

Case Study

Pennsylvania College of Optometry
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**Case Number Three:
Solutions for enhanced comfort in daily-life for a low-vision patient suffering from albinism.**

by
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Abstract:

Albinism is a heterogeneous group of diseases. It is genetically induced with different patterns of inheritance. Ocular findings include photophobia, iris transillumination, nystagmus, high myopia, foveal hypoplasia and fundus hypopigmentation. This case report describes solutions for enhancing vision and vision related tasks for a patient suffering from albinism.

Key words: *albinism, low-vision, longpass edge filtering lens*

Introduction

Albinism is a genetically determined heterogeneous group of disorders involving deficiency in the enzyme tyrosinase, which mediates the conversion of tyrosine to melanin. The two main types are: Oculocutaneous and ocular. Oculocutaneous albinism may be either tyrosinase-negative or tyrosinase-positive.¹

Ocular Albinism:

The pattern of inheritance in ocular albinism is either X-linked or autosomal recessive. The skin and hair in ocular albinism appear normal but still the typical ocular signs and symptoms are present.

By observation, ocular albinism can only be seen at the structures of the eyes. Nevertheless Creel et al, 1974, found abnormal macromelanosomes in the dermis and epidermis in X-linked ocular albinos. No such abnormalities have been reported in the autosomal recessive form which makes the skin biopsy with determination of macromelanosomes a diagnostic tool to find out about the type of disease present.

Oculocutaneous Albinism:

The hereditary pattern of oculocutaneous albinism is usually autosomal recessive. The tyrosinase-negative and the tyrosinase-positive form represent different genotypes.

The tyrosinase-negative type is also called complete albinism. These individuals are incapable of synthesizing melanin. The tyrosinase-positive type is called incomplete albinism. Tyrosinase positive albinos can synthesize variable amounts of melanin and are therefore harder to diagnose.

Tyrosinase-negative:

Have straw- or platinum blond hair and pale skin. Tyrosinase-negatives burn in the sun and never tan. They have typically light blue to pink appearing irides which transilluminate completely.

Tyrosinase-positive:

Has also light hair and eye color. But the color of the hair can darken with age.

In both types ocular findings include: reduced central vision, nystagmus, iris transillumination, decreased pigmentation of the fundus, poor foveal development and ill-defined macular landmarks.²

Hermansky-Pudlak (HP) and Prader-Willi Syndromes (PW):

These two syndromes represent life-threatening combinations with albinism.

HP is the combination of oculocutaneous albinism with defective platelet function leading to an increased bleeding tendency, easy bruisability and gum bleeding. Puerto Rican albinos seem to be at a greater risk for HP. Aspirin use can be life threatening in HP patients, and death can be caused by fibrotic restrictive pulmonary disease.

PW-syndrome is characterized by muscular hypotonia, hypogonadism, obesity, mental retardation, short stature and diminished sensitivity to pain. In 1982 a previously missed component of the PW syndrome was recognized, that of oculocutaneous albinism. Nine patients were found with PW who also had decreased tyrosinase activity with light hair and skin, decreased pigmentation of the iris stroma, and variable amounts of iris translucency. However none had reduced vision, nystagmus, photophobia, or foveal hypoplasia. Therefore the disease was named "albinoidism".²

Case Report

At 9. January 2003 U.B. a 52 year old white female (retired bakery employee) presented to our contactlens-practice with a complaint of strong photophobia, reading difficulties and RGP-lens intolerance. The conditions affected both eyes and existed since several month.

She had a 20 year history of RGP lens wear with no incidents of infections or inflammations. Her last exam with an ophthalmologist was two weeks prior to her visit at our institute. As an infant she was diagnosed with albinism. Otherwise she has no systemic disease and no family history of any eye- or systemic disease. She reported an allergy to penicillin. She was oriented to time, place and person.

