Degenerative Myopia: A Case Series Report
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ABSTRACT

Background: Degenerative myopia is identified by excessive and progressive elongation of the globe in combination with refractive error of greater than -6.00D and an axial length greater than 26mm. Though it has long been debated whether school myopia is of solely genetic origin, influenced by environmental factors, or a mixture of the two, degenerative myopia is thought to be more genetic in cause.

Case Study: This case series was conducted on a family of seven degenerative myopes, comparing clinical findings as well as A-scan and visual evoked potential (VEP) alternating pattern testing. Data was gathered from the most recent examinations to determine the most common degenerative myopic findings within the family unit. VEP alternating pattern testing was conducted with best-corrected spectacle lenses at time of testing. Only one subject was tested before and after being fit with contact lenses.

Conclusion: Upon clinical comparison of the seven family members, genetic inheritance appeared to be the most obvious causative factor of myopia progression in this family. Though it is still debated whether environmental factors take part in the early phases of degenerative myopia, heredity has been found to be the dominant predicting factor for the extent of the myopic changes. Further testing through a larger scale study is warranted to determine whether contact lens wear improves VEP amplitude and latency recording when compared to spectacle wear in degenerative myopia patients.

Keywords: A-scan, degenerative myopia, progressive myopia, visually evoked potential

Introduction

Degenerative myopia (i.e., progressive or pathologic myopia) has been a topic of scientific debate for many years. Aristotle (c. 384-322 BC) was the first to recognize and to describe the difference between short and long sight. He believed that animals with prominent eyes did not see well when looking at a distance, but animals with smaller, more recessed eyes were better adapted to see far away because movement was able to take a straighter course and was not scattered into space. Other philosophers through the ages have attempted to explain the reasons for myopia, including Kepler (1611), who deducted that occupation influenced refraction. It was not until 1854 that Von Graefe would correlate refractive error and the pathology of degenerative myopia.¹

The current definition of high myopia is an eye with a refractive error of greater than -6.00D and an axial length of the globe greater than 26mm.² Approximately 0.5% of the general population is affected, and of those with myopia, 30% have been found to have high myopia.² Degenerative myopia is identified by excessive and progressive elongation of the globe in combination with the above characteristics of high myopia. Excessive elongation of the globe is often associated with secondary changes involving the retina, choroid, optic nerve head, and sclera. Most common clinical findings often include a pale, tigroid fundus, chorio-retinal atrophy, lacquer cracks, choroidal neovascularization, sub retinal

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hemorrhages, and Fuchs spot. Complications include staphyloma, retinal detachment, foveal retinoschisis, and peripapillary detachment.\(^1\,^2\) Maculopathy is the most common cause of visual loss.\(^2\)

The exact cause of myopia is yet to be determined. It is believed that both environmental factors (near work, schooling, lifestyle, and outdoor activities) and genetic factors play a role in influencing myopic changes. Recent studies suggest that specific genes regulate scleral remodeling, a crucial factor in axial elongation, and may be the key to high, progressive myopia.\(^3\,^4\)

**Case Series Report**

The following case series was conducted on a family of seven high myopes (six siblings and their mother) in order to study correlations of degenerative myopia within a family unit. Goals were to determine similarities in best-corrected VEP measurements between family members and to discuss current literature on the etiology of and treatment options for degenerative myopia.

The following is a brief overview of each patient’s most recent pertinent clinical findings. Findings not noted are to be considered as normal (Table 1).

**Patient 1**

Patient 1 had been fit in contact lenses previously and was being followed by low vision and contact lens specialists. She had been diagnosed with degenerative myopia, blindness/low vision in the better eye and moderate impairment in the lesser eye.

**Patient 2**

Patient 2 had been fit with contact lenses one year prior and was also being followed by contact lens specialists. She had been previously diagnosed with progressive high myopia.

**Patient 3**

Patient 3 had previously been diagnosed with progressive high myopia. He presented for his one-week follow-up for finalization of his contact lens prescription. Visual acuity remained stable.

