# The Clinical Diagnosis & Treatment of Achromatopsia

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## ABSTRACT

Congenital achromatopsia (meaning without color vision) or rod monochromacy is a rare hereditary vision disorder that affects 1 in 33,000 people in the United States. It is a stable condition that is often overlooked or misdiagnosed, because so many eye care professionals are unaware that this condition exists. It is classified according to the mode of inheritance as either autosomal recessive (AR) or X-linked (XL). The AR achromats are further divided into complete achromats and incomplete achromats. Regardless of the inheritance pattern, almost all persons affected have reduced visual acuity, severe photophobia, poor to no color discrimination abilities, nystagmus and a reduced photopic electroretinogram (ERG). Unfortunately these patients must not only deal with the ocular complications, but also social stigmas that accompany this condition. Despite these serious signs and symptoms, there are ophthalmic options available to improve the quality of life for these patients.

## **INTRODUCTION**

Congenital achromatopsia is so rare that it occurs in .003% of the population in the United States<sup>1,2</sup>. Rod monochromatism is not a disease that is well documented, therefore many people suffering with this condition go misdiagnosed for years, and do not benefit from the proper treatment or care. There are two forms of inheritance patterns, autosomal recessive (AR) and X-linked (XL). Some sources say AR is the most common form of the disease while others say XL is the most common<sup>3,4</sup>. The AR inheritance pattern is further divided into a complete form and an incomplete form<sup>2</sup>.

Complete achromats have only one type of functional photoreceptor, rods, and are thus unable to discriminate colors based on hue. They can also be referred to as monochromats, rod monochromats, or typical complete achromats<sup>2</sup>. Incomplete achromats, on the other hand, do have some residual cone function, which is usually so reduced that color vision may be manifested only under certain conditions (large fields, restricted range of light levels)<sup>4</sup>. This form of achromatopsia is also known as atypical achromatopsia (which should not be confused with the third type of achromatopsia described below). Clinically, complete and incomplete forms are indistinguishable<sup>2,4</sup>.

The last form is X-linked achromatopsia (XL), also known as atypical achromatopsia, blue cone monochromats, or blue monocone monochromats. These individuals have two types of functional photoreceptors: cones with short wavelength sensitivity (blue cones) and rods. They do possess reduced amount of dichromatic color vision at low illuminances, but in higher levels they essentially monochromats<sup>2</sup>.

#### HEREDITY

There are two forms of inheritance seen in achromatopsia: autosomal recessive (AR) and X-linked (XL). The AR form of achromatopsia is seen in the offspring only if each parent passes on the defected or abnormal gene to the child. The child would then possess two abnormal genes resulting in achromatopsia. The chance of two carrier individuals (each person possessing one normal gene and one abnormal gene) producing a child with achromatopsia is 25%. Each time these same carrier individuals mate, they have a 25%

chance of having a child with achromatopsia, 50% chance of a child being a carrier, and 25% chance that a child will be normal for color discrimination<sup>1</sup> (see Figure 1).

If a person with normal color vision mates with a carrier individual, none of the children will possess the trait of achromatopsia. However, there is a 50% chance the offspring will be carriers for the trait, and the other 50% could be normal for color discrimination (see Figure 2).

When two achromatic individuals mate, all children will possess the trait of achromatopsia (See Figure 3).

**Autosomal Recessive Achromatopsia Punnett Squares** 

	Figure	1
	N	n
N	NN	Nn
n	Nn	nn

Figure 2		2	Figure 3
	Ň	N	
1[	NN	Nn	n nn
11	Nn	Nn	n I nn

n

nn

nn

NN= normal color vision

Nn = carrier for the achromatic trait

nn = achromatic individual

Figure 1- punnett square for two carrier individuals. Figure 2- punnett square for a carrier individual and a person normal for rod /cone function. Figure 3- punnett square for two achromatic individuals.