Her uncorrected visual acuity was fingercounting @ 150 cm OU. Uncorrected near acuities could not been assessed. Best-corrected visual acuity with glasses was 20/400 OD and 20/200 OS. The refraction was $-1.75 -4.50 @ 8^\circ$ OD and $-4.25 -3.25 @ 175^\circ$ OS at a corneal apex to lens distance of 14mm OU. For reading a monocular add-power of 12dpt (3x) OS resultet in a near visual-acuity of 20/100 (2.0M) which was sufficient for newspaper- and book-reading. A monocular add-power of 20dpt (5x) OS resultet in a near visual-acuity of 20/50 (1.0M) which allowed the patient to read small print such as timetables for short period of times. Pinhole acuity at distance was 20/200 OD and 20/130 OS. Color vision testing with pseudoisochromatic plates showed no color vision deficiency OU. Pupils were equally round and reactive to light, no afferent pupil defect was noted OU. Confrontation fields were full to finger count OU. Extraocular muscles were unrestricted in all gazes, and cover test demonstrated orthophoria at distance and near. Due to the law of practice in Switzerland no Goldman Tonometry could be performed. Anterior segment evaluation by slit lamp examination revealed a quiet bulbar and palpebral conjunctiva OU. An even tear film with tear break up time of 16 seconds OU. Clear lashes OU. The corneas were clear and no staining was noted anywhere. Irides were blue OU and showed a Grade 4 (complete) transillumination upon slitlamp examination. The anterior chamber appeared clear without cells or flare and the anterior chamber angles were estimated by the Van Herrick method with the slit-lamp as 4 nasally and 4 temporally OU. Both lenses were evaluated by slit-lamp with undilated pupils and have been noted as clear with no opacities in any region. A horizontal pendular nystagmus was noted which was reduced with light.

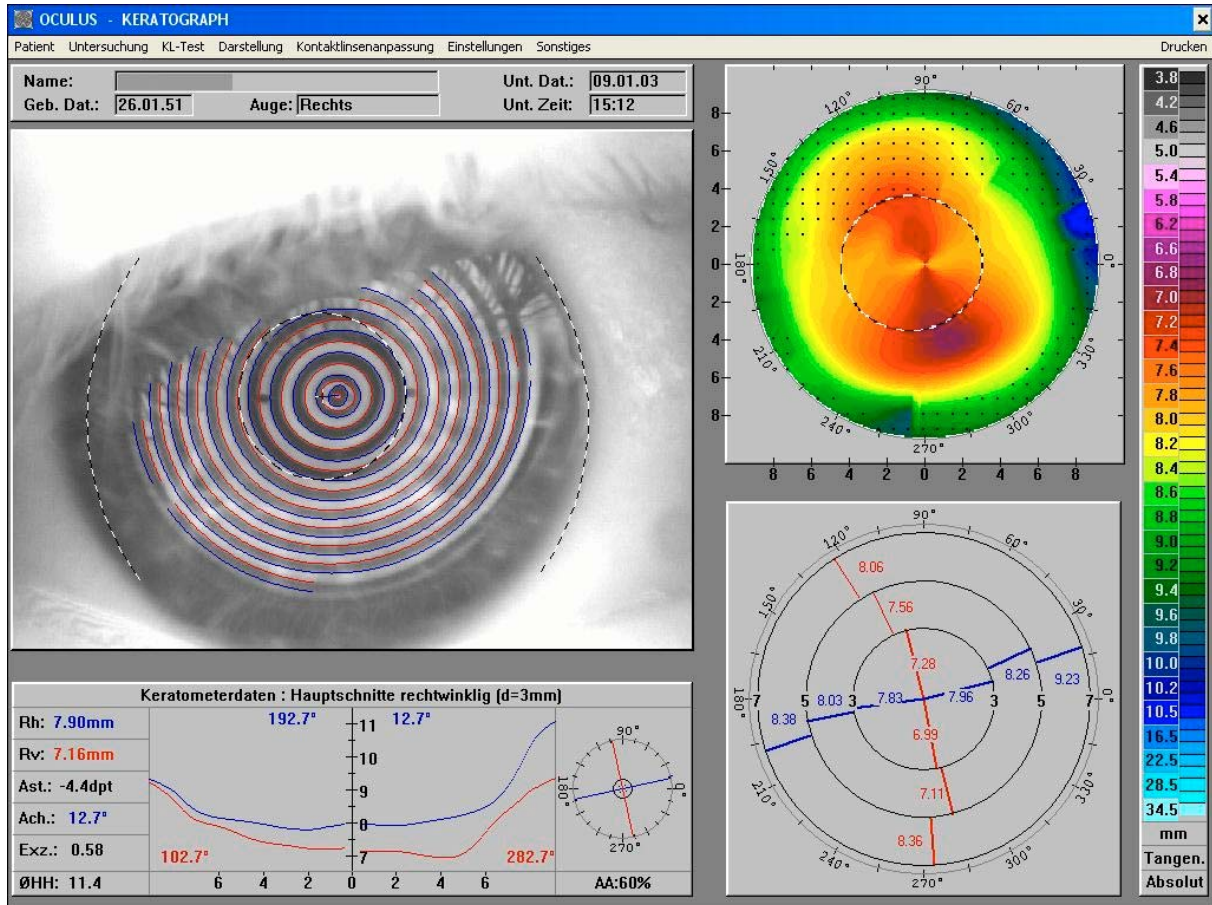
The evaluation of the posterior segment by slit lamp with 90D lens and undilated pupils revealed a optic disc hypoplasia OU with a cup-to-disc ratio of approximately .2/.2 OU. The neuroretinal rims were healthy and intact. A lack of pigment was noted throughout the fundus and prominent choroidal vessels were present. The Retinal vessels showed an arterial-venous ratio of 2/3 OU. Due to the strong nystagmus the macular region could not be observed through the undilated pupils (Optometrists in Switzerland are not allowed to use diagnostic drugs). Usually with this type of disease one would expect that both eyes present with ill defined macular margins, foveal hypoplasia and no foveal reflex.

