UNIVERSITY OF LATVIA
FACULTY OF PHYSICS AND MATHEMATICS
DEPARTMENT OF OPTOMETRY AND VISION SCIENCE

ACHROMATOPSIA PROBLEM TREATMENT WITH CHROMATIC CONTACT LENSES

MASTER THESIS

Author: **Claudio Aldegheri**
Student card number: Ca15001
Supervisor: docent, Dr.phys. **Sergejs Fomins**

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ABSTRACT

AIM: Evaluation of filtered contact lenses in the treatment of achromatopsia

METHOD: The study is carried on a group of 8 subjects, 6 men and 2 women, between 8 months and 25 years old, over a period of time ranging from 2012 to 2016. After an ophthalmologic analysis and the diagnosis of achromatopsia, I made the pre-application, application and post-fitting procedure for filtered CL. I evaluated recovery time after glare with and without CL and then I gave patients a satisfaction questionaire.

CONCLUSIONS: Filtered CL have reduced symptoms like photophobia, have allowed a recovery time after glare into the standards and have improved many psychological aspects of life.

Key Words : Achromatopsia, filtered contact lenses, photophobia, glare, light

Thesis is written in English on 38 pages. It contains 2 figures, 6 tables and 36 references.

ANOTĀCIJA

MĒRĶIS: Novērtēt filtru kontaktlēcu (KL) pielietojumu ahromatopsijas simptomu korekcijai.

METODE: Pētījumā piedalījās 8 dalībnieki (6 sievietes un 2 vīrieši) vecumā no 8 mēnešiem līdz 25 gadiem, periodā no 2012. līdz 2016.gadam. Pēc oftalmoloģiskās pārbaudes un ahromatopsijas noteikšanas, tika veikta speciālās krāsainās KL pielaikošanas procedūra.
Pacientu atbildes tika noteiktas ar anketas palīdzību un noteikts apžilbšanas laiks ar un bez piedāvātām KL.

SECINĀJUMI: filtru KL samazināja fotofobiju; apžilbšanas laiks kļuvis salīdzināms ar normālā redzes gadījumu; dalībnieki atzinīgi novērtē psiholoģiskos un sociālos ieguvumus ar izrakstītām filtru KL.

Atslēgas vārdi: ahromatopsija, filtru kontaktlēcas, fotofobija, apžilbšana

Maģistra darbs ir uzrakstīts uz 38 lapām. Tas satur 2 attēlus, 6 tabulas un 36 literatūras avotus.

1.1. Cones and Color Vision

The process of vision begins thanks to particular cells, located in the retina, called photoreceptors, which are cones and rods. Photoreceptors have the capacity to absorb the light and then to convert it into electrical signals. From these cells, the signal is sent to the occipital cortex (the posterior part of the human brain) through the optic nerve. Into the brain the electric signals are analyzed, processed and then transformed into images that allow us the ability to see. Rods are located in the peripheral region of retina; they are highly sensitive to movement and they allow us to see in dark or low lighting conditions (scotopic vision).
Cones capture with very fine precision details and colors. They are located in the fovea, that is the central portion of the retina, and are deputated to the fotopic vision.

Human are able to perceive colors and discriminate objects thanks to the distribution of the wavelengths of the light that they reflect to the eye. While differences in luminance are often enough to distinguish objects, color adds another perceptual dimension that is especially useful when differences in luminance are little or non-existent.
Colors can give a perceptual dimension to objects, and that is useful in different situations, like in low or no brightness conditions. A difference in luminance, instead, make us only able to distinguish the shape and main characteristics of the object.

Colors obviously give us a quite different way of perceiving and describing the world we live in. So, the humans perceive colors after a sensations produced and generated by the brain, after a stimulation of cones. These contains a pigment which is sensitive to certain wavelenghts that come from light sources.

The perception of colors depends on many factors, such as the pigments of the object, the wavelenght of luminous ray that hits him, and the subject itself.
There are three types of cones that differ in the photopigment they contain. Each of these photopigments has a different sensitivity to light of different wavelengths, and for this reason are refered to as “blue”(420 nm) “green”(530 nm), and “red,”(560 nm) or, also, short (S), medium (M), and long (L) wavelength cones, terms that can describe their spectral sensitivities.

To have complete informations about the surrounding world, the brain needs the interation of at least two different types of cells that must be sensitive to different wavelenghts in order to respond to every different stimulus.
The ability to distinguish all colors depends greatly on how much light enters the retina.
If a large amount of light reaches the retina, surely the brain will be able to analyze and process a big number of informations.
So we will have a better ability to distinguish colors.

1.1.1. Types of color vision disorders

Colours-related visual problems may have two different origins: one due to an altered absorpion of the light from the rod pigments, and one caused by an hereditary deficiency of photoreceptors used to perceive colors.
They are rarely caused by injuries of central portion that transduces informations about color perception. Color blindness represent, therefore, an inability to distinguish certain wavelenghts (Swanson, Cohen, 2003). It occurs when one or more types of cones are absent, or present but damaged, or incapable of sending a correct signal that has to be processed into the brain. When a pigment is injured, the subject is defined as protonormal, istead, when all pigments are completely absent, they are called protonotops.

TRICHROMATIC: The vision of colors is perfectly normal and the three pigments of rods are all present correctly. The subject is able to see the three primary colors.