The XL form of achromatopsia differs in the mode of inheritance from that of the AR form, in that, the gene has been linked to the X chromosome. The exact loci is still unknown, but researchers have found that three defects in the X chromosome result in this form of achromatopsia<sup>1</sup>. XL achromatopsia occurs more commonly in males<sup>1</sup>. Affected

fathers can not pass the gene to their sons, but each of their daughters will be at least a carrier individual (Figures 5, 6 and 8). Affected mothers will always give the faulty gene to their son, and their daughter will always receive at least one defective gene (Figures 7 and 8). Carrier mothers have a 50% chance of giving the faulty gene to their son or their daughter (Figures 4 and 5). Similar to autosomal recessive hereditary patterns, if both parents suffer from achromatopsia, the child will be achromatic for visual function (Figure 8).

## X-Linked Achromatopsia Punnett Squares

F	igure 4	
	Xn	Xa
Xn	XnXn	XnXa
Y	XnY	XaY

Figure 7

Xa

XnXa

XaY

Xn

Y

XaY	Y	Xn
, e por le por	Fig	ure 8
Xa		Xa

F	igure 5	6 282 - 9 - 9 - 9 - 9
	Xn	Xa
Xa	XnXa	XaXa
Y	XnY	XaY

Xa

XaXa

XaY

Xa

XaXa

XaY

Figure 6			
	Xn	Xn	
Xa	XnXa	XnXa	
Ŷ	XnY	XnY	

XnXn = f	female	with	normal	color	vision
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XnXa

XaY

XnXa = carrier female for the achromatic trait (normal color vision)

Xa

Y

XaXa = achromatic female

XnY = male with normal color vision

XaY = achromatic male

Figure 4- punnett square for a carrier female and a normal male.

Figure 5- punnett square for a carrier female and an achromatic male.

Figure 6- punnett square for a normal female and an achromatic male.

Figure 7- punnett square for an achromatic female and normal male.

Figure 8- punnett square for two achromatic individuals.

(Normal and carrier meaning chromatic for color vision and abnormal meaning achromatic for color vision.)

Individuals who are carriers for the trait do not have any symptoms or show any signs for achromatopsia upon examination. There is no way of knowing that they carry the defective gene until they produce a child who has the condition. A pedigree can then be analyzed to determine the inheritance pattern and help in explaining the chances of producing other children with this trait.

The exact gene, or possible combination of genes, that causes achromatopsia for both types of inheritance has not been discovered. It is assumed that the defected gene(s) is involved in the way cones are made and/or the way they function<sup>1</sup>. Much research still needs to be done before the exact location in the chromosome map is found, and a cure may be discovered.

#### Signs and Symptoms

Characteristic signs and symptoms of congenital achromatopsia, for all forms of inheritance, include photophobia (which increases as the level of illumination increases), decreased visual acuity, decreased photopic ERG (usually normal scotopic ERG), inability to discriminate colors, and nystagmus. The most disabling feature for people with achromatopsia is not the decrease in visual acuity or the absence in color vision, but rather it is the blinding affect from higher levels of illumination which leaves many achromats almost nonfunctional<sup>1</sup>.

#### **Photophobia**

The normal human eye contains two types of photoreceptors: rods and cones. The cones are used for spatial resolution, color discrimination, detecting contrast changes and viewing objects in higher illuminances<sup>5</sup>. The rods, on the other hand, enable a person to see in dim illumination and are used to detect a stimulus that is 3 log units (1000 fold) dimmer than that detectable by cones<sup>5</sup>. This gives a person a high sensitivity to light, allowing for visual detection at low illuminances<sup>6</sup>. The rods also sum information over space, but are poor at resolving detail and discriminating color<sup>5</sup>. RM possess only one type of normal functioning photoreceptor; the rod. In scotopic conditions they are able to function at almost "normal" levels using their rods. However, in photopic conditions achromats function poorly. In normal (chromatic) patients, under photopic conditions, the cones slow down rod activity and allow for vision. Patients with achromatopsia do not have any cones, or have few dysfunctional cones, therefore they must depend on their rods for night vision as well daytime vision. The rods become saturated at higher light levels, putting the retina in a dark adaptation state despite daylight illuminances<sup>7</sup>. Because the rods become saturated, they are unable to function, resulting in poor visual acuity. Dazzlement is the correct description of the effect of photopic conditions on a rod monochromat, a flooding of vision with light such as to remove all sense of visual contrast. This poor visual acuity occurs in retinal illuminances over 1000 trolands<sup>8</sup>. (A standard screen projector is equivalent to 80 trolands, whereas a moderately sunny day can be around 1000 trolands.) Furthermore, photophobia that occurs in RM is not accompanied by the usual pupil constriction, injection or watering of the eyes<sup>8</sup>. Since these usual signs and symptoms of photophobia do not occur, and because most of the