There has never been an evaluation for low-vision-aids performed with the patient. She was nonetheless using binoculars to orient herself in unknown environments. In addition she was wearing progressive lenses with the following prescription: $-1.75 -4.50 @ 15^\circ$ Add. 3.00 OD, $-3.25 -4.00 @ 180^\circ$ Add. 3.00 OS.

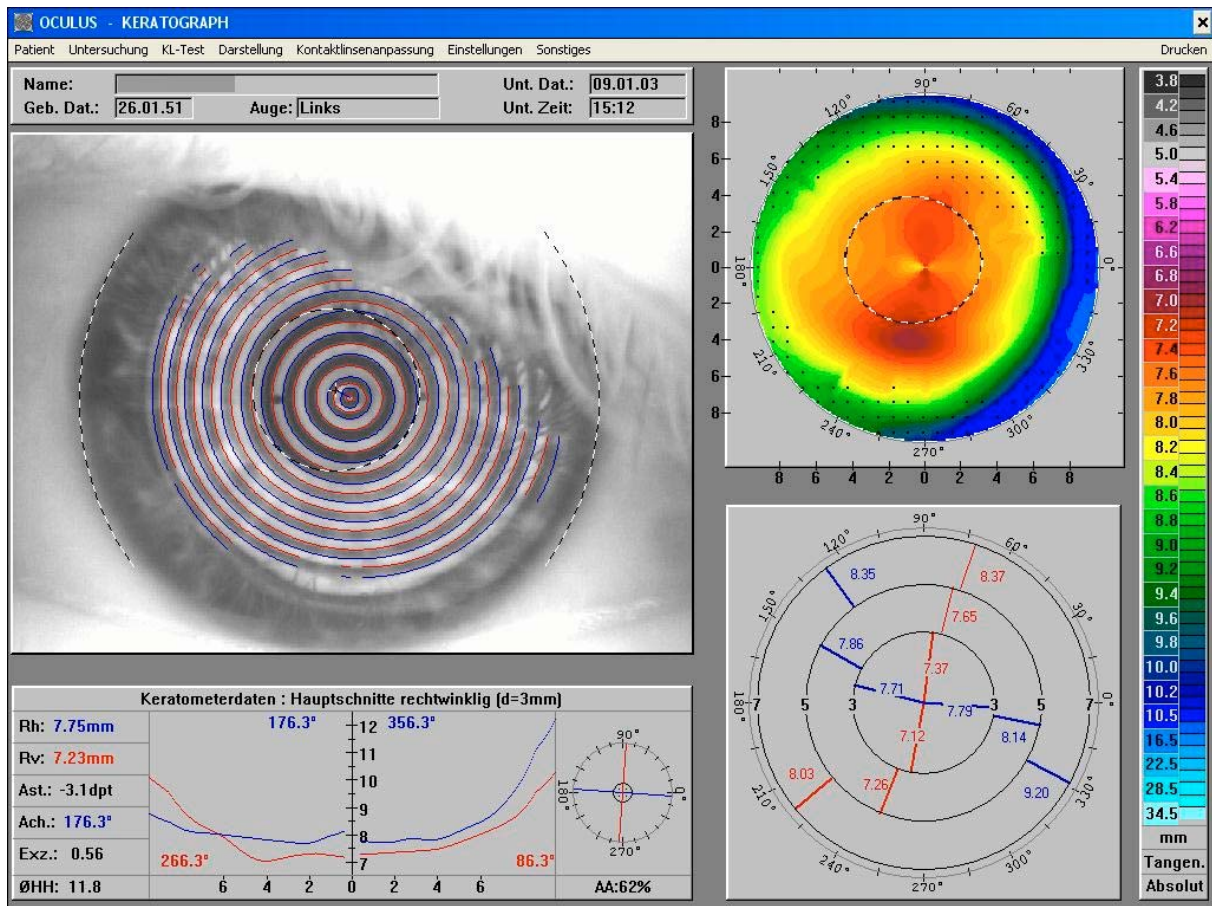
The patient possessed RGP lenses with unknown parameters. According to the patient the lenses were manufactured and fitted in 1997. The lenses have not been worn for several month before this initial exam due to intolerance.

