CASE REPORT

# Vision Therapy and Vision Rehabilitation in a Child with ABCA4 Retinal Dystrophy

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## ABSTRACT

### Background

Diagnosis of gradual onset visual impairment can be complicated, with some testing results inconclusive. Additionally, patients with visual impairment can have concurrent binocular vision and accommodative problems that need attention. We report successful application of vision therapy in a young patient to reduce asthenopia when reading. When visual acuity became more reduced, macular pathology was diagnosed and vision rehabilitation was provided.

### Case Report

An 18-year-old patient was followed for the last 10 years. During that time, she was successfully treated with vision therapy to alleviate blurred near vision and horizontal diplopia, which improved reading performance. Later, she required vision rehabilitation as her vision declined due to macular changes. Finally, the diagnosis of ABCA4, likely cone-rod dystrophy was made. Counseling the child on the visual diagnosis was managed uniquely due to significant concurrent life events that challenged routine patient education.

### Conclusions

Young patients with visual impairment also need to be assessed for binocular vision and accommodative disorders. Vision therapy and rehabilitation can be provided to these patients for best outcomes and enhanced quality of life. Diagnosis and delivering the serious news of vision loss can be complicated in pediatric populations. Families will take the lead in how and when the news is delivered to the child.

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**Keywords:** convergence insufficiency; inherited retinal disease; vision rehabilitation; vision therapy

## INTRODUCTION

Blurred vision, eyestrain, diplopia, and difficulty reading can present as symptoms of both accommodative and binocular vision disorders. Vision therapy is used to successfully treat convergence insufficiency and accommodative disorders.[1,2] Macular pathology can cause similar visual symptoms and should be ruled out before commencing vision therapy. It is important to realize that macular pathology and binocular/accommodative disorders occasionally occur simultaneously.[3] Vision therapy has been shown to be successful in patients with visual impairment.[4] We present a child who successfully underwent vision therapy for convergence and accommodative disorders. However, her intermittently blurred vision worsened