<table>
<thead>
<tr>
<th>Table 1: Comparison of Patient Data</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Patient number</strong></td>
</tr>
<tr>
<td>---------------------</td>
</tr>
<tr>
<td>1</td>
</tr>
<tr>
<td>2</td>
</tr>
</tbody>
</table>

(continued)
<table>
<thead>
<tr>
<th>Patient number</th>
<th>Patient Demographics</th>
<th>Entering Findings</th>
<th>Spectacle Rx</th>
<th>Contact Lens Rx</th>
<th>Anterior Segment</th>
<th>Posterior Segment</th>
<th>Axial Length (A-Scan)</th>
</tr>
</thead>
<tbody>
<tr>
<td>3</td>
<td>13yo African American Male</td>
<td>VA: sc Distance OD: 20/400 OS: 20/400 Near OU: 20/100 IOP: OD: 18mmHg OS: 20mmHg</td>
<td>OD: -9.50-3.00x180 OS: -9.50-4.00x007 VA: Distance OD: 20/25+ OS: 20/25- OU: 20/25</td>
<td>Coopervision Biofinity Toric (8.7/14.5) OD: -8.00-1.75x170 OS: -8.00-1.75x180 VA: Distance/Near OD: 20/25 OS: 20/25</td>
<td>Negative pertinent findings</td>
<td>Disc: choroidal crescent, malinsertion C/D: 0.5x0.5 OD,OS Retina: flat Macula: flat and intact, +FLR Periphery: (-) holes, tears, or retinal detachment OD,OS</td>
<td>OD: 26.39mm OS: 26.74mm</td>
</tr>
<tr>
<td>4</td>
<td>12 yo African American Female</td>
<td>Entering VA through habitual Spec Rx OD: -11.50-0.50x060 OS: -8.75-0.75 x 150 Distance OD: 20/125 OS: 20/40 Near OD: 20/200 OU: 20/80 OU: 20/70 IOP: OD: 11mmHg OS: 12mmHg</td>
<td>OD: -11.50-0.50x060 OS: -9.00-1.00x160 VA: Distance OD: 20/100 OS: 20/20</td>
<td>Coopervision Biofinity OD: -10.00sph (8.6/14.0) OS: -8.00-0.75x160 (8.7/14.5) VA: Distance/Near OD: 20/150 OS: 20/30 OU: 20/30 Near OD: 20/50 OS: 20/25+ OU: 20/25</td>
<td>Negative pertinent findings</td>
<td>Disc: pink/distinct C/D: OD: 0.2x0.2 OS: 0.25x0.25 Retina: flat Macula: flat, +FLR Periphery: flat, intact, (-) holes, tears x 360</td>
<td>OD: 27.03mm OS: 26.38mm</td>
</tr>
<tr>
<td>5</td>
<td>11 yo African American Female</td>
<td>VA: cc through habitual Spec Rx OD: 20/400 OS: 20/50 OU: 20/50 Near OD: 20/100 IOP: OD: 15mmHg OS: 14mmHg</td>
<td>Habitual Spec Rx: OD: -18.25-2.50x180 OS: -10.00-1.00x180 Final Spec Rx: OD: -19.00-2.50x180 OS: -11.00-1.00x180 +2.50 add VA: Distance OD: 20/400 OS: 20/30 OU: 20/30 Near OU: 20/25</td>
<td>Proclear XR multifocal OD: -15.50-1.25x180 +2.00 add OS: -10.00 +2.00 add VA: Distance OD: 20/150 OS: 20/30 OU: 20/30 Near OD: 20/50 OS: 20/25+ OU: 20/25</td>
<td>Conjunctiva mild injection bulbar and palpebral OD, OS</td>
<td>Disc: malinsertion OU C/D: OD: 0.45x0.45 OD,OS Retina: degenerative myopia changes, tigroid fundus Macula: flat, +FLR Posterior pole: OD: staphyloma around ONH Periphery: operculum at 9 o’clock OD</td>
<td>OD: 29.67mm OS: 26.33mm</td>
</tr>
<tr>
<td>6</td>
<td>9 yo African American Female</td>
<td>VA cc: Distance OD: 20/50 OS: 20/40 OU: 20/40 Near OD: 20/25+ OS: 20/25+ OU: 20/25+ IOP: 12mmHg OD,OS</td>
<td>Habitual Spec Rx: OD: -16.50-1.00x180 OS: -13.25 Final Spec Rx: OD: -16.50-1.00x180 OS: -13.25 No change in spec Rx</td>
<td>To be considered at future appointment</td>
<td>Negative pertinent findings</td>
<td>Disc: pink/distinct C/D: OD: 0.3x0.3 OD,OS Retina: flat, intact Macula: flat, +FLR Periphery: (-) holes, tears x 360, white without pressure OU inferior</td>
<td>OD: 27.55mm OS: 27.23mm</td>
</tr>
<tr>
<td>7</td>
<td>9 yo African American Male</td>
<td>VA cc: Distance OD: 20/20- OS: 20/25+ OU: 20/20 Near OD: 20/20 OS: 20/20 OS: 20/20 IOP: OD: 17mmHg OS: 18mmHg</td>
<td>Habitual Spec Rx: OD: -10.50-1.50x160 OS: -11.00-0.50x060 Final Spec Rx: OD: -11.00-1.50x165 OS: -11.00-0.50x065 VA: Distance OD: 20/20 OS: 20/20 OU: 20/20</td>
<td>To be considered at future appointment</td>
<td>Negative pertinent findings</td>
<td>Disc: pink/distinct Choroidal crescent OU C/D: 0.35x0.35 OD/OS Retina: tigroid funds Macula: flat, intact, +FLR Periphery: flat, intact x 360, (-) holes/ tears, white without pressure temp and nasal OD, white without pressure nasal OS</td>
<td>OD: 28.67mm OS: 28.10mm</td>
</tr>
</tbody>
</table>
published indicating cataract formation to be a further complication of myopia.\textsuperscript{5,7}