Keratography was performed and revealed the following data:

OD:



OS:



Differential Diagnosis for this case includes the following:

- Aniridia
- Congenital optic atrophy
- Leber's congenital amaurosis
- Achromatopsia
- Pigment dispersion syndrome
- Albinism

Aniridia is a rare bilateral condition which occurs as a result of abnormal neuroectodermal development secondary to a mutation in the PAX6 gene on the short arm of chromosome 11. The degree of aniridia can vary from partial to total absence.¹

Congenital Optic Atrophy is also called hereditary optic atrophy. This is a very rare and heterogenous group of diseases with bilateral optic atrophy as its hallmark. Systemic abnormalities are absent.¹

Leber's Congenital Amaurosis is a group of disorders with onset in infancy or early childhood of severe visual impairment, sluggish pupils, nyctalopia, and nystagmus. May have range of fundus abnormalities from no fundus changes to retinal pigment epithelial granularity, vascular attenuation, tapetal sheen, yellow flecks, salt-and-pepper fundus, chorioretinal atrophy, or a retinitis pigmentosa appearance. Associated with hyperopia, mental retardation, deafness, seizures, skeletal abnormalities, posterior subcapsular cataracts, keratoconus and renal/muscular abnormalities. Mapped to chromosome 17p.³

Achromatopsia is an inherited condition that affects approximately 1 in every 33,000 Americans. It is also known as rod monochromatism. This condition is associated with color blindness, visual acuity loss, extreme light sensitivity and nystagmus. It is a condition found throughout the world with varying incidence. There are two primary forms, the complete achromatopsia and incomplete achromatopsia. Achromatopsia is an autosomal recessive inherited condition.⁴

Pigment Dispersion Syndrome refers to a pathologic increase in the trabecular meshwork pigment, associated with characteristic midperipheral spokelike iris transillumination defects. Normally, the amount of pigment in the trabecular meshwork increases with aging, but does not exceed grade 2. Abnormal pigment dispersion is grade 3 to 4 trabecular pigmentation with increasing radial transillumination defects or increasing corneal endothelial pigmentation with time.⁵

Albinism has already been described in detail above.

Diagnosis

Due to the hair and skin color in addition to the above mentioned ocular signs and symptoms the patient was diagnosed to suffer from tyrosinase-negative oculocutaneous albinism.

Plan

Different steps were to be taken to solve the problems related to the patient's disease.

Glasses/Contactlenses:

Because of the low central visual acuity the patient was most comfortable with variable degrees of eccentric fixation. The rim of her glasses were hindering free head and eye movement for eccentric fixation. Since the RGP lenses have not been well tolerated a custom-made soft-lens fitting was considered.

Far Vision Aid:

It was decided that the binoculars already in use by the patient were sufficient for the required distance-vision tasks.

Near Vision Aid:

Since the patient was unable to drive a car she was obliged to read timetables. In addition a vision aid for regular reading tasks was required. The best solution, evaluated by trial, was the fitting of two different monocular microscopes. Her dominant eye was the left eye. Therefore the left eye was corrected for the near tasks. Two pairs of specialty glasses were ordered. The type used was Eschenbach Noves Mono⁶ which incorporates two different types of plus lenses, one of them diffractive, to reduce aberrations and to reduce the center thickness of the lens. The microscope for regular near work was ordered with a magnification of 3x (12dpt) and the one for timetable-reading with 5x (20dpt.).