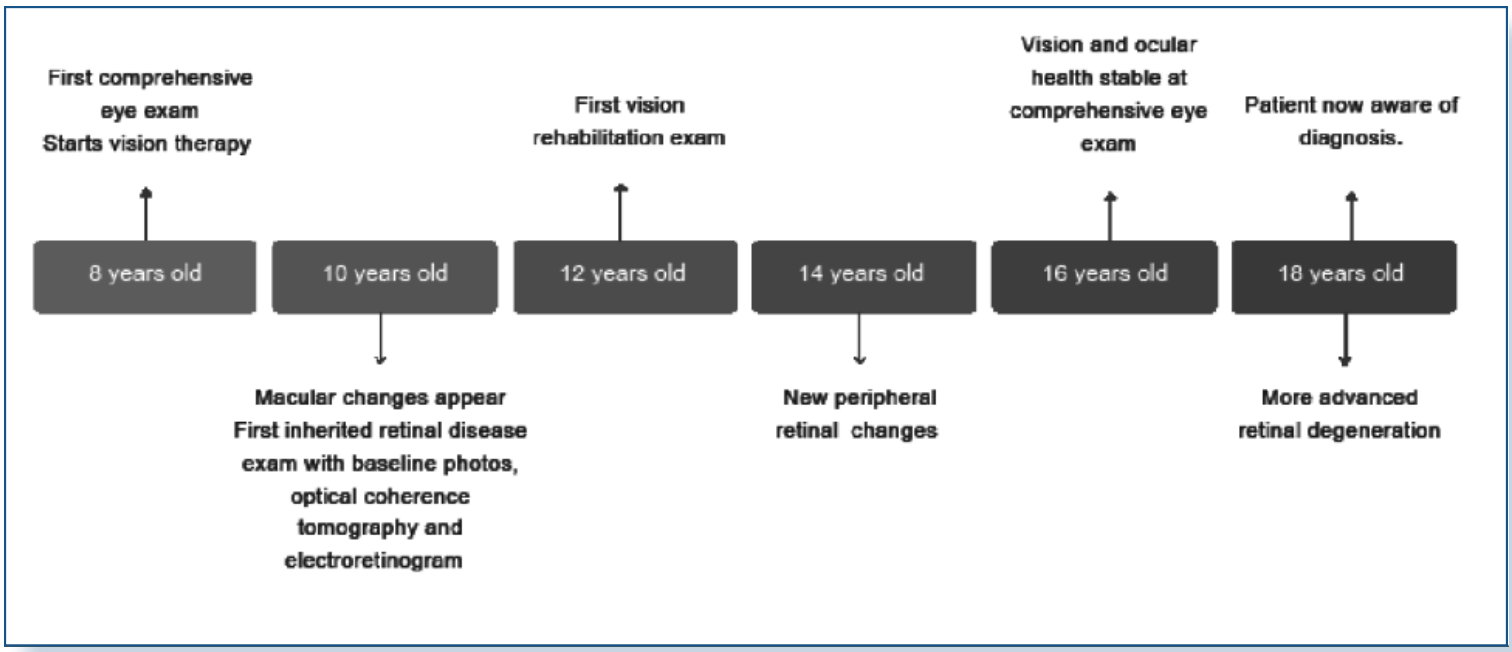



***Figure 1.*** Timeline of patient care.

and only later was she found to have an inherited retinal disease. Delivering serious news in the pediatric population is complicated and difficult. In this case, for several years after the patient's retinal diagnosis, her legal guardian declined to inform her of the diagnosis to allow her to deal with other major psychological trauma in her life. No identifiable health information was included in this case report.

## CASE PRESENTATION

Figure 1 provides an overview of this case report. An 8-year-old female presented to the Pediatrics/Binocular Vision Center of the Illinois Eye Institute complaining of reduced reading performance. She became sleepy when reading and occasionally experienced horizontal diplopia and blurred vision during near work. The child's medical history was unremarkable. There was a family history of macular degeneration causing vision loss in her maternal grandfather and maternal aunt before age 40. Her mother was seriously ill, contributing to a stressful home environment. Entering Snellen visual acuity with habitual spectacles for mild astigmatism was 20/40 right eye, 20/50 left eye at distance and 20/20 each eye at near. Pupils, extraocular muscles, and confrontation fields were normal. Cover test through habitual correction revealed orthophoria at far and 14 Δ intermittent alternating exotropia at near with low frequency and good control. Stereopsis was 20 sec. arc lateral disparity and 250 sec. arc global. Dry refraction yielded visual acuity of 20/25 in each eye at distance through +0.25 -1.00 x 180. Cycloplegic refraction with 0.5% cyclopentolate, 0.5% tropicamide and 2.5% phenylephrine spray revealed +1.25 -1.00 x 180 bilaterally. Intraocular pressures, anterior segment, and dilated fundus examination were unremarkable, with healthy optic nerves, cup/disc ratios of 0.3 and clear maculae. The dry refraction findings were prescribed for full-time wear; the patient was to return for 1 month follow-up.

The patient returned 4 months later, wearing her new spectacles. Aided acuities were 20/40+ at distance and 20/30 at near. Through these spectacles, cover test showed 2 Δ exophoria at far and 25 Δ intermittent alternating exotropia at near. Near point of convergence was 7 centimeters with alternating suppression and no recovery, using an accommodative target, and 7/10 centimeters (break/recovery) using a penlight with red lens. Worth 4-dot showed fusion at 40 centimeters and alternating suppression at greater distances (hence, central suppression based on smaller angular subtense of the target). Prism bar vergences at near measured x/10/4 for divergence and x/10/6

for convergence. Minus lens accommodative amplitudes were 4.00 right eye and 4.50 left eye. Accommodative facility binocularly using +/-2.00 D lens flippers showed suppression through plus (unfortunately accommodative facility was not tested monocularly). The Developmental Eye Movement test yielded normal results for the patient's age. Reduction in corrected distance acuity was attributed to over-use of accommodation to control the exotropia; thus, refraction was not repeated.

## VISION THERAPY

The patient was diagnosed with intermittent alternating exotropia at near, of the convergence insufficiency type. This diagnosis was based on the small exophoria at distance and the large intermittent exotropia at near with poor convergence along with symptoms of intermittent diplopia, blur, and sleepiness with close work. (Unfortunately, a Convergence insufficiency Symptom Survey (CISS) was not administered.) She also was diagnosed with accommodative insufficiency, based on low accommodative amplitudes. Fluctuations in best-corrected acuity were attributed at that time to the accommodative disorder. Vision therapy was recommended for these conditions. Full time use of glasses was recommended.