DICHROMATIC: There is a deficiency of one of the three cones, and the other two are perfectly normal. In this case, the retina is able to respond to only two of the three primary colors. We can distinguish three subcategories of dichromates, depending on which of the three normal pigments results absent.

- Protanopes: We are in presence of a reduced brightness of red, orange and yellow, compared to normal. This the most common condition.
- Deuteranopes: This is a particular dichromatic case, in which the person confuses red from green. This dynsfuncion affects the green-cones.
- Tritanopes: In this condition, the individual in unable to distinguish blue from yellow.

There are two other categories represented by monochromates and acromates. Subjects that are part of first category possess only a cone and presents a severe reduction in visual acuity, photophobia and nystagmus. Achromates can only perceive white and black colors. Typically, the complete achromatopsia is associated to a reduced visual acuity, photophobia and pendolar nystagmus.

1.2. Achromatopsia

1.2.1. Clinical characteristics

Achromatopsia is defined by a visual, congenital and pathological visual condition (Millidot, 2014). It consists of a rare hereditary defect that is present from birth and that is not degenerative. According to the primary cause of this disability, we can identify three types of achromatopsia:

- Congenital achromatopsia
- Dystrophy of cones (or degenerative achromatopsia)
- Celebral achromatopsia

As stated above, in the human retina are present two types of cells, cones and rods, called photoreceptors, which are susceptible to light. Cones are focused on the fovea and are specialized in daytime vision. They allow us to adapt to light, distinguish different colors and to discriminate details. Rods are located in the peripheral region of the retina and are specialized in scotopic vision. They are much more sensitive to light, however they saturate quickly when light increases and they don't allow color vision or to distinguish the more finest details.

All the individuals affected by achromatopsia present a wrong color discrimination along all three axes of colour vision that correspond to the three classes of cones. In them, the cones work little or not work at all, so they can only rely on rods to see.

Many individuals present a complete achromatopsia (total monochromatism of rods), related to a total dysfunction of all three cones. It represents the most severe condition. Patients rarely present an incomplete achromatopsia (partial monochromatism of rods), in which one or more cones is partially working. Symptoms are similar to those of the complete achromatopsia, but are basically less severe. Finally, we have the monochromatism of blue – cone, in which a part of cones is active and always sensitive to blue color.

Achromatic patiens have four major visual symptoms and clinical conditions: avversion to light (photophobia), reduced or complete loss of ability to distinguish colors, reduced visual acuity (poor vision of far details), nystagmus (involuntary movement of one or both eyes), small central scotomy, eccentric fissation and anomalous or absent photopic electroretinorgam (recording of the electrical activity of the eye under a light condition in which the cones would normally work). Let's see now these symptoms:

a) Day blindness: it's the primary and most significant and cripping symptom that causes a strong intolerance to the light sources in the achromates. This has a strong influencce on the life of the individuals. In fact, instead of improving their vision, every source of light darkens. That is why the eye of the achromates cannot addapt to a lighter light than twilight.

b) Reduced visual acuity: since the cones are concentrated in the macula, they are responsible for the perception of central vision. A lesion or damage at this level produces a reduction in visual acuity that cannot be corrected with conventional glasses. Proximal vision is usually less negatively influenced than the far one. Patients with achromatopsia often show a wide range of refractive errors that may vary from extreme hyperopia to an high myopia.

c) Color blindess: the second major symptom is the lack of the perception of the color band. The perception of achromatic patients is unreliable. Many of them have learned, during growth, to associate certain colors to objects and to recognize them according to the intensity of light (Sharpe et al, 1999).

d) Nystagmus: another characteristic symptom is the presence of nystagmus, an oscillatory, ryhtmic and involuntary oscillation / movement of the eyeballs, in a specific direction of the visual axis, more frequently on the horizontal one. It causes strong variations in vision and it usually occurs during the first few weeks of life.
   Nystagmus can be influenced by:

   - distance, and it increases when trying to focus on distance
   - illumination level, lower in the dark and better with strong lights
   - fixation, it worsens when trying to focus on something
   - age, it tends to decrease with age

When the presence of this condition is suspected, the ophtalmologist may add further diagnostic tests such as optical coherence tomography (OCT), autoflourescence of the fundus and visual field. Many achromatic individuals exhibit a normal appearence of the eye, while others present bilateral macular changes such as lack of foveal reflex, retinal veins constriction and abnormalities in the macular pigmentation. It's important to consider that is not possible to diagnose achromatopsia only considering the ocular fundus. In fact, Electroretinogram (ERG) may show an absence or a marked reduced response to light while the scotopic response would be normal or partially abnormal.

1. 2.2. Genotype - phenotype correlations

Achromatopsia has been associated with the mutation of five genes (GNAT2-1P13, PDE6C-10Q24, PHE6H-12P13, CNGA3-2Q11.2 and CNGB3-8Q21.3), that code for key components of the cone-phototransduction cascade. The most common mutations are those in CNGN3, followed by those in CNGA3. The other ones are more rare.