exam is performed under lower levels of illuminance (which is not as disturbing to the achromatic patient), it may not be as apparent to the examining physician that a patient is photophobic.

To decrease the amount of illumination falling on the retina, and thereby improving visual comfort and acuity, red lenses may be used. Rod photoreceptors are poor at detecting light from the red range of the spectrum, therefore, RM are relatively insensitive to red light (most do not even perceive the color red)<sup>1</sup>. Thus, glasses that only allow red light to enter the eye, create a situation that is more visually pleasing to the achromat by preventing the rods from being saturated. Some RM, however, also prefer tints in dark brown, dark amber or other shades of red<sup>1</sup>.

"...In order to see at all outdoors in the daytime, I had to blink rapidly and continuously. When I opened my eyes even briefly, I could not see the world around me, because I was blinded by light. It was like having strong floodlights aimed at my eyes. The squinting and blinking allowed me to obtain just enough visual information to find my way around in reasonable safe surroundings...." The frequent blinking is a way to rely on the afterimage that remains on the retina to help in mobility<sup>9</sup>.

"... The most debilitating, handicapping, and distressing consequence of achromatopsia is the hypersensitivity to light. The practical problems of light aversion and feeling clumsy in intense light constitute more of a hindrance than not being able to experience colors or to discern minute detail...<sup>9</sup>"

## Visual Acuity

Fortunately, this is a non progressive disease with visual acuities in the range of 20/50-20/200 for all hereditary patterns<sup>3</sup>. The lower the illuminance level, the better the acuity and the better the visual functioning avoiding dazzlement. The use of tints (described later) in spectacles and contact lenses can aid in improving the acuity, particularly in higher levels of illumination<sup>8,10</sup>.

## **Refractive Error**

Achromatic patients do show different refractive errors from those of chromatic patients. On the whole, achromatic patients tend to have higher refractive errors with a tendency towards myopia. The highest amounts of myopia are more common in the XL group<sup>2</sup>. In addition, astigmatism is more common in achromats than with nonachromats. In the general adult population, 16% have astigmatism over 1.00 D. Among monochromats, over 80% of the AR population have 1.00 D or more, while in the XL group, 47% have one or more diopters of astigmatism<sup>2</sup>.

#### Nystagmus

Nystagmus is another condition that frequently occurs in patients suffering from achromatopsia. The amplitude averages 3 degrees and is pendular in young children and jerk type in adults<sup>11</sup>. Precise recordings have shown that the jerk type nystagmus has smooth movements with exponentially decreasing velocity in the slow phase. Differentiation in detecting increasing and decreasing velocity trajectories can not be made without the use of eye movements recordings<sup>12</sup>. In addition, occlusion of either eye does not change the amplitude or direction of the fast phase in patients with RM, as it does in other conditions<sup>11</sup>. Many patients report that the amplitude decreases with increasing age, and in many cases disappears completely<sup>11, 12, 13</sup>. Another interesting characteristic of the nystagmus found in RM is that the frequency and amplitude decreases with eyes opened in the dark<sup>12</sup>.

It is important to be able to differentiate nystagmus due to RM from those due to other causes. Congenital nystagmus differs from RM because it has a higher amplitude and has an increasing velocity of smooth movements with and without fast components or saccades<sup>11, 12</sup>. Latent nystagmus is characterized by jerk type waveform with decreasing slow phase velocity, and occlusion of either eye changes the direction of the jerk<sup>11, 12</sup>. Spasmus nutans patients typically have asymmetry of nystagmus amplitudes and frequency between the two eyes, whereas those patients with RM have symmetrical amplitude and frequency<sup>11</sup>. Patients with oculocutaneous albinism, or severe ocular albinism, have nystagmus with much larger amplitudes<sup>11, 12</sup>.