The vision therapy program utilized weekly in-office vision therapy supplemented with daily home therapy. For the first 3 months of vision therapy, the patient performed monocular accommodative activities intended to improve amplitude and flexibility (plus lens push-away, near/far rock, minus lens rock, plus/minus lens flippers). She experienced intermittent difficulty clearing plus lenses and seeing distance targets clearly. She also worked on gross convergence skills using binocular push-up activities with suppression monitoring, such as beads and string. Similar activities were prescribed for home. Participation in weekly office visits was excellent. Compliance with home therapy was moderate (home activities were completed 4-5 times per week); there was a great deal of stress at home due to the mother's illness. The patient's uncle arranged for professional tutoring in reading and math, which improved the child's school performance.

Vision therapy continued for 6 additional months, during which time the patient made slow progress despite moderately good compliance. In addition to accommodative activities, she worked on convergence and divergence using activities such as Vectograms, Aperture Rule, and free-space fusion cards (Bernell Corp., Mishawaka, IN). The patient's home situation became more stressful, as her mother passed away, and the patient was subjected to a custody battle between family members. Nevertheless, the patient reported that her initial diplopia and sleepiness had decreased. She continued to experience fluctuating distance vision with acuities ranging from 20/25- to 20/60 through habitual spectacles on different visits. Cover test showed orthophoria at far and 16Δ exophoria at near. Near divergence range was x/16/14; convergence range was 18/30/25. Pull-away accommodative amplitudes were 12.5 diopters in the right eye and 14 in the left. The vision therapy program ended due to the disrupted home situation and the patient's psychological distress. The patient received grief counseling.

The patient returned for comprehensive eye examination 3 months later, with corrected acuity of 20/50 in each eye at distance and 20/40 at near. Refraction provided no improvement. Cover test showed 2 Δ exophoria at far and $10^{\Delta}$ exophoria at near. Near point of convergence was 5/7 centimeters, using an accommodative target. Because of the inconsistencies in visual acuity testing, the patient underwent another thorough ocular health evaluation, including dilated fundus examination. Ocular health findings, remained completely normal. Psychogenic vision loss was suspected, based on tubular results on tangent screen visual fields. Follow-up in 2 months was recommended with electrodiagnostic testing to be considered.

At a follow-up two months later (age 10) best corrected distance acuities were 20/40 right, 20/70 left. Near point of convergence was to nose; near divergence range was x/16/14, convergence was x/25/16. Push-up accommodative amplitudes

measured 8 diopters in each eye. For the first time, the fundus examination revealed mild macular retinal pigmented epithelium atrophy and flecks in the posterior pole bilaterally. Optical coherence tomography demonstrated retinal pigmented epithelium disruption in both maculae. The patient was referred for an inherited retinal disease examination and vision rehabilitation. Due to the psychological trauma the patient was experiencing from her family situation, her uncle (now her legal guardian) did not want her to be told of her retinal diagnosis or prognosis. We believed the macular disease had been developing for some time but had not previously been evident on fundus examinations.

## INHERITED RETINAL DISEASE EXAMINATION

The inherited retinal disease examination at the Illinois Eye Institute included vision loss history, pedigree analysis, dilated fundus examination, electroretinogram, and review of recent optical coherence tomography. History indicated reduced visual acuity, but the patient denied photophobia, nyctalopia, scotoma, or color vision problems. Pedigree analysis showed likely autosomal recessive juvenile macular degeneration, with both maternal aunt and maternal grandfather losing central vision before age 40. Dilated examination revealed atrophic macular changes with drusenoid-like changes throughout the posterior pole; the peripheral retina was unaffected. The electroretinogram was not well tolerated but did show abnormal scotopic and photopic responses. Optical coherence tomography review confirmed retinal epithelium disruption and thinning of the macular region.

Following the inherited retinal disease examination, there was a gap in care for 1.5 years. During that time, five phone conversations were documented regarding the diagnosis and options. Subsequently, the patient returned for comprehensive eye examination. At that visit, her best corrected visual acuity was 20/70 in each eye, and the family consented to having a Vision Rehabilitation consultation.