1.2.3. Differential diagnosis

As stated above, achromatopsia is easily recognizable by symptoms such as reduced visual acuity, nystagmus, reduced or complete inability to distinguish colours and other electroretinographic and psychophysical evidences.
However, there are other retinal disorders that may be confused with achromatopsia.
Let's see them:

1) Blue-cone monochromatism

Like achromatopsia, monochromaticity of blue cones is defined by symptoms such as nystagmus, low visual acuity, eccentric fixation, possible retinal abnormalities and a low or absent color perception (Sharpe et al 1999). However, unlike in achromatopsia, the peak of the photopic brightness function is 400 nm, not 507 nm, and the response of electroretinogram is positive with a blue flash on a yellow background. This is because blue cones are active together with rods. Typically, this pathology affects men; the test that allows to specify the distinction with achromates consists of a special target of four colors or the test with two colored filters.

2) Cone dystrophy

In presence of this particular condition, symptomatology generally consists of four characteristics alterations: reduction of visual acuity, difficulties in colors discrimination, visual disturbance and discomfort in full light conditions (photophobia). The most characteristic form origins in adolescents and young adults and then tends to become worse progressively until mature age. The diagnosis is usually based on a clinical evaluation of the patient (there is frequently the presence of a central scotoma), electrophysiological investigations, OCT. With the fluoroangiography it's common to see the "bull's eye" aspect, which origins from the alternance of iper and ipo alterations at the macular level. It is not easy to distinguish this pathology from achromatopsia, especially if it occurs early in childhood. The discriminator is represented by the progression of the same, a factor which characterizes and allows to distinguish that condition from achromatopsia.

3) Red-green color vision disorder

This condition, usually manifested in male individuals, is not related to physical or clinical abnormalities. Many protanopes and deuteranopes don't show any particular difficulties in giving a name to the colors, so that individuals with low levels of the desease don't realize they are affected until they are subjected to diagnostic tests. Among those affected, about 8% is male while only 0,5% is female. This visual disorder affects in a lower percentage African and Asian subjects. The test used to diagnose this pathology is the Ishihara plates. It should be specified that the final diagnosis of these abnormalities requires the use of an anomaloscope.

4) Tritan yellow-blue disorders

Tritan disorders hit the blue cones and are often defined as yellow-blue deficiencies. They are very rare and usually incompletely manifested, so the nature of their manifestation could be very limited. Other yellow-blue cone disorders, similar to this one for many aspects, may result from aging or from disorders of the choroid, retinal pigment, retina or optic nerve. They are often progressive and degenerative and associated with other symptomps such as reduced visual acuity (Sharpe et al, 1999).

5) Celebral achromatopsia

Celebral achromatopsia is not caused by a cone malfunction, but by celebral cortex injuries (Bouvier et al, 2006) secondary to cortical trauma, fever, diseases (tumors). In this case, however, a loss of color perception is not associated with a reduction in visual acuity.

1.2.4. Treatment of manifestations

People with achromatopsia don't have a normally functioning and active cone system and are forced to use ophtalmic or contact filtered lenses to adapt to all levels of higher light intensity to those of twilight. In those patients vision worsen proportionally to the increase in luminous intensity.

1.3. Contact lenses for Achromatopsia

Although there are no possible medical treatments or therapies for achromatopsia, there is the possibility to correct the disease and to improve color vision using special colored contact lenses (CL).

These lenses were avaiable only in a hard material and were very uncomfortable in the past. Now, they can be found in soft materials as an alternative to filtered opthalmic lenses. They give much more comfort and can be customized to perfectly fit every achromatic patient.

The geometric properties of the lenses of each patient must be defined according to different parametres and ensure a correct level of light intensity, the best filtering color and the right coloring area. The psychological and cosmetic benefits that these lenses allow to obtain are very significant and positive for people suffering from achromatopsia. With this solution, patients no longer have to worry about wearing protective but unaesthetic glasses.
In Figure 1.1 is possible to observe a young girl affected by achromatopsia. In the photo on the left you can notice the facial expression and the discomfort she feels when she doesnt't wear colored contact lenses. Notice the strong wrinkle of the eyelids used to reduce the amount of light that comes into her eyes.

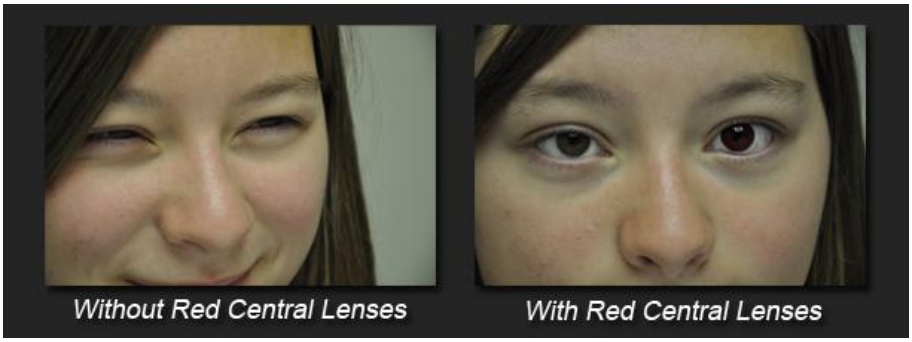

Figure 1.1. Girl with achromatopsia without (left) and with filtered contact lenses (right).
Source: www.achromatopsia.com

In the picture on the right she is wearing colored contact lenses and she is able to keep the eyes open in a natural way. It's important that the level of dimming of colored lenses doesn't limit vision in lighter environments. The goal is to reduce glare, allowing a comfortable vision under every light conditions and into every environment. Today it's possible to determine and customize all types of filter to apply to the lenses.