"...At 6 months our son was diagnosed with congenital nystagmus. His aversion to bright light and poor distance vision were obvious to us at an early age. We took him to several ophthalmologists who 'specialize' in nystagmus. They said that, because his nystagmus was mild, he could see fairly well, that he wasn't photophobic, and we should just let him wear a hat outside. We then took him to an optometrist- a low vision specialist- who did a more thorough job of testing his vision and declared him legally blind. He said he suspected achromatopsia. This was the first we had heard of this condition. Although way back we were told there was a slight chance he could be colorblind, we now strongly suspected it, since he didn't know his colors. The possibility that all his visual difficulties could be related to one eye condition was a revelation..., $9^{n}$ 

#### **Color discrimination**

Not surprisingly, people with achromatopsia do not possess the ability or have very limited ability to discriminate colors. Rods do not enable a person to detect color differences, but can help in detecting differences in brightness<sup>5</sup>. The rods are most sensitive to wavelengths near 505 nm, therefore these wavelengths appear brightest. The farther away the wavelength is from 505 nm, the dimmer it will appear to the RM. Green colors appear the brightest, yellow-orange colors are a little dimmer, and red appears the dimmest<sup>5</sup>. Several tests are used to determine the extent of color vision and help in the classification of the disorder. These tests include the Farnsworth-Munsell D-15, Berson test, Ishihara Pseudoisochromatic Plate (PIP), and the Sloan test.

The Farnsworth-Munsell D-15 test is used to differentiate among various types of color anomalies. The patient is to arrange a set of 15 color samples in order of color appearance, with the origin of the arrangement being fixed by a color sample permanently mounted in a box with a black interior. Each color sample is embedded in a black plastic cap and subtends 1.5 degrees at 0.5 meters. Each cap is numbered on the back according to proper location on the color circle. The test is scored by recording the sequence of the numbers in the cap arrangements by the test subject<sup>4</sup>. Incomplete and complete achromats tend to produce a scotopic axis which lies between deutan and tritan axis, while XL achromats tend to produce an axis that is similar to deutans<sup>4</sup>. There is so much individual variation that these serve only as guidelines and are not used to classify the type of achromat.

The Berson test is unique because it can differentiate AR from XL monochromats. The test consists of four test plates, each plate contains three identical blue-green and one blue-violet patch made of Munsell papers that are scotopically matched. The patient's task is to locate the blue-violet patch among the blue-green patches from the four test plates. Missing any one of the four plates is considered failing the test<sup>4</sup>. The XL achromats are able to pass the Berson test, but the AR achromats do not, failing at least one plate.

The Ishihara Pseudoisochromatic Plates (PIP) consist of a arabic numeral composed of one chroma surrounded by a background of another chroma. These two different chromas fall on a line in color space that can be confused as being the same by color defectives. The saturation of the chromas is varied to define the depth of color deficiency<sup>10</sup>. The achromatic patients tend to color match on the basis of luminosity differences and not hue discrimination. Any correct responses by the RM are random and do not allow for any classification. Therefore, RM do not pass the PIP which provides a quick diagnosis of color deficiency, but does not categorize the defect<sup>4</sup>.

The Sloan achromatopsia test can also be used to test for color abnormalities. The observer is shown a two degree circular bipartite field, in which one half of the field contains a yellow (589 nm) test light of variable luminances and the other half contains a

mixture field of red (670nm) and green (545 nm)<sup>4</sup>. The patient is asked to match the luminance of the yellow field to that of a pure red, pure green and a mixture of red and green<sup>4</sup>. Although the end results show that AR and XL both produce too much variation on the test to separate these two forms, it can be used to discriminate achromats from nonachromats.