Visually evoked potential (VEP) testing was performed under binocular and monocular viewing conditions on each family member in order to compare and contrast findings within the family. Testing was performed on the Diopsys unit with medium contrast. Phase alternating checkerboard patterns were used with recordings taken at 8x8, 16x16, 32x32, and 64x64 check sizes. Patients 2, 3, 4, 6, and 7 demonstrated normal VEP recordings, with findings ranging from zero to moderate binocular summation.

Patient 1 had decreased amplitude findings at all checker sizes when viewing through her left eye. She also demonstrated late latency findings while viewing the 8x8 checkerboard size monocularly with both the right and left eye. Latency findings were also delayed while the patient was viewing the 16x16 checkerboard pattern monocularly with the right eye.

Patient 5 was tested before the initial contact lens fit while wearing her spectacle prescription and then again a month later while the multifocal contact lenses were in place. Findings revealed increased amplitude, as well as improved latency with contact lenses binocularly and monocularly during several of the recordings. Two of the initial right eye recordings were determined to be unreliable due to reduced amplitude during the 8x8 and 16x16 measurements. Moderate binocular summation was noted during most of the size recordings throughout testing.

Table 2 represents the normalized values of latency recordings for patient 5 with spectacle correction on February 1, 2012 and then with multifocal contact lens correction on February 27, 2012.

**Discussion**

Extensive genetic testing has been performed in an attempt to identify the inheritable factor causing myopia. Research indicates that high
myopia may be inherited as an X-linked recessive, autosomal recessive, or autosomal dominant trait. At this time, multiple genetic myopia loci have been detected and mapped, ranging from single-nucleotide polymorphisms relating to collagen make-up to genetic predisposition for increased testosterone levels. Genetic testing has not identified a specific myopia gene, but literature regarding the intricate interplay of the genetic factors contributing to scleral remodeling has been published. During the development of myopia, active remodeling occurs in the sclera. Research indicates that specific genes involved in the degradation of the extracellular matrix may be the inheritable factors leading to globe elongation and essentially myopia. Publications both for and against this theory have been released; therefore, more research is needed before a specific inheritance factor can be determined.

It is often a difficult task to delineate between degenerative myopia and simple myopia. Though degenerative myopia is more likely to be genetically driven, there may be environmental factors that impact the progression and development of the condition. Simple myopia, further classified as school myopia (onset during school age and stabilizing around 15-17) and adult-onset (which develops in young adults), may be driven by multiple factors. Several theories based on the disruption of the emmetropization process have been proposed. One such theory suggests that excessive accommodation may result from overacting intraocular muscles and can influence emmetropization. Such theories are supported by research that has been conducted on Eskimo populations after the introduction of Western educational systems, as well as Asian studies that have followed the refractive error of students undergoing rigorous educational programs emphasizing extended near work activities. Based on these studies, culture, education, and increasing near demand on students seem to influence myopia prevalence.