1.3.1. Benefits of filtered contact lenses

Colored contact lenses may provide a series of benefits and important changes in many situation of everyday life of the achromatic patient. Using that device, their life will probably be easier, safer and more advantageous.

Here is a list of those benefits:

1. Elimination of the debilitating inside glare

Contact lenses are much more reliable in controlling light and glare than eyewear with filters. With the use of tinted eyewear, the vision still fluctuates when light comes out of the edges of the frame. Contact lenses are fitted directly on the eye to fully block the light that could enter in the retina and damage the visual system.

2. More stable vision

Using a proper filtered contact lenses is possible to control the light in a much more efficient and stable way than using other devices. Vision is much more stable and safer under different levels of lighting. The constant ups and downs of vision are smoothed out.

3. Better functional visual field and travel vision

When the proper filtered contact lenses are prescribed, the necessity to squint the eyes is dramatically reduced.

This squinting can reduce the visual field to the point of limiting safe travel or can make difficult searching for things such as looking for items in a bright condition. With contact lenses is possible to have a wider visual field. Considering this, individuals may also be more confident in traveling.

4. Better cosmetic appearance

The cosmetic benefits are also very significant. The constant squinting and the use of dark tinted eyewear have a significant impact on those affected by achromatopsia. It's possible to fit the filtered contact lenses from the very first months of life of the achromates. This factor is especially crucial in children who are often the subject of bullying. It's probably that patients under age ten may initially require help from parents in the daily insertion and removal of the lenses. Contact lenses should always be fitted considering the visual and also cosmetic benefits. Patients don't need to wear red contact lenses to gain benefits. In fact, we often use brown lenses rather than red or use the darker NARZ contact lens that don't appear red on the eye.

5. The social and psychological impact

Removing the dark glasses worn inside and the constant squinting, and improving the consistency of vision, it's possible to have a dramatic impact on a person's confidence and self image.

6. Bright sunlight requires sunglasses

Filtered contact lenses allows the achromatic patient to have control of inside light, but the same contact lenses cannot provide full sun protection outside. With filtered contact lenses, the outside glare will be easier to control just with regular sunglasses. If contact lenses are too much dark and are fitted to fully control the outside glare, they would be too dark to be worn inside.

7. Night vision

Filtered contact lenses may be too dark in outside activities at night. The lenses can be revomed and the patient can wear transparent contact lenses or glasses to improve the condition and to have a more comfortable, safe and clear vision.

8. Changing Environmental Lighting

Once an achromat is fitted with filtered contact lenses, the lighting sources at home or work may need to be adjusted slightly higher. What was good without the contact lenses may now result too dark.

9. Color Vision

Filtered contact lenses can sometimes accentuate certain colors in incomplete forms of achromatopsia where some color vision still exists. This depends on the type of filter used and it's sometimes a secondary benefit, but is not possible in many cases.

10. Reading Vision

A control of the light with contact lenses can improve the quality of reading. Contact lenses will not eliminate the near working distance for reading, in fact the acromatis patient who needs to read must still hold reading materials close to have an angular magnification. Achromates may need to add light to read while in the past they may have preferred to read in a very low light level. High powered reading glasses may still be needed for reading.

7. Driving Issues

The achromat may need some help in certain situation of drive, for example when far from the traffic lights or from a signal. Many individuals have developed strategies to handle traffic lights, like using filters to enhancing one color or subtracting another one.

Patients with achromatopsia respond well to magnification with bioptic systems, that is a combination of prescription eyewear with a small telescopic system that allows many achromatic individuals the option of bioptic driving.

The central point, however, is the serious nature of color blindness; the amount of vision loss determines the possibily to be linecsed or not. In presence of complete achromatopsia, patients will have problems with traffic lights, brake lights and difficulties in reading certain colored signs or images.

In case of incomplete achromatopsia, patients still have a small capacity of perceiving colors, and that permitt the color detection of traffic lights or signals with the use of filters and red contact lenses. This may increase the detection of traffic lights and signs, allowing some incomplete achromates to become bioptic drivers. However, there could be certain situations that can cause difficulties in traffic lights recognization and discrimination, like in presence of a traffic light that is backlit by the rising or setting sun.

1. 4. Glare

Light causes a reduction of the retinal image contrast, with a consequent reduction in visual acuity. If we increase brightness without changing the contrast of the image, our eyes will interpretate it all as a loss of contrast sensitivity with a loss of details. This condition leads to a reduction of the visual ability to effect whitening of a picture.
Glare sensitivity, as previously mentioned, is very high in achromates and, in general, the visual acuity is extremely low. Glare occurs as a result of an overexposure of the rods and can be defined as an annoying and debilitating phenomenon of the eye that causes visual difficulties in the presence of a bright sun or artificial and reflected light. This may be caused by the presence, in the visual field, of an area with a very high luminance contrast, compared to the average luminance of the surrounding field (luminance adaptation).

There are two types of glare:

1. Debilitating glare, that consists of an instant worsening of visual function. This type of glare is obtained by looking at an area which increases brightness without changing the contrast of the image, that our eye interprets as a loss of contrast sensitivity.

