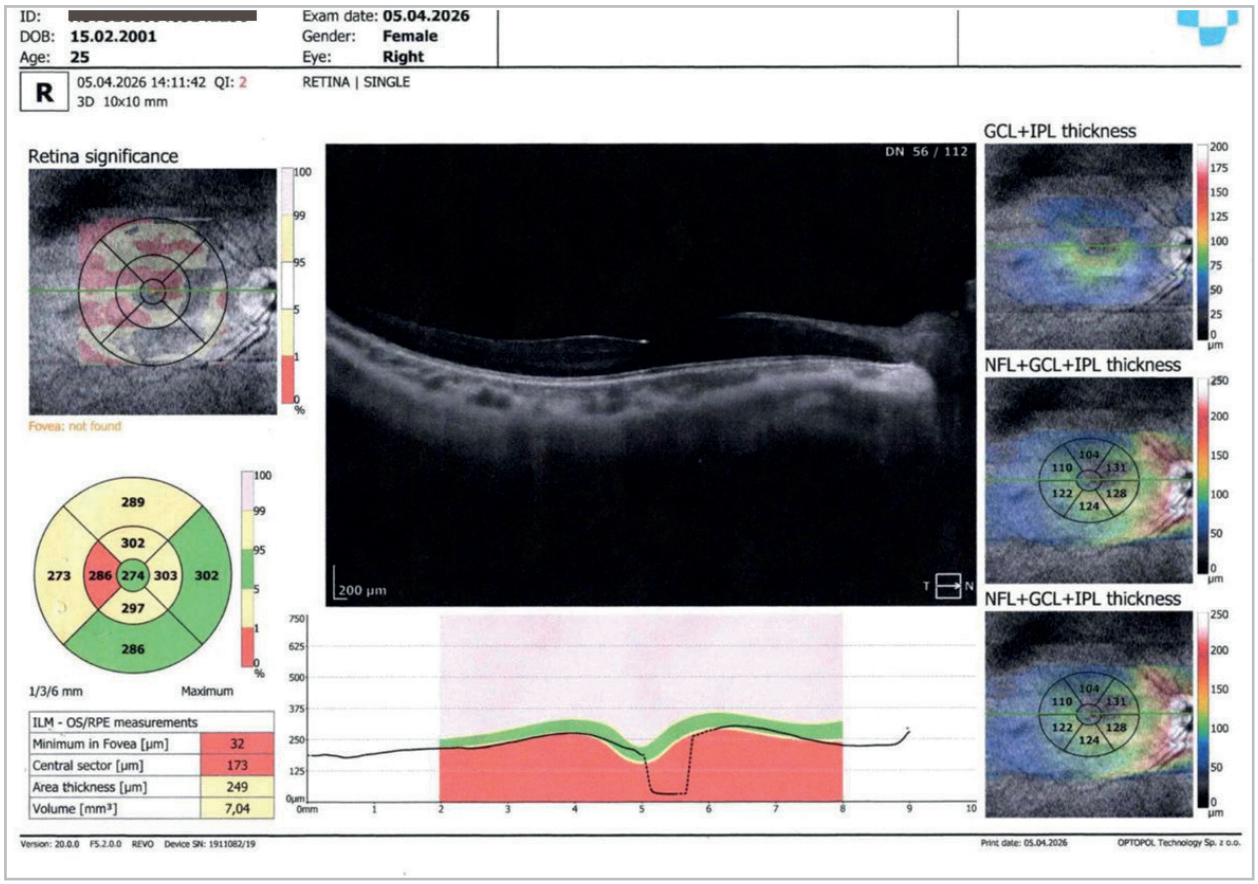


Fig 4. Optical Coherence Tomography (OCT) image of the patient with Down syndrome.