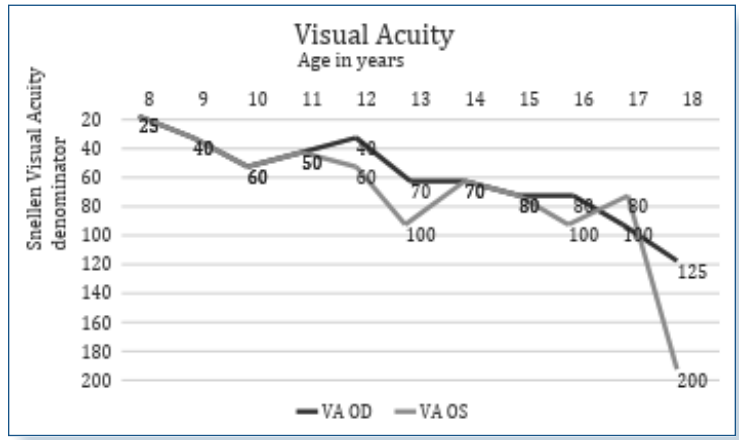



***Figure 2.*** Best corrected distance visual acuity showing the gradual decline as patient ages.

## VISION REHABILITATION

The patient was seen five times in the Vision Rehabilitation clinic over the course of the next 6.5 years. Figure 2 shows the gradual decline in her visual acuity over the course of her care. Ancillary testing was difficult to obtain due to the legal guardian's hesitation. The patient continued to engage in psychological counseling at school and family counseling outside school. The primary concern was blurry distance vision. She reported no fatigue with near work. It is possible that her previous work in vision therapy had helped with reading ability. Her grades were above average at a gifted school with no accommodations. She was best corrected to 20/70 in each eye at distance and .4/2M at near. Her habitual near working distance was recorded as 20 centimeters; she pulled material closer to take advantage of relative distance magnification. The predictions for devices were 3.5x distance magnification and +10 D equivalent power at near. Trials of optical and electronic magnification found a preference for a 3.25x25 handheld telescope (20/25 vision), 2.1x mounted telescopes, and +4 D reading glasses with base in prism (.25/.8M). Although the patient did not report near fatigue at the initial Vision Rehabilitation examination, it is common for children with visual impairment to have reduced accommodation and to pull reading material closer to gain a larger retinal image size.[5,6] These two factors can lead to trouble with reading. The

+4 reading glasses were accepted by the patient and put in place as her visual acuity was expected to get worse and near fatigue to increase. These devices were dispensed, and a school report was completed recommending an Individual Education Plan with a Teacher of the Visually Impaired, front row seating, testing accommodations, Orientation and Mobility consultation, and assistive technology.

At the next visit 1.5 years later, the patient's main concern was continued blur at distance with new concerns of fatigue after 20 minutes of near work, but no diplopia. She denied any problems with glare or mobility. School performance was stable. She reported having the Individual Education Plan with the previous recommendations in place. Refraction was stable and distance acuities were now 20/80 in each eye with .2/1M at near. Her magnification devices were still appropriate. Bar magnifiers with an equivalent power of +6 diopters were added to help her read printed material. Accommodation and confrontation visual fields were normal. Dilated fundus examination showed continued atrophic degeneration of macular area and new findings of diffuse bilateral peripheral temporal retinal pigment clumping and dropout. Optical coherence tomography results were like previous, showing expanding atrophic outer retina. The legal guardian deferred additional testing.

The patient returned 13 months later with continued concern of blur. She denied nyctalopia. She reported that her devices were helpful and she benefited from front row seating and testing accommodations. Best corrected vision was 20/80 in each eye at far and .2/1.6M at near. She was now an 8th grader and doing well in school. Refraction was stable, accommodation normal and confrontation visual fields full. Cover test showed orthophoria at far and 20Δ constant alternating exotropia at near. As her visual acuity reduced with increasing macular changes, her fatigue with reading at near was increasing. She continued to maintain a 20-25 centimeter near working distance and use