Glare reduction:

To reduce glare the patient was wearing regular sunglasses up to date. The patient wasn't comfortable with the sunglasses in many situation because they were too dark. But without sunglasses she was nevertheless very sensitive to light. For the purpose of evaluation a trial set of three edge filtering glasses was ordered. All three filters were longpass filters with a stopband up to 420nm, 530nm or 550nm and a passband with up to 98% transmissibility in the longer wavelength range. The brand name of the lenses were: "HC Geor" with the filtering edge at 420nm (Figure 1), "HC BB80" with the filtering edge at 530nm (Figure 2) and "HC BB86" with the filtering edge at 550nm (Figure 3). All lenses where coated with an superantireflex coating plus UV absorbing coating. HC Geor absorbs 100% of UVA and UVB whereas the other two lenses absorb UV light between 500 and 650nm to 99.9%. The lenses were manufactured in Switzerland by Reize Optics.⁷

For technical details see the charts in the Appendix.



Figure 1



Figure 2



Figure 3

Follow-Up #1, 15. January 2003

Contactlenses:

The data of the soft lenses dispensed were as follows:

TorisL GM3 (dynamic stabilized soft lens with 58% water content)

OD: 8.40 -1.63 -4.00 @ 173° 14.00

OS: 8.40 -4.00 -2.75 @ 180° 14.00

The lens fit was aligned with the cornea with a good movement of the lens after blinking (1.5mm vertically). The axis of lens stabilisation was 10° OD and 180° OS. The visual acuity was measured as 20/130 OD and 20/130+OS. An overrefraction of +1.00 was measured OS and a modified lens was ordered and sent to the patient. The patient was instructed in the handling and care regimen of soft lenses.

Near Vision Aid:

The two glasses (3x and 5x) were dispensed and the patient instructed how to use the devices.

Glare reduction:

Three pairs of filtering glasses were dispensed. The patient was instructed in the use of them.

Follow-Up #2, 31. January 2003

Contactlenses:

The contactlens fit was equal to the previous exam. Visual acuities were measured as 20/130 OD 20/130+ OS. The overrefraction was plano OU. The patient was instructed to follow the care regimen and a recheck was scheduled in one year for a contactlens checkup.

Near Vision Aid:

The patient reported good near vision with both of the reading glasses for the appropriate task. The patient was advised to report if she finds other needs in daily living. In this case other low-vision devices could be tried.

Glare reduction:

The patient reported excellent comfort with the longpass edge filter "HC Geor". The other two filters made too dark of an impression and subjectively reduced vision. She reported to wear the filtering glasses everywhere when leaving the house. A frame was selected and a custom made pair of lenses ordered and fitted into the frame.

Discussion

An albino patient presents to the practitioner with a wealth of problems to be solved. The reduction of visual acuity, especially in the oculocutaneous type, is an extreme restriction for the patient in daily life. Also the problem with glare and visual field restriction in wearers of glasses needs to be addressed. Nevertheless albinism is not a life-threatening disease with a good prognosis.

Conclusion

Due to the multiple factors distressing albino patients several approaches have to be made at the same time.

In albinos contactlenses can be an excellent way of correcting ametropias, especially high myopias and astigmatisms without restricting the patients visual fields. Thus allow the patient to bring the head and eye into the best position for eccentric fixation.

Other Low-Vision aids such as binoculars, telescopes and microscopes have to be introduced to the albinotic patient.

Longpass edge filtering lenses can bring enormous relief to patients with complaints of photophobia. Especially filters with a stopband up to a maximum of 420nm are excellent because not too much light is blocked from reaching the eye but glare is significantly reduced because of the elimination of the short wavelength range of the light spectrum.

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www.lowvision.org/achromatopsia_and_color_blindnes.htm
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www.reize.ch

Appendix

Spectrophotometer Test

Sample :	Date :	5.6.02 15:21
HC GEOR mit DIASAR	Instr. :	Shimadzu
	Name :	dbl-1534



COLOURIMETRIC OBSERVER ACCORDING CIE

D65	2° Observer	x=	0.4166	y=	0.4278	Y=	77.7
	Cielab 1976	L* =	90.64	a* =	3.71	b* =	56.30
	10° Observer	x=	0.4246	y=	0.4237	Y=	74.4
	Cielab 1976	L* =	89.11	a* =	8.43	b* =	55.55