"...At buffets or smorgasbords, I can't tell tuna casserole from blueberry muffins. Once I took something I thought was potato salad and it turned out to be some kind of marshmallow Jell-O...<sup>9</sup>"

"...In my early childhood, when I used coloring pencils or crayons, I always broke all the "rules" about the correct colors to use. I would happily color the sky green, the grass and leaves orange, the sun white, and so on. I was always corrected in these choices by those who knew better, and so eventually I gave up painting and coloring. I would memorize the colors of my clothes and other things. I learned the rules for correct use of colors and the most probable colors of various things...<sup>9</sup>"

#### Electroretinogram (ERG)

The results from electroretinogram testing of achromatic patients can be used to help in the diagnosis of this condition. Normal subjects have equal dark-adapted rod and cone amplitudes, rod/cone ratio (R/C ratio) equal to one, to scotopically matched blue and red stimuli, whereas those with achromatopsia have a R/C ratio less than one<sup>14</sup>. ERG testing can be used to differentiate rod monochromacy from progressive cone disease which has a R/C ratio of more than one<sup>14</sup>. More specifically, those patients with rod monochromacy do show abnormal cone mediated activity when a red flash or a white flash is presented under photopic conditions. The rod mediated activity is normal when presented with a dim blue flash or white flash under scotopic conditions<sup>15</sup>. Simply put, achromats will have a reduced photopic ERG, but relatively normal scotopic ERG<sup>13, 16</sup>.

Differential diagnosis chart for Rod Monochromacy:<sup>3,4,5,6,7,8,16,17,28</sup>

Differential Diagnosis	Differentiating Characteristics
Acquired forms	Onset later in life
	Usually sudden loss of color vision
	2° to: trauma, migraine, cortical lesions, Alzheimer
	Normal acuity
Congenital Color Deficiency	Normal acuity
	Axis of D-15 does not match that of the achromats
	No photophobia
	No nystagmus
Reitinitis Pigmentosa	Decrease night vision
	Loss of peripheral vision which proceeds to central
	vision loss
	Progressive condition
	Good vision in higher levels of illumination
Progressive Cone Disease	Progressive disease
	Acquired decrease in central acuity (usually within
	the first decade of life)
	Decrease in color vision when acuities decrease to
	20/40
	Fundus: Bulls eye lesion at macula, diffuse pigment
	clumping on retina, choroidal vascular atrophy, optic disc pallor
	Visual Fields: scotoma- relative defect that deepens, enlarges and
	coalesces
	ERG- R/C ratio greater than one
Congenital Nystagmus	Larger amplitude and higher frequency
	Increasing velocity of smooth movements with and
	without fast components or saccades
Latent Nystagmus	Jerk nystagmus with decreasing slow phase velocity
	Occlusion of either eye changes the direction of the
	jerk
Spasmus Nutans	Asymmetric amplitude and frequency between the
	eyes
Oculocutaneous Albinism	Good color discrimination
	Pale skin, hair and irides

## **Treatment**

There are many treatment options available to improve the lifestyle of the achromat. It is important to note that one solution will not work in all situations, and what works for one person may not be the solution for another. Many times, trial and error is the only way to find out which treatment method will work best for each individual.

The use of tinted lens (either spectacles or contact lenses) is the most common method used to reduce the glaring effects of bright lights for the achromat. Red filters (without blue transmission) are optimal for complete AR achromats, whereas, dark magenta filters (allowing some blue transmission) work best for XL achromats. Incomplete AR achromats are best helped with red-brown filters<sup>2</sup>. The use of grav and brown tints do not have the same effect as the above lenses, because they do not block out as much of the spectrum as the red lenses. Using selective cut off filters, overall retinal illumination can be brought to mesopic levels for increased patient comfort<sup>10</sup>. The brighter the conditions, the darker the tint needs to be in order to keep the rods from being saturated. The achromat may need to switch several different pairs of glasses of various tints throughout the day depending on the lighting, therefore, it is much easier to change glasses than it is to insert and remove different contact lenses<sup>9</sup>. However, contact lenses do work great when outdoors on bright sunny days. Contact lenses with a red tint provide the best visual comfort reducing illumination to tolerable levels. The X-Chrom lens is a rigid gas permeable (RGP) red contact lens that transmits light near the red range of the spectrum (590nm-700nm), with an overall transmission between 15-40%<sup>23</sup>. The Alberta II lens is another red RGP contact lens that is similar to the X-Chrom lens, but allows 5% more