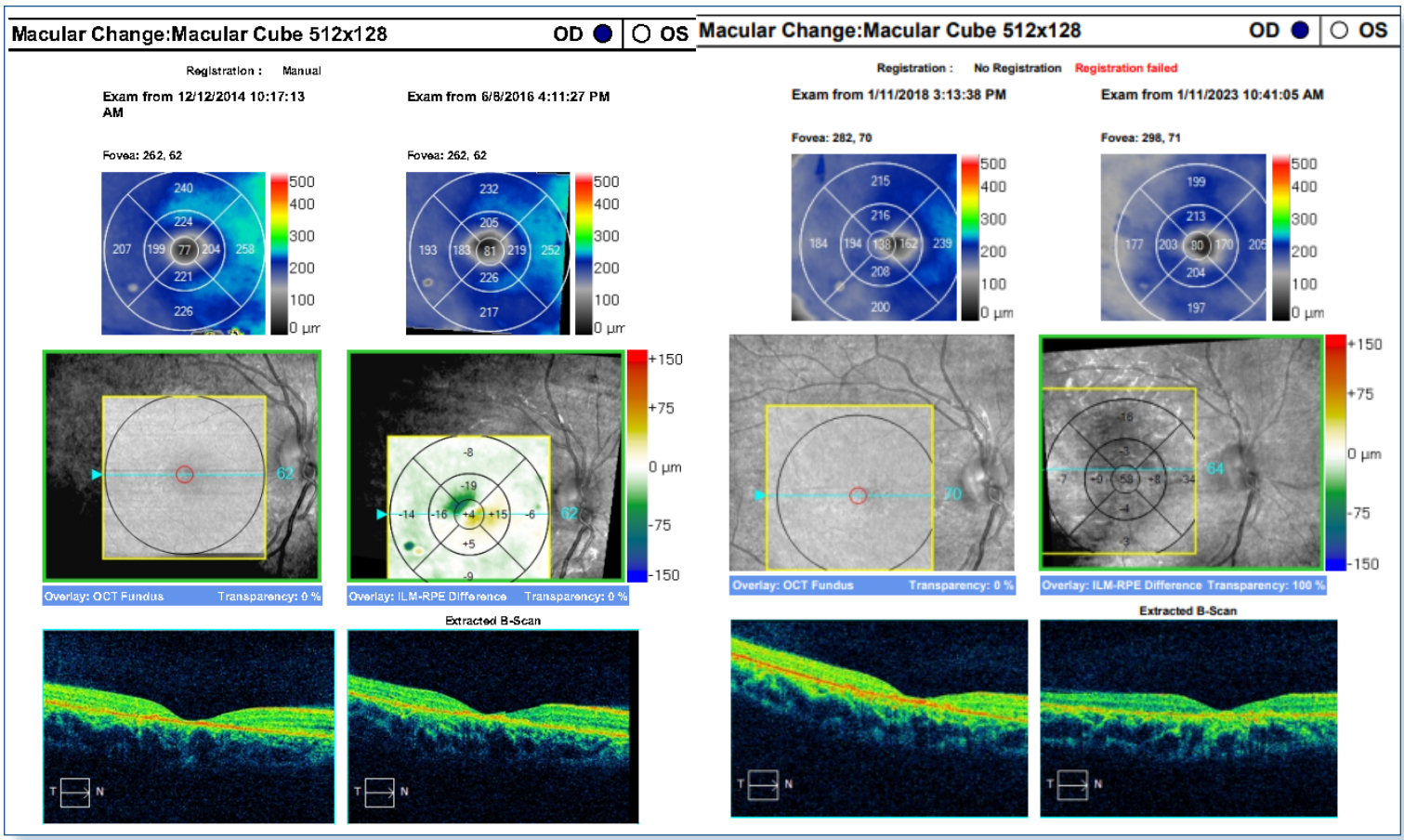



***Figure 3.*** Serial images of the ganglion cell analysis of the macular cube from 2014 to 2023 of the right eye. Significant thinning was evident in 2014 at 77 microns and was stable to 80 in 2023. Foveal pit anatomy shows atrophy. Eccentric viewing by patient may have caused slightly eccentric mapping of the fovea.