2. Discomfort glare, that is manifested as a sense of visual discomfort that doesn't always cause severe deficit in vision, but after some time may cause visual fatigue, stress, difficulties in concentration, loss of attention and increased probability of error in different situations.
The "recovery time after a glare" is the value, in seconds, required to recover a sufficient vision after facing an intense increase in brightness that is capable of generating glare. This condition, which makes unable to see centrally for a few seconds / minutes, is due to the saturation of the photoreceptors.

1.5. Backgrounds

The use of filtered contact lenses to treat this kind of disease has been studied and analyizes in different study, mostly with the attempt to compare them to other solutions and devices, like ophtalmic colored lenses. In 1995, Schiefer tried to treat with centrally tinted contact lenses a 9 years old girl suffering from an incomplete achromatopsia. He found out that, besides correcting the ametropia, the use of contact lenses also reduced light exposure and glare.

Park and Sunness (2004) conducted a study on 23 patients with achromatopsia, cone disorders and severe photophobia. Those patients were fitted with absorptive red soft contact lenses, that immediately appeared to solve the avversion to light, and dramatically improved their visual function. Their use allowed also 8 of the patient to become able to drive. He found that red contact lenses successfully relieved the symptom of photophobia in patients with cone disorders.

Few years later, Sharmack et al. (2007) made a study on two patients, affected by reduced visual acuity, complete color blindness, nystagmus and severe photophobia. Both were fitted with centrally tinted contact lenses. Althought they havent't had an important visual acuity improvement, they had an huge reduction in photophobia and light sensitivity. They had a decrease in the dimension of central scotoma, a magnification of pheripheral visual field and had achieved an improvement in visibility of long wavelenght stimuli in bright illumination.

Also Rajak et al. (2006) studied how the use of those lenses could improve the quality of life. They treated three children (5-13 years old) suffering from cone dystrophy, reduced visual acuity and high photophobia with Lunelle ES70 Solaire 70% brown contact lenses. After the use of these lenses, children started to show much more self-confidence and a greater interaction with other kids and no more bullying.

2. THE STUDY

2.1. Purpose

Color blindness is a rare defect of color vision. The cones of achromates work too little or don't work at all, so these people can just rely on the rods. The disease creates many situation of discomfort and that is very disabling and limiting for the patient. A lot of activities that seem to be simple for a normal person, are extremely difficult for the achromates and require a lot of efforts and attentions. Many achromates, from an early age, have to squint the eyelids, to misure and control the amount of light that enters into the retina. Such attitudes and preventive strategies often create discomfort from a psychological point of view.

There is no cure for color blindness. The professional has to find a way to minimize the negative effects of the disease. In the past, the optometrist made use of colored ophtalmic lenses in order to make less painful the exposure to light, to improve contrast, increase visual acuity and to guarantee a normal life to the patient.

One of the solutions that were later identified, is the compensation through the use of colored contact lenses. They allow to compensate the disease, exactly as with the ophthalmic filters, but adding a series of important benefits, aesthetic and functional, to the patient.

The purpose of this work is to analyize how the use of contact lenses can improve the life of patients affected by achromatopsia, especially in school and pre-school ages.

2.2. Procedure and protocol

The fitting procedure consists in three steps (A-B-C):

A. The pre-application phase allows to select the lens that best fits ocular characteristics considering the corneal parameters, refractive status, the conditions of tearing and of use. It is divided into 5 steps:

1. functional visual analysis;
2. evaluation of visual acuity and contrast sensitivity test;
3. corneal topography and acquisition of corneal paramenters;
4. fluoroscopic evaluation;
5. qualitative and quantitative tear film's evaluation;

B. The application phase involves the realization and the delivery of customized contact lenses and the clinical evaluation and test of the same. In case of necessity, I used trial lenses to assess any adjustments and to design and calculate the final lens. During this first period there might be required more trial lenses to define the final solution. The patient had to come to the controls with the lenses worned almost for few hours, and also with the proper containers and glasses usually alternated with contact lenses.

C. The post-fitting phase consists into programmed cheks of the patient and the lenses, every six months after the contact lenses fitting.

Before my study, all partecipants were wearing colored contact lenses for at least one year, and they have been subjected to a questionaire that I developed to determine the degree of satisfaction with the use of contact lenses and how they have been able to improve their lifestyle. After that, I proceeded with the evaluation of the recovery of visual acuity after glare, with and without contact lenses.

In Table 2.1. we can find the questionaire developed by me to evaluate how the subjects who partecipated at this study lived te disease before and after the use of colored contact lenses. The results demonstrate a complete satisfaction (100% of 7 subjects, not 8 because patient no.1 results not evaluable because he is 8 months old).

Before the study, the totality of subjects was using protective sunglasses; this created an umcomfortable situation in the relationship with other people and made them feel different and with an handicap. These difficulties are completely disappeared using of colored contact lenses, and we can see this total satisfaction in Table 2.1.

QUESTIONAIRE	YES	NO
Did you use protective sunglasses to reduce glare before the use of contact lenses?	100%	0%
Did you feel discomfort and difficulties in using that protective device?	100%	0%
Has this discomfort diminished after the use of contact lenses?	100%	0%
Are you satisfied with the use of contact lenses from a psychological point of view and from relationship with other people?	100%	0%
Do you feel more confident about yourself after the use of contact lenses?	100%	0%
Would you recommend another achromate this type of corrective solution?	100%	0%

Table 2.1. Before and after CL using questionaire, with patient's answers.