transmission in the blue range of the spectrum. Soft contact lenses with the red or magenta tint are also available through Optical Designs, Adventures in Color and Kontur Contact Lens<sup>23</sup>. The amount of tint can be varied in order to best suit the needs of the individual. Many achromats will use contacts in outdoor settings and then switch to glasses when returning indoors. It is obvious that there is always a trade off, solutions will always have some advantages and some disadvantages.

"... I've never tried contacts, nor do I plan to do so. In any given day I may take my glasses off and then put them on again 20-30 times, at least. That would not be possible with contacts...<sup>9</sup>"

"...I love going for walks, jogs, etc., with my tinted contacts on. The world just looks bigger. I really feel more 'mobile' with them on...<sup>9</sup>"

In addition to using tints to reduce the amount of retinal illuminance, side shields and hats can also effectively reduce the amount of glare. There are many different styles of frames available that reduce peripheral glare including the "Digi" sportlens, "Glacier goggles", 'driver' sunglasses, Zurich Shields, 'instant sunglasses', 'SuperVisors' and solar shields. Clip-on and slip-in spectacle tinted lenses are also available to use as an additional tint to help reduce illumination that enters the eye<sup>1</sup>. One drawback to using side shields is the reduction of peripheral vision, which may be too limiting to achromats in certain situations<sup>1</sup>. "...Side shields on spectacle frames shut out unwanted light, but they also prevent motion detection in the peripheral visual field, which is important for moving about safely, so I don't use them....<sup>9</sup>"

Reading can be challenging for achromats, but not totally impossible. To help improve the near visual acuities, there are many types of magnifying devices available. Hand held and stand magnifiers of various powers are the simplest devices used to make reading easier. Telemicroscopes, microscopes, half-eyes, high plus spectacles, and closed circuit televisions are a few low vision devices that can also be introduced to the achromat in helping to decide which product works best for the RM. Large print books may also be helpful to some achromats, allowing for larger retinal image size while reading. This, however, may not be the case with every achromat, as some find the increased size too tedious to read at a normal pace. An ideal situation would be to increase the size of the letter just enough to make the letter more visible, without making it so large that only one letter can be read at a time<sup>1</sup>. The large type books may be more appropriate for the presbyopic patients who need the extra magnification due to the decreasing amounts of accommodation in their systems<sup>1</sup>.

"...I didn't like large print books back when I was in school. Beside the fact that the writing was in appropriately large for me, an open large print book on my desk was just another huge expanse of white- more glare for me to deal with...<sup>9</sup>"

"...When traveling in unknown surroundings, I always carry a small 8 power, monocular, which I can conceal in my hand and which I use for reading street names, destination signs and other information I cannot get close to....<sup>9</sup>"

"...For certain tasks such as reading newspaper want ads, I use a closed circuit TV. It could also be useful when working on circuit boards or when making repairs of something composed of small parts. I prefer to use a 4X magnifier to 'spot read' telephone books or maps...<sup>9</sup>"

To improve upon distance visual acuities, a telescope can be mounted in the spectacles. A telescope can be used to spot check various objects located in the distance that would otherwise be out of focus. The field of view is limited when viewing through the scope, therefore it is advised to look through the scope occasionally. Tints can also be incorporated into the lens to reduce the blinding affects of bright lights. The M-lens (M Technology), a 4X magnification, is one type of telescope that is mounted behind the lens, making it more cosmetically pleasing, which can also incorporate a tint for comfort. Other telescopes (in various powers) are available that can be mounted on the front of the lens.