Animal studies focusing on form deprivation have concluded that when eyes are sutured or occluded (translucent), axial elongation and myopia will occur. Growth due to image degradation appears to be mediated by the retina and occurs in the sclera. Scleral research has stemmed further biochemical research attempting to identify the exact chemical cascade controlling scleral growth. Studies involving dopamine, fibroblast growth factor, muscarinic antagonists, retinal dehydrogenase, and many more chemical factors have been performed. Unfortunately, not one specific factor has been identified as the primary contributor to scleral control during axial elongation and the development of myopia. With continuing research, pharmaceutical options may be possible for the myopic patient in the near future.

### Table 2: VEP Comparison of Spectacle vs. Contact Lens Wear for Patient 5

<table>
<thead>
<tr>
<th>Size</th>
<th>Eye</th>
<th>Amplitude</th>
<th>Normative percentile through contact lenses</th>
<th>Amplitude</th>
<th>Normative percentile through contact lenses</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td></td>
<td></td>
<td>Feb 1, 2012</td>
<td></td>
<td>Feb 27, 2012</td>
</tr>
<tr>
<td>8x8</td>
<td>OU</td>
<td>12.8</td>
<td>73.56%</td>
<td>25.6</td>
<td>86.49%</td>
</tr>
<tr>
<td>8x8</td>
<td>OD</td>
<td>Unreliable initial recording 2.1</td>
<td>12.07%</td>
<td>17</td>
<td>57.43%</td>
</tr>
<tr>
<td>8x8</td>
<td>OS</td>
<td>17.4</td>
<td>100.00%</td>
<td>29.6</td>
<td>100.00%</td>
</tr>
<tr>
<td>16x16</td>
<td>OU</td>
<td>26.6</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>16x16</td>
<td>OD</td>
<td>Unreliable initial recording 0.9</td>
<td>3.38%</td>
<td>14.5</td>
<td>46.93%</td>
</tr>
<tr>
<td>16x16</td>
<td>OS</td>
<td>14.7</td>
<td>55.26%</td>
<td>18.5</td>
<td>59.87%</td>
</tr>
<tr>
<td>32x32</td>
<td>OU</td>
<td>37.6</td>
<td>100.00%</td>
<td>39.6</td>
<td>100.00%</td>
</tr>
<tr>
<td>32x32</td>
<td>OD</td>
<td>6.5</td>
<td>17.29%</td>
<td>9.2</td>
<td>23.23%</td>
</tr>
<tr>
<td>32x32</td>
<td>OS</td>
<td>30.1</td>
<td>80.05%</td>
<td>19.7</td>
<td>49.75%</td>
</tr>
<tr>
<td>64x64</td>
<td>OU</td>
<td>28.8</td>
<td>100.00%</td>
<td>30.5</td>
<td>100.00%</td>
</tr>
<tr>
<td>64x64</td>
<td>OD</td>
<td>5.3</td>
<td>18.40%</td>
<td>7.7</td>
<td>25.25%</td>
</tr>
<tr>
<td>64x64</td>
<td>OS</td>
<td>23.4</td>
<td>81.25%</td>
<td>31.1</td>
<td>101.97%</td>
</tr>
</tbody>
</table>

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Treatment Options for the Degenerative Myope

There are many treatment options for the myopic patient. Attempts to arrest the progression of myopia have ranged from the use of different spectacle lenses to surgical procedures involving scleral reinforcement.\textsuperscript{11,18,19}

Multiple corrective lens theories have been tested for their effectiveness in the decrease of myopia. The use of spectacle lenses to correct myopia is no longer thought of as a process that induces myopic progression. Studies indicate that there is minimal difference in myopia progression between full prescription wear, distance-only wear, and non-wear.\textsuperscript{11,18,19} A review conducted at the Ohio State University found that under-correction of myopia may actually lead to an increase in myopia progression.\textsuperscript{20}