2.3 Selection of subjects

The data collection was carried out on a group of subjects consists of children of school and preschool age, residents in Verona, made of two girls and six boys, between 8 months and 25 years old.

The recruitment was carried out to the optometric study inside the store "Benetti Optics" (VR), in a range of time which goes from 2012 and 2016. The selected subjects were treated and initially visited by Dr. Elena Gusson, ophtalmologist, Dr.Ilaria Tomaello (orthoptist) at Eye clinics of Borgo Roma and Borgo Trento (VR) hospitals, and by Dr. Menegotti Alessia (orthoptist) and Dr. Stramare Giuliano (opthalmologist), who works at Negrar hospital's (VR) Eye clinic.

After a first general ophthalmologic analysis and the diagnosis of color defiency, they were sent to my study for the treatment of the disease through the use of opthalmic filters and / or contact lenses.

In Table 2.2. there is a scheme of the age, sex and prescription of the patients who partecipated at my study.

That misurations have been made by these professionals, in some cases in a partial sedatiom because of the poor collaboration of patients.

I had to refer to the oculistic prescriptions because achromatic patients are included in the Italian Health National Service, which provides the total refound of the contact lenses's forniture under the visual acuity of 2/10 and the next Acceptance Certificate made in the same Hospital structure.

What I was able to to was select the best typology of contact lenses for each of my patient.

SUBJECTS	AGE	SEX	RX	
			OD	OS
1	8 m	M	+0,00 - 3,25 ax 180°	+0,00 -3,25 ax 180°
2	10	M	-3,5 D	-3,5 D
3	25	M	+2,5 D	+3,5 D
4	8	F	-2,75 -2,75 ax 20°	-2,75 -2,75 ax 160°
5	15	M	+2 -1,25 ax 180°	+2,75 -1,75 ax 170°
6	21	M	-1,5 D	-0,75 D
7	5	M	-9 D	-3 D
8	11	F	Neutral	Neutral

Table 2.2. Description of the sex, age and prescription of the subjects.

2.4. Colored Contact Lenses

The subjects have been treated with colored contact lenses produced by the company Cantor&Nissel, called ChromaGen. In the Table 2.3. there is a description of the parameters of each lenses used for the patient of my study, and in Table 2.4 the product specification of each lens.

Before the use of these lenses, I used Lunelle 70 Essilor, that were the only one possible solution for the achromatic patiens because they were the only lenses similar, in color, to brown 6 and were good for sports.

PATIENT	**RB1 (mm)**		**DIAMETER (mm)**		**TYPE OF LENS**	**PRODUCT**	**COLOUR**
	OD	OS	OD	OS			
1	8,4	8,4	16,0	16,0	Semestral	Hydrolens 67	Brown 8
2	9,0	9,0	14,0	14,0	Semestral	Hydrolens 67	Brown 8
3	8,6	8,6	14,0	14,0	Semestral	Hydrolens 67	Brown 8
4	8,6	8,6	14,0	14,0	Semestral	Hydrocyl 67	Brown 5/8
5	8,8	8,8	14,0	14,0	Semestral	Hydrocyl 67	Brown 7
6	8,6	8,6	14,0	14,0	Semestral	Hydrolens 67	Brown 6
7	8,8	8,8	14,5	14,5	Semestral	Hydrolens 67	Brown 6
8	8,2	8,2	12,0	12,0	Semestral	Hydrolens 67	Brown 6

Table 2.3. Description of the lenses used in the study for every subject

PRODUCT SPECIFICATION						
MATERIAL	**WATER CONTENT**	**PERMEABILITY (Dk)**	**BASE CURVE (mm)**	**DIAMETER (mm)**	**POWER RANGE**	**CENTRE THICKNESS (mm)**
Filcon II 2	67%	30 x 10-11	6.60 to 10.40 (0.20 steps)	11.00 to 17.50 (0.50 steps)	- 30.00D to +30.00D (0.25D steps)	14.50 - 3.00 is 0.14

FEATURES	**BENEFITS**
	large parameter range
spherical blended bicurve front surface	a graduated aspheric lenticular zone ensures minimum thickness in any power
spherical blended bicurve back surface	every prescription optimised for handling charateristics and oxygen trasmissibility
lathe cut	computer controlling lathes ansuring excellent reproducibility

Table 2.4. Characteristics of the lenses used for the study. Source http://www.chromagens.us/

About ChromaGen™

(from http://www.chromagens.us/)

ChromaGen is a system of eight colored haploscopic filters (Figure 2.1.) of a known density and color hue which, when prescribed to sufferers of ASD™, has been proven to improve these disorders. ASD™ is an umbrella term that includes dyslexia, color deficiency, dyspraxia (lack of coordination; clumsiness), and other learning related difficulties. Chromagen Lenses are supplied (ChromagenOphthalmic Lenses and Chromagen Contact Lenses) for Color Blindness (Color Deficiency) and for Dyslexia.

ChromaGen was developed by David Harris at the Corneal Laser Center for Color Blindness at Clatterbridge Hospital in Cheshire, England. Dr. Chaaban Zeidan, an optometrist from Tamworth, England together with Cantor & Nissel further developed ChromaGen for dyslexia, resulting in patent approval in 1997.