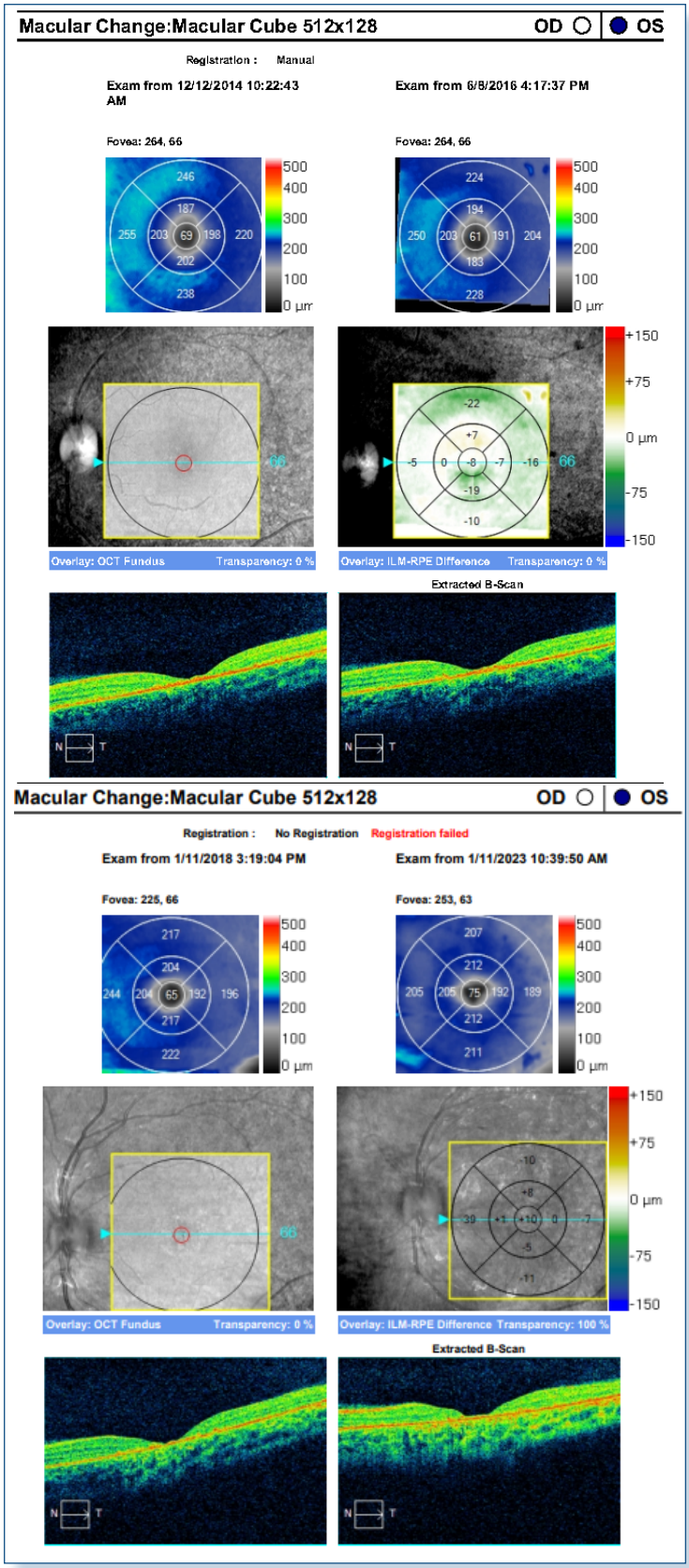



***Figure 4.*** Serial images of the ganglion cell analysis of the macular cube from 2014 to 2023 of the left eye. Significant thinning was evident in 2014 at 69 microns and was stable to 75 in 2023. Foveal pit anatomy shows atrophy. Eccentric viewing by patient may have caused slightly eccentric mapping of the fovea.

the +4 reading glasses. To help compensate, print size was enlarged on her screens (phone/tablet/computer) to enable her to read. She also started to prefer white print on a black background for visual reading. Additionally, likely the poor acuity and close working distance required when using high plus reading glasses had led to a return of this large exo posture. Fortunately, she did not experience diplopia due to suppression. Dilated fundus examination revealed consistent results with both central and peripheral degenerative changes. Recommendations included: prescription glasses, updated school report for services, continued use of devices; additional resources were discussed including technology and audio learning. The legal guardian deferred additional ocular health testing.

The most recent visit occurred at age 18. The patient's main concern was whether she could obtain a driver's license. She was a high school senior, class president and in the top 5% of her class. She was active in many extracurricular activities including dance and theater. She still used her optical magnification devices occasionally but relied more on assistive technology consisting of enlargement and contrast enhancement while using computers and her smartphone. The family was now openly discussing the diagnosis of an inherited retinal disease and was open to genetic testing.

Refraction was stable and best corrected vision at distance was reduced to 20/125 right and 20/200 left with .23/2M at near. She had significant loss of log contrast sensitivity to 0.64 right, 0.52 left and 0.57 both eyes. Confrontation visual fields remained full. Dilated fundus examination revealed consistent results. Optical coherence tomography, seen in Figures 3 and 4, found thinning of the ganglion cell layer and macular atrophy consistent with ABCA4.[7] Fundus photos, seen in Figure 5, showed diffuse macular atrophy, mildly attenuated vessels and granular hypopigmentation to mid-periphery and temporal pigment clumping. All these findings are consistent with severe ABCA4.[8,9] A buccal genetic sample was collected. Recommendations included: prescription glasses, updated school report, continued use of devices, discussion of accommodations and services at

the university level. There was a discussion about driving and not meeting the state requirements. Alternative transportation services were discussed. The plan was to follow up in the clinic after the genetic results were received.

