



**Bachelorarbeit des Studiengangs
Augenoptik und Hörakustik**

Katharina Breher

**Visual Consequences
Of Albinism**

Prüfer: Vertr.prof. Dr. Steffen Kreikemeier

(Hochschule Aalen, Deutschland)

**Zweitprüfer: Dr. Willard Bleything,
Distinguished University Professor of
Optometry and Public Health**

(College of Optometry, Pacific University,
Oregon, USA)

Abstract

Purpose: The purpose of this thesis is to provide a comprehensive literature review about albinism as an inherited metabolic disorder of melanin synthesis along with those related conditions impacting the visual system. As such, it addresses eye care emphasizing the visual consequences of albinism along with diagnostic and treatment options.

Methods: Background knowledge about ocular development is given as well as information about etiological biochemical and genetic processes. The current classification, clinical findings and their assessment and management options are presented based on recent results of research. In conclusion, two case reports are described as examples of visual care options.

Results: Melanin plays a big role in the retinal and chiasmal development. Melanin biosynthesis can be disrupted by different genes in various ways which leads to the current classification of albinism. Clinical findings include fundus hypopigmentation, nystagmus, iris transillumination, photophobia, foveal hypoplasia, excessive chiasmal decussation, reduced visual acuity, high astigmatism (with-the-rule), strabismus and decreased stereopsis. Treatment options to improve visual acuity, fixation and binocularity are (tinted) prescription lenses and contact lenses, low vision aids, surgical procedures and vision therapy. Medication and supplementation for increased pigmentation are currently being tested on mice.

Conclusions: Albinism is caused by genetic mutations resulting in ocular and cutaneous hypopigmentation. It establishes various phenotypes that require different therapy approaches in order to improve vision and therefore quality of life.

Keywords: albinism; vision; genes; melanin; vision therapy; low vision

Vision therapy and biofeedback

Vision therapy (VT) is a relatively new approach to the visual consequences of albinism including treatment procedures for fixation, accommodation and vergence, binocularity and eventually stereopsis. A main goal here is to improve reading skills and to develop depth perception, necessary in many daily living requirements. The therapy procedures are similar to those used in non-albinotic children, however, larger targets are used when necessary. VT for albinotic infants younger than 2 years of age consists of visual stimulation with high contrast toys. These toys should be placed in different directions of gaze to lower the risk of strabismus. Parents should also encourage their child to reach for objects. Visual perceptual training with puzzles, books and games is particularly helpful in children older than 2 years.⁹³ For older children and even adults, Table 7 gives an overview about various VT procedures and the visual skills they train as primary or secondary aim. For the description or other possible training methods see “Applied Concepts of Vision Therapy” by Leonard Press.⁹⁷

ophthalmopathy but worse than in cataract, glaucoma or early macular degeneration. Notwithstanding, individuals with albinism are able to live independently and feel well accepted in society.¹⁰⁷ Kutzbach et al also evaluated the neuro- and motor development including school performance in children with albinism. Their findings reveal that the majority of children have normal balance, fine and gross motor skills despite their visual impairment. Nevertheless, there are some difficulties in motor coordination, such as eye-hand coordination resulting, most likely, from nystagmus and strabismus.¹⁰⁸ Furthermore, the research group showed a higher prevalence of attention deficit hyperactivity disorder (ADHD) in albinism whereas the association to albinism is unknown and more research is required.¹⁰⁹ Still, most of the evaluated children perform at grade level in math and even reading. However, studies emphasize the importance of support in the form of special education “vision impaired” teachers, individualized education programs and more time for visually based tasks.^{108,110}

One of the most important elements for affected children is parental support. They can only understand and accept their situation if their parents do. Therefore, it is important that parents reach out for further information and interdisciplinary counseling, such as in health care, support groups, books or the internet. This also avoids the risk of