Unlike other programs, ChromaGen offers a range of eight colored filters, as opposed to other lenses such as X-chrom and ColorMax, which only offer lenses for those with red/green color blindness and are not FDA approved. This product is available with a prescription in both spectacle and contact lens modality exclusively through ChromaGen Europe for worldwide delivery. With the Chromagen Lenses you can pass the Ishihara Test and other color blindness tests: the Farnsworth and the Lantern Color Vision tests in Employment Test.

ChromaGen™ and Colour Deficiency

Color deficiency, also known as color blindness, is an optical condition in which affected people have a deficiency in one or more of the primary colors (red, green, or blue) recognized through the retina. There are approximately 3.5 million people afflicted with color blindness in the United States. 10% of males and 0.6 to 0.8% of women are color deficient.

Color deficiencies can be divided into the following categories: Deutrans (lack in green receptors), Protans (lack in red receptors), Tritans (lack in blue receptors), Monochromatics (sees only 1 pigment), and Achromatics (has no functioning cones). Defective color vision can range from near-normal ability to distinguish colors, where typically the chemical for seeing red or green is slightly altered, to a high degree of confusion, where the chemical balance is considerably altered. In trials, over 97% of color blind people reported a significant enhancement to their color vision and their everyday lives.

For people suffering from color deficiency or “color blindness”, ChromaGen haploscopic filters work by changing the wavelength of each color going into one or both eyes, which enhances color perception and color discrimination. Improved color vision may also lead to improved safety through earlier identification of hazard warning lights and brake lights on the roads.

Tinted contact lenses (Chromagen™) (Cantor and Nissel) require the affected individual to select a preferred filter from a group of colors ranging across the spectrum. The filters may minimally improve color discrimination. Chromagen™ contact lenses are not FDA approved but have FDA marketing clearance for use in red-green color deficiencies.

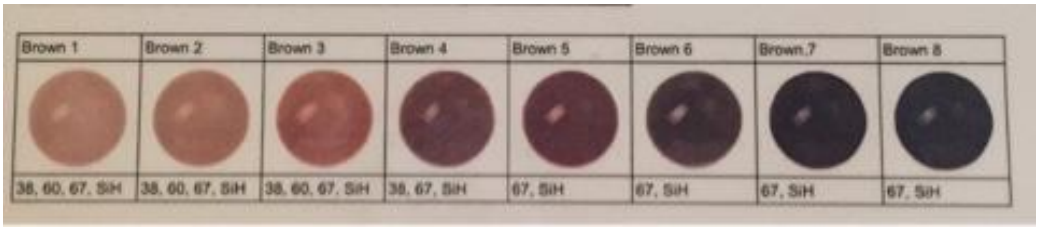

Figure 2.1. ChromaGen lense's tints avaiable on all Hydrocyl, Hydrofit and Hydrolens designs in selected materials. (source: CSD.333| Issue 1 –April2013)

2.5. Recovery time after glare

I decided to submit such test to my patients, with and without the use of colored contact lenses, to evaluate and quantify objectively how the use of the same could reduce the discomfort caused by strong light in achromates subjects .

In Italy, drivers are divided into two groups according to the characteristics of the vehicles for which the driver's licence is required. I decided to take as a guide the test normally used for the evaluation of this aspect of the vision in issuing driving licenses for vehicles in the categories AM, A, A1, A2, B1, B, and BE (Group 1 driving licence).

During the exam for the driving licence, in Italy, the test is made out monocularly, and both eyes are corrected for the possible ametropy. After an adjustment in the dark for a maximum one minute, one eye is covered and a luminous source is located at a distance of 3 cm from the contralateral eye for ten seconds, asking the subject to fix it.
After removing the light source and lighting the room light, is asked to read instantly the line corresponding to 2/10 for licenses for Group 1 in maximum 60 seconds , and the 4/10 line for Group 2 licenses, in maximum 30 seconds.
The line is considered read if 50% of the letters are recognized on time.

During the test, that has been conducted monocularly, I used an electronic eye chart placed three meters from the patient, with Sloan symbols in linear progression, and a light source (penlight) that would produce an illuminance of 200 lux at 20 cm, placed in orthogonal position. The environment remained obscured only during the adjustment period in the dark.

After a period of adaptation in the dark for about one minute, I occluded one eye and put the penlight on the controlateral, for about 10 seconds, asking the patient to fix the light.
Then, I turned on again the lights in the room, and measured the time required to recover the visual acuity of 2/10, in a maximum time of 60 seconds.
I made this procedure two times for every patient, and take the medium value between the two misurations.

As expected, in the case of patients with no colored contact lenses it was not even possible to perform the test under this condition, because of the excessive discomfort caused by the strong light stimulation.

On the contrary, once colored contact lenses have been worn, recovery times have significantly changed. That have provided the possiblility of carrying out the test in all subjects without any discomfort, providing perfectly even results included in the standard maximum values (60 seconds).

I have found a considerable difference in the test made with and without the filtered contact lenses. In fact, 100% of subject (100% of 7, not 8, because the 8 months baby is not evaluable) improved their response. The result show that 100% of achromates (100% of 7, not 8, because the 8 months baby is not evaluable) experience reduced glare comparable to normal vision. This is showed in the Table 2.5.
This test results not evaluable for the patient no.1, because he is 8 months old.