EUROPEAN STANDARD EN 1836 : 1997 EN 8980 : 1997 Pass

Transmittance (D65)	T V	77.7%	Filter category	1	Pass
Red	signal transmittance T sign	97.5%	Recognition of signal light Q	1.25	Pass
Yellow	signal transmittance T sign	92.6%	Recognition of signal light Q	1.19	Pass
Green	signal transmittance T sign	68.9%	Recognition of signal light Q	0.89	Pass
Blue	signal transmittance T sign	58.7%	Recognition of signal light Q	0.75	Pass
Blue light	(380-500nm) T sb	23.6%			
UV	(280-380nm) T SUV	0.0%	(500-650nm) T V min	41.8%	Pass
UVA	(315-380nm) T SUVA	0.0%			Pass
UVB	(280-315nm) T SUVB	0.0%			Pass

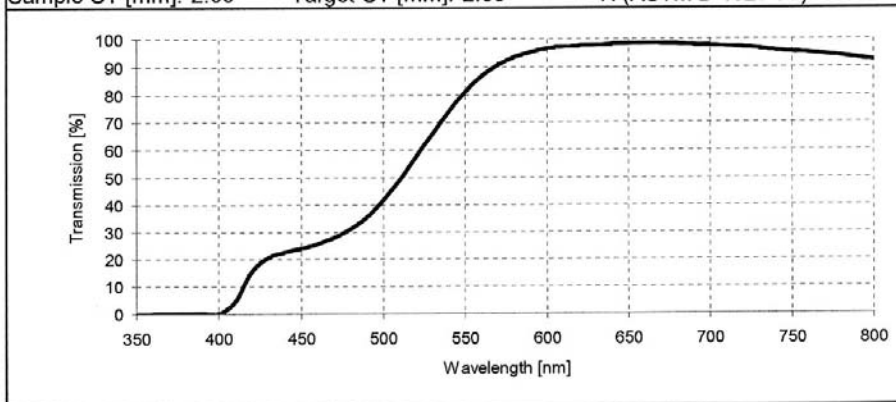
AMERICAN STANDARD ANSI Z80.3-1996 Pass

Transmittance (C)	T V	78.0%	Filter:	Cosmetic Use	Pass
UVA	(315-380nm)	0.0%			Pass
UVB	(290-315nm)	0.0%			Pass
Red	signal transmittance T sign	98.1%			Pass
Yellow	signal transmittance T sign	92.5%			Pass
Green	signal transmittance T sign	68.9%			Pass
Colour distortion			x	y	
	D65		0.4166	0.4278	
	2° Observer	Yellow	0.5836	0.4153	
		Green	0.2698	0.5190	

AUSTRALIAN STANDARD AS 1067.1 - 1990 Fail

Transmittance (C)	T V	78.0%	Filter:	Fashion	Pass
Erythema UV(300-320nm)	T e	0.0%	Red factor	1.26	Pass
Near UV (320-400nm)	T u	0.0%	Violet factor	0.27	Fail

Sample CT [mm]: 2.00 Target CT [mm]: 2.00 YI (ASTM D 1925-70): 85.19



[nm]	[%]
280	0.0
290	0.0
300	0.0
310	0.0
320	0.0
330	0.0
340	0.0
350	0.0
360	0.0
370	0.1
380	0.1
390	0.1
400	0.1
410	4.6
420	15.6
430	20.8
440	22.8
450	24.1
460	25.8
470	28.0
480	31.2
490	35.6
500	41.8
510	49.3
520	57.7
530	66.0
540	74.1
550	81.3
560	87.0
570	91.0
580	93.7
590	95.5
600	96.7
610	97.3
620	97.7
630	98.0
640	98.3
650	98.4
660	98.5
670	98.5
680	98.3
690	98.0
700	97.7
710	97.4
720	97.1
730	96.5
740	95.9
750	95.4
760	95.0
770	94.6
780	94.0
790	93.2
800	92.5

Spectrophotometer Test

Sample :	Date :	5.6.02	15:26
HC BB80 mit DIASAR	Instr. :	Shimadzu	
	Name :	dbl-1535	