Several studies on multifocals and bifocals have been documented, with varying conclusions. Theories behind the use of multifocal lenses stem from the thought that myopes, having reduced accommodative response at near, experience retinal blur, which can be related to uncoordinated eye growth.\textsuperscript{11,21} Studies show that bifocal lenses may help to slow myopia progression rates more than single vision lenses, especially in children with esophoria and high accommodative lag.\textsuperscript{22-26}

Contact lens use is believed to help increase peripheral awareness and may improve the quality of the retinal image while reducing peripheral blur.\textsuperscript{11} Theoretically, contact lens use should help to decrease peripheral image defocus. If peripheral defocus stems axial elongation, then it is predicted that myopia progression should be slowed with contact lens use. Studies have shown varying results with contact lens versus spectacle use in myopia progression.\textsuperscript{11,26,27} Bifocal contact lenses are another lens option that is currently being investigated. In a recent trial conducted by Lam et al., a two-year double-blind study was conducted on 221 children aged 8-13. The study compared myopia and axial length progression between children given single vision contact lenses and multifocal contact lenses (add power of +2.50). The study concluded that myopia progressed 25% more slowly in children wearing multifocal contact lenses. Axial length was also less in the multifocal contact lens group when compared to the single vision contact lens group.\textsuperscript{28}

Orthokeratology uses reverse geometry corneal reshaping techniques to reduce myopia up to -6.00D. There has been significant evidence developed over the past 3-5 years showing orthokeratology to have efficacy in reducing myopia progression. In a recent review article, Smith and Walline\textsuperscript{29} documented eight studies that demonstrate that orthokeratology contact lenses slow axial length growth compared to single vision contact lenses,\textsuperscript{30} single vision spectacles,\textsuperscript{30-35} and single vision gas permeable lenses.\textsuperscript{36} The axial length difference ranged from 0.02 to 0.42 mm. While there have been multiple cases of severe microbial keratitis related to orthokeratology overnight use since 2001,\textsuperscript{37,38} the rates are as high as wearing soft contact lenses at night.\textsuperscript{39}

In the above case series, the patients reported greater visual satisfaction and improved comfort with contact lens use. Patient 5 was the only patient in her family to be tested with Diopsys VEP before and after her multifocal contact lens fit. Normalized results indicated improved amplitudes with multifocal contact lens wear during most pattern sizes when compared to testing through spectacles. At the time of writing, the other family members have not been tested both with and without contact lens wear to determine whether the findings would be similar with single vision contact lenses; this will be considered for future studies.

Atropine is a non-selective muscarinic antagonist that inhibits accommodation. Atropine use to slow myopia progress has been theorized to decrease axial elongation and to inhibit form deprivation. Dopamine, which is thought to have a role in the control
of eye growth, is also affected by the use of atropine. Studies indicate that atropine use to decrease myopia progression would have to be in high doses and would induce severe retinal side effects.\textsuperscript{11,26,40} Another therapeutic agent currently being studied is pirenzepine, a selective antagonist of muscarinic M1 receptors. Pirenzepine, when compared to atropine, is less likely to produce mydriasis and cycloplegia. Several studies have shown a positive correlation between pirenzepine use and a decrease in myopia progression. When looking at some of the studies more closely, problems make the results look dubious: large patient dropout rates and the high dosage of the drug needed for effect.

Surgical considerations for scleral reinforcement have been researched specifically for pathological myopia. During the surgical process, reinforcing material is placed over the posterior pole to prevent thinning and enlargement of a posterior staphyloma involving the macular area.\textsuperscript{41} Scleral reinforcement surgery is more common in China and Russia; the efficacy and safety of the procedures are currently being studied in the United States.\textsuperscript{41-45}

**Conclusion**

Myopia is a condition for which cause and treatment have long been debated. Though current research has still not been able to identify one specific factor causing degenerative myopia, or the most effective way to slow progression, new theories are being studied. Until conclusive research is published, the exact cause and most efficient treatment for degenerative myopia remain open to debate. Though research indicates a genetic predisposition for progressive myopia, environmental factors should not be ignored. All treatment options must be considered and researched before deciding which is the most appropriate for each patient.

**References**


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