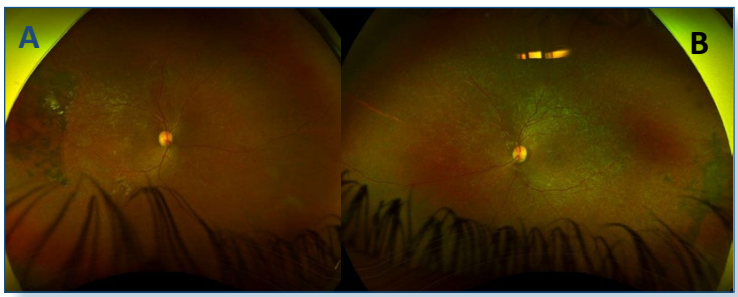



***Figure 5.*** Fundus photograph of (A) right and (B) left eyes at age 18 showing mildly attenuated vessels, macular atrophy, widespread granular hypopigmentation of the retinal pigment epithelium and pigment clumping temporally.

Genetic testing was done using the My Retina Tracker® program through the Foundation Fighting Blindness, revealing that the patient is heterozygous for ABCA4 c.4954T>G, p.(Tyr1625Asp). Reviewing this genetic variant in NIH National Library of Medicine, National Center for Biotechnology Information, ClinVar, it is classified to be pathogenic.[10] In review of all clinical testing, this case appears to be ABCA4 retinal dystrophy leaning towards a cone-rod dystrophy. The patient was contacted multiple times for follow-up but has not yet returned to the clinic.

## DISCUSSION

Convergence insufficiency is a common binocular vision disorder in which the patient shows an exodeviation greater at near with inadequate convergence ability for near work. Common symptoms include blurry vision, diplopia, difficulty reading, and eye strain. All these symptoms were evident with our patient. This condition responds well to vision therapy.[11,12] In this case, vision therapy did help but ultimately could not address the fluctuating visual acuity. It was considered during the vision therapy program that her fluctuating near vision might be attributed to an accommodative disorder, with overaccommodation potentially occurring secondary to the large near exodeviation and her efforts to achieve fusion.[13] Nonorganic vision loss from psychosocial stress can also cause vision loss in children.[14,15] This was considered as our patient had the significant psychological trauma of the illness and death of her mother. Treatment for nonorganic vision loss includes mental health counseling, which the patient was already actively receiving. Although ocular disease had also been suspected due to fluctuating visual acuity, there was no funduscopic evidence for many months. In retrospect, OCT could have been performed sooner, but at the time of diagnosis, OCT was not commonly used for children due to lack of access and lack of normative data.

When managing suspected vision loss, it is critical to collect data to confirm the diagnosis. Clinical data may be ambiguous, or in this case, difficult to obtain. In our case there were two barriers: the legal guardian's reluctance to allow additional clinical testing and the nature of the child's condition changing over time, making it difficult to confirm or verify the diagnosis. Since there was no ocular health emergency, the family's decision was respected. We carefully followed the child, supported the family, and encouraged ongoing mental health therapy.

When inherited retinal disease is suspected, clinical data collected should include family pedigree, history with emphasis on pattern of visual loss, visual acuity, contrast sensitivity, color vision, visual field including far peripheral with kinetic perimetry and central with microperimetry, optical coherence tomography, fundus photos, electrophysiology, genetic testing and counseling. In this case, testing supported a diagnosis of inherited retinal degeneration likely involving both cones and rods. The genetic testing panel initially reported a variant of uncertain significance, but upon further investigation, the variant was documented to be pathogenic. More thorough genetic testing, beyond the free testing available, may provide more information. Other testing was deferred by the legal guardian. Ideally, repeated electrophysiology and visual field testing would be helpful for clinical diagnosis.