COLOURIMETRIC OBSERVER ACCORDING CIE

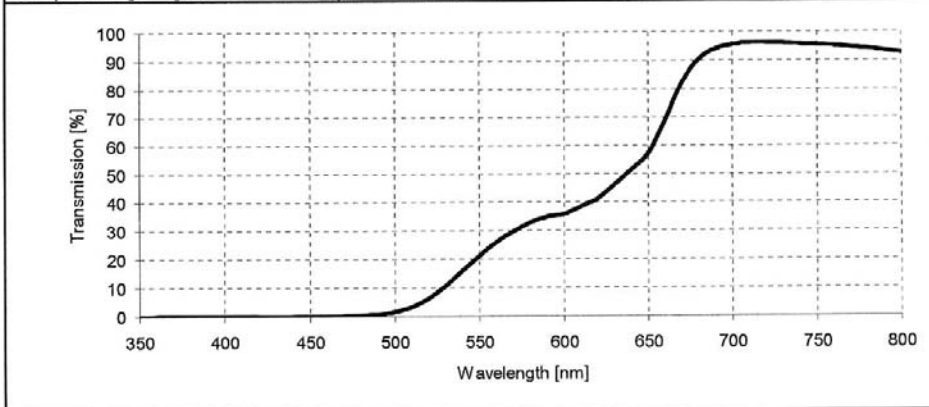
D65	2° Observer	x= 0.5438	y= 0.4481	Y= 22.9
	Cielab 1976	L*= 55.01	a*= 25.97	b*= 91.18
	10° Observer	x= 0.5534	y= 0.4407	Y= 21.3
	Cielab 1976	L*= 53.29	a*= 29.33	b*= 91.69

EUROPEAN STANDARD		EN 1836 : 1997	EN 8980 : 1997	Fail
Transmittance (D65)	T V	22.9%	Filter category 2	Pass
Red	signal transmittance T sign	45.0%	Recognition of signal light Q 1.96	Pass
Yellow	signal transmittance T sign	35.3%	Recognition of signal light Q 1.54	Pass
Green	signal transmittance T sign	15.1%	Recognition of signal light Q 0.66	Pass
Blue	signal transmittance T sign	14.7%	Recognition of signal light Q 0.64	Pass
Blue light	(380-500nm) T sb	0.1%		
UV	(280-380nm) T SUV	0.0%	(500-650nm) T V min 1.7%	Fail
UVA	(315-380nm) T SUVA	0.0%		Pass
UVB	(280-315nm) T SUVB	0.0%		Pass

AMERICAN STANDARD		ANSI Z80.3-1996	Pass
Transmittance (C)	T V	23.2%	Filter: General Purpose Pass
UVA	(315-380nm)	0.0%	Pass
UVB	(290-315nm)	0.0%	Pass
Red	signal transmittance T sign	53.4%	Pass
Yellow	signal transmittance T sign	35.1%	Pass
Green	signal transmittance T sign	15.3%	Pass
<u>Colour distortion</u>		x	y
	D65	0.5438	0.4481
2° Observer	Yellow	0.6113	0.3879
	Green	0.3851	0.5926

AUSTRALIAN STANDARD		AS 1067.1 - 1990	Fail
Transmittance (C)	T V	23.2%	Filter: General Purpose Pass
Erythema UV(300-320nm)	T e	0.0%	Red factor 2.17 Pass
Near UV (320-400nm)	T u	0.0%	Violet factor 0.00 Fail

Sample CT [mm]: 2.00 Target CT [mm]: 2.00 YI (ASTM D 1925-70): 154.28



[nm]	[%]
280	0.0
290	0.0
300	0.0
310	0.0
320	0.0
330	0.0
340	0.0
350	0.0
360	0.0
370	0.1
380	0.1
390	0.1
400	0.1
410	0.1
420	0.0
430	0.0
440	0.0
450	0.1
460	0.1
470	0.2
480	0.3
490	0.7
500	1.7
510	3.4
520	6.4
530	10.8
540	16.3
550	21.4
560	26.0
570	29.9
580	33.0
590	34.9
600	36.0
610	38.6
620	41.4
630	46.3
640	51.7
650	57.3
660	69.0
670	82.4
680	90.5
690	94.1
700	95.7
710	96.2
720	96.3
730	96.0
740	95.7
750	95.4
760	95.0
770	94.6
780	94.1
790	93.4
800	92.7

Spectrophotometer I est

Sample :	Date :	5.6.02	15:29
HC BB86 mit DIASAR	Instr. :	Shimadzu	
	Name :	dbl-1536	