"...Both our children have glasses that have telescopes mounted on the inside. They wear these to all events such as concerts, circus, sporting events. They both say that it is great to be able to applaud and watch the event at the same time. With their hand held scopes, they could not do this...<sup>9</sup>"

Driving is another issue that can be a major obstacle for achromats. Many have been told at young ages that they would never be able to drive, therefore many do not even attempt it. However, distance visual acuity can be improved with the use of telescopes or bioptics that would meet legal standards of visual acuities for driving, and therefore allow one to obtain a license. State laws vary throughout the country and various restrictions may be placed on the license. The following states allow the use of a telescope for driving: Alaska, Arkansas, California, Delaware, Georgia, Hawaii, Idaho, Illinois, Kansas, Maine, Massachusetts, Michigan, Missouri, Nevada, New York, Texas, Vermont, Virginia, and Wyoming. The use of tints in addition to the use of telescopes can make driving even easier improving acuity and decreasing photophobia. Occupational therapists offer programs that train the individuals on the use of the telescopes in daily living situations and also prepare them for use of the telescope while driving. Although few achromats choose to drive, it is one option that should be presented to all individuals.

... "I obtained a driver's license in one state under a relatively new bioptics program, but my current state of residence does not support this program. I am fairly comfortable driving under certain lighting conditions, wearing red contacts (which allow me to distinguish red) and bioptic glasses. The glasses I use have a slim sidemounted periscope across the top of the frame, instead of the telescope-in-the-lens type. These work well but are significantly more expensive than the telescope type. I find red contact lenses work well for red enhancement but are poor for acuity...<sup>9</sup>"

...From a man with 20/100 visual acuity: "I obtained a Georgia driver's license under a new bioptics program. I am fairly comfortable driving under light condition, wearing bioptic glasses and red contact lenses, which allow me to distinguish red. I find red contact lenses work well for red enhancement but are poor for acuity...<sup>9</sup>. Achromats can also work with an occupational therapist who is able to offer suggestions to improve their orientation and mobility skills. These include sensory training, orientation to surroundings in and out of the home, making the home more suitable to the achromatic patient, self-protection, and other activities of daily living<sup>1</sup>. Occupational therapists working in conjunction with optometrists can also teach achromats how to use telescopes, bioptics, and many other low vision devices.

#### CONCLUSION

Rod Monochromacy can be a devastating disease to those who suffer from it, but it does not have to be that way. With proper diagnosis and treatment, these patients can function at near normal levels. A red or amber tint can alleviate the photophobia and improve their orientation and mobility skills. A high power magnifier can be used to enlarge small print for easier reading. Telescopes can be implemented to increase visual acuity at distance and possibly enable a RM to drive a vehicle. There are also many organizations available for support groups to help with the emotional problems. Know the signs and symptoms of the disease- severe photophobia, poor to no color discrimination, nystagmus, decrease photopic ERG, decreased visual acuity, and possible hereditary factors. Recognize these and you can improve the lifestyle for the rod monochromat patient, which will prove to be a rewarding experience for you as well. Resources Available To Help the Achromatic Individual<sup>1</sup>

- Achromatopsia Network- editor Frances Futterman The Achromatopsia Network
   P.O. Box 214, Berkeley, CA 94701-0214
   WWW: http://www.achromat.org
   E-mail: Futterman@achromat.org
- Vision Resource Foundation 818 Mt. Auburn St., Watertown, MA 02172
- Prevent Blindness
  4299 California St. #101, San Francisco, CA 94118-1314
  1-800-338-3041 (in CA) or 1-415-387-1689
- National Association for Parents of the Visually Impaired P.O. Box 317, Watertown, MA 02272 1-800-562-6265
- National Association for the Visually Handicapped 3201 Balboa St., San Francisco, CA 94121 1-415-221-3201 (for western states) 22 W. 21<sup>st</sup> St. 6<sup>th</sup> floor, New York, NY 10010 1-212-889-3141 (for eastern states)
- 6. The American Fondation for the Blind 1-800-232-5463
- National Library Service for the Blind and Physically Handicapped The Library of Congress 1291 Taylor St. N.W., Washington D.C. 20542 1-800-424-9100

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