Once a diagnosis has been determined it is important to keep apprised of emerging treatments

and clinical trials in which a patient may opt to participate if they are eligible. For ABCA4 retinopathies, there are multiple treatments being studied including gene therapy and visual cycle vitamin A modulation. Both treatments are currently in clinical trials.[16]

One study, "A Phase 2 Multicenter, Double-Masked, Randomized, Placebo-Controlled Study to Investigate the Long Term Safety, Tolerability, Pharmacokinetics and Effects of ALK-001 on the Progression of Stargardt Disease" is referred to as the TEASE study. It is evaluating modifying the vitamin A cycle to prevent retinal degeneration. The investigational drug ALK-001, Gildeuretinol Acetate, is a deuterated vitamin A derivative and is being administered orally for 24 months versus a placebo at eight study locations.[17] Another study also investigating the vitamin A cycle is "Phase 3, Multicenter, Randomized, Double-Masked, Placebo-Controlled Study to Evaluate the Safety and Efficacy of Tinlarebant in the Treatment of Stargardt Disease in Adolescent Subjects" and is referred to as the DRAGON study. This study is investigating the oral drug tinlarebant versus a placebo for 24 months to slow retinal lesion growth. The DRAGON study has sites worldwide.[18]

A study investigating gene therapy is entitled "ACDN-01-001: Open-Label, Single Ascending Dose Study to Evaluate the Safety, Tolerability, and Preliminary Efficacy of Subretinal ACDN-01 in Participants With ABCA4-related Retinopathy." This study is referred to as STELLAR and is currently in recruitment at 10 locations with a study duration of 5 years. It seeks to evaluate the use of ACDN-01, an investigational RNA exon editor gene therapy, to restore full length ABCA4 mRNA in patients with ABCA4-related retinopathies.[19]

Low vision rehabilitation has been shown to maximize visual function and improve quality of life in the pediatric population.[20,21,22] During our patient's visits in the low vision rehabilitation clinic, magnification, and assistive technology were provided. Additionally, she was connected to her school's vision services and taught the importance of self-advocacy.

The legal guardian requested that the diagnosis of vision loss not be discussed with the patient. During the visits, no retinal diagnosis was discussed; communication included simply that her eyes needed a little more help including magnification and technology. The child never questioned what was done in the examinations or any of the recommendations. Sometime between the final two visits, the patient had been told about her diagnosis and by age 18 she appeared well-adapted and was thriving. Her guardian confirmed that she was doing well at school, socially, and at home.

A clinician relies on their background, experiences, and training to develop patient communication skills. Importantly, clinicians need to be aware of protocols that have been developed to deliver serious news. Guidelines from these protocols can be tailored to each situation. This will lead to productive interactions and increased ability of the patient to understand their diagnosis and participate in their ongoing care. It is also important to remember that families will reference their own culture, beliefs, and values when considering additional testing and telling their children about the diagnosis. When involving a child, first deliver the serious news to the parent/guardian, who should decide how to deliver the news to the child.

Several well-known communication protocols for delivering serious news have been developed, including SPIKES[23], ABCDE[24] and BREAKS.[25] Common themes amongst these protocols include creating a private, quiet space and allowing enough time for the conversation in person. Explore what the family understands and provide clear information without medical jargon so it can be digested. Provide as much information as the family wants, without overwhelming them. Acknowledge and validate the family's emotional responses with empathy. Provide resources and next steps to the family, a roadmap of where they are going. How the provider delivers the serious news of vision loss can have a lasting impact on how the patient accepts their visual impairment.[26]

## CONCLUSION

This case illustrates the importance of treating the visual problems that are diagnosed and being aware of changes in vision that uncover additional problems requiring management. This case shows that binocular vision and accommodative problems can coexist and be successfully managed in patients that also have visual impairment. The case also highlights the challenges of delivering serious news in the pediatric population, while being sensitive to a family's desire to control the information, helping the pediatric patient to understand and accept the information when they are ready. Our patient received continued mental health counseling and vision rehabilitation and currently is a thriving university student.

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Dr. Matchinski graduated from the Illinois College of Optometry in 1995 and completed a Low Vision Rehabilitation residency at the William Feinbloom Low Vision Rehabilitation Center at the Pennsylvania College of Optometry. Upon completion of her residency, she returned to Chicago and has been providing clinical vision rehabilitation care at both the Illinois Eye Institute's Rosenbloom Center and The Chicago Lighthouse. She is the inaugural recipient of the Rosenbloom Family Professor in Low Vision Rehabilitation at ICO. Dr. Matchinski has published and lectured both nationally and internationally on Low Vision Rehabilitation and teaches the vision rehabilitation course at ICO. She has held chair positions in the ASCO Low Vision Educators Special Interest Group, AAO Low Vision Diplomate Program and AAO Low Vision Section. She has also been president of both VOSH Illinois and VOSH International.