PATIENT N.	RECOVERY TIME AFTER GLARE	
	WITHOUT CL	**WITH CL**
1	Not evaluable	Not evaluable
2	Not evaluable	17 sec
3	Not evaluable	18 sec
4	Not evaluable	13 sec
5	Not evaluable	21 sec
6	Not evaluable	15 sec
7	Not evaluable	17 sec
8	Not evaluable	16 sec

Table 2.5. Results of the recovery time after glare test

2.6. Discussion

Colored contact lenses have been used by patients with achromatopsia for many years to improve their lifestyle and to reduce the negative impact of the symptoms. Reducing the amount of light that reaches the retina, the saturation of the rods is reduced while maintaining a minimal function of the cones.

Filtered contact lenses can largely improve the life of patients affected by achromatopsia, both from a functional and aesthetic point of view. Compared to devidces such as eyeglesses with selective filters, contact lenses allows the patient to limit glare from periphery of the lens and to drammatically reduce the reflexes. The reflection of light in the inner part of the lens can cause further stress. In patients with nystagmus they allow to maintain a correct alignment between visual axis and the optical lens center. This is not possible with other compensating solutions.

Contact lenses can provide greater performance. They can be customized choosng the intensity of color thet better suits the lifestyle of the person. Usually is chosen a medium - intensity color to ensure an optimal use in both high brightness and darker conditions. In particular and high light conditions, the achromate can use both glasses with contact lenses.

These lenses allow to limit the symptoms like photophobia, reduced or complete loss of ability to distinguish colors, reduction in visual acuity and nystagmus. They also improve the lifestyle and reduce the negative psychological impact that the disease has over the people with achromatopsia. The cosmetic benefits of filtered contact lenses are great and have permitted a better and more comfortable life for many patients.

As for the only one patients that could not be evaluated because he was 8 months old, I was able to get informations from his parents. It was not possible to test recovery time after glare and, of course, even evaluate the questionaire.
Parents say, however, that, without filtered contact lenses he was always very much covered in the stroller, with umbrella, hat and dark glasses. After using the contact lenses the baby had much less discomfort and when he come outdoors with his parents, the cap was enough to protect him without trouble.

Before the use of the lenses, the baby always cried when he came out and there was a light. Now things are much more better and he doesn't cry anymore as often as before.

While providing a better and easier lifestyle, contact lenses can potentially damage the structures of the eye surface. That is why the professional should keep in mind that the use of contact lenses still poses risks, so he must make sure that the patient uses the lenses properly in terms of hours of use, cleaning and maintenance, in order to reduce the appearance of complications and to avoid excessive dependence on their use.
The risk of complications are almost reduced by a proper training by the professional to the patient in the manteinance of the contact lenses.

This kind of compensation results, from my research and from most of others author's previous works, the ideal solution for the treatment of achromatopsia.

PATIENT	DIAGNOSIS	DEGREE OF SATISFACTION
1	Megalocornea associated with blepharospasm, photophobia, incomplete achromatopsia	No longer use of penalizing glasses, no more eyes and head down. Less discomfort.
2	Complete achromatopsia	The patient used glasses with selective filters before this study. Much more self-confident
3	Macula on piano with pigmentary retinal systrophy, cone dysfunction, visus with correction 1/20, rotatory nystagmus	Resumed studying after quitting because was uncomfortable in dealing with other people. Now is graduated at University
4	Severe photophobia, achromatopsia	After the use of contact lenses has succeeded to deal with the world of work with greater positivity
5	Blepharospasm, total dystrophy of cones (ERG), complete achromatopsia	More self-confidence and more confident with other people.
6	Total achromatopsia, visus 1/20	The use of CL has permitted a better approach to others and to everyday life.
7	Congenital glaucoma, high photophobia, incomplete achromatopsia, has done several operation	Better head position and posture and higher esteem
8	Late diagnosis because of an hernia, oculo-anathomic albinism, nystagmus, alteration of the pigmented epithelium	Initial discomfort caused by the change of the eye color, from dark blue to dark brown. This has been overcome after the excellent performance and improved quality of life

Table 2.6. Diagnosis (left) and results (right) obtained after using colored contact lenses

2.7. Conclusion

1. After the use of the filtered contact lenses in my study, all subjects give only positive marks on questionaire specially indicating the question about the decrease of discomfort after the use and the satisfaction from a psychological point of view and from relationship with other people.

They all report to feel much more confident about themself and they would recommend another achromate this type of corrective solution.

2. Colored contact lenses allow to limit the symptoms like photophobia, reduced or complete loss of ability to distinguish colors, reduction in visual acuity and nystagmus.

3. Regarding the test for recovery after glare, the result show that 100% of achromates (100% of 7, not 8, because the 8 months baby is not evaluable) experienced reduced glare comparable to normal vision.

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I certify with my signature, that research has been conducted independently, there have been used only in reference list mentioned sources of information, and electronical copy correspond to printed version.

Author: **Claudio Aldegheri**

Student card number: Ca15001

Recommend/unrecommend thesis for defensing

Supervisor: docent, Dr.phys. Sergejs Fomins

Opponent: professor Dr.phys. Maris Ozolins

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Dean’s authorized person: _____

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