COLOURIMETRIC OBSERVER ACCORDING CIE

D65	2° Observer	x=	0.5648	y=	0.4330	Y=	14.0
	Cielab 1976	L*=	44.26	a*=	28.90	b*=	86.67
	10° Observer	x=	0.5740	y=	0.4250	Y=	13.0
	Cielab 1976	L*=	42.74	a*=	31.70	b*=	88.12

EUROPEAN STANDARD EN 1836 : 1997 EN 8980 : 1997 Fail

Transmittance (D65)	T V	14.0%	Filter category	3	Pass
Red	signal transmittance T sign	29.2%	Recognition of signal light	Q 2.09	Pass
Yellow	signal transmittance T sign	23.4%	Recognition of signal light	Q 1.67	Pass
Green	signal transmittance T sign	7.8%	Recognition of signal light	Q 0.55	Fail
Blue	signal transmittance T sign	8.0%	Recognition of signal light	Q 0.57	Pass
Blue light	(380-500nm) T sb	0.0%			
UV	(280-380nm) T SUV	0.0%	(500-650nm) T V min	0.1%	Fail
UVA	(315-380nm) T SUVA	0.0%			Pass
UVB	(280-315nm) T SUVB	0.0%			Pass

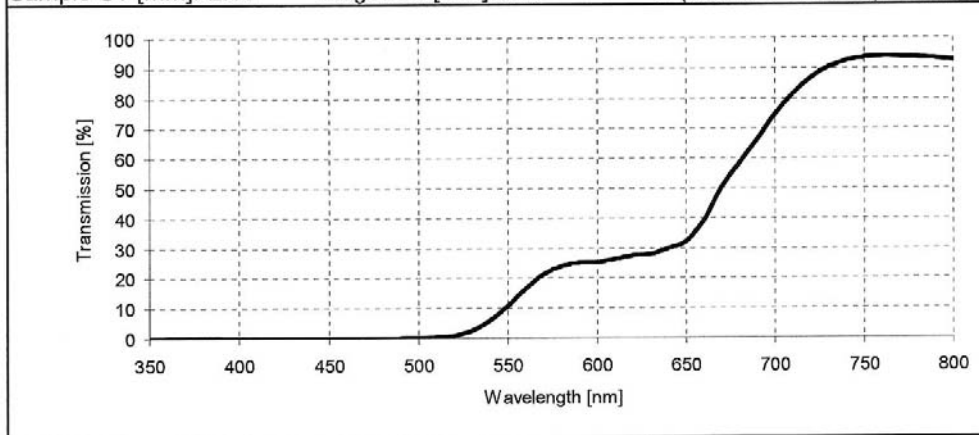
AMERICAN STANDARD ANSI Z80.3-1996 Pass

Transmittance (C)	T V	14.3%	Filter:	General Purpose	Pass
UVA	(315-380nm)	0.0%			Pass
UVB	(290-315nm)	0.0%			Pass
Red	signal transmittance T sign	32.8%			Pass
Yellow	signal transmittance T sign	23.2%			Pass
Green	signal transmittance T sign	8.1%			Pass
Colour distortion			x	y	
	D65		0.5648	0.4330	
	2° Observer	Yellow	0.6103	0.3890	
		Green	0.4376	0.5565	

AUSTRALIAN STANDARD AS 1067.1 - 1990 Fail

Transmittance (C)	T V	14.3%	Filter:	General Purpose	Pass
Erythral UV(300-320nm)	T e	0.0%	Red factor	2.18	Pass
Near UV (320-400nm)	T u	0.0%	Violet factor	0.00	Fail

Sample CT [mm]: 2.00 Target CT [mm]: 2.00 YI (ASTM D 1925-70): 166.27



[nm]	[%]
280	0.0
290	0.0
300	0.0
310	0.0
320	0.0
330	0.0
340	0.0
350	0.0
360	0.0
370	0.1
380	0.1
390	0.0
400	0.0
410	0.0
420	0.0
430	0.0
440	0.0
450	0.0
460	0.0
470	0.0
480	0.0
490	0.0
500	0.1
510	0.3
520	1.0
530	2.6
540	6.0
550	11.0
560	16.6
570	21.4
580	24.3
590	25.3
600	25.4
610	26.6
620	27.7
630	28.1
640	30.2
650	32.4
660	39.3
670	50.5
680	58.5
690	66.4
700	74.8
710	81.7
720	87.0
730	90.6
740	92.6
750	93.5
760	93.9
770	93.8
780	93.6
790	93.2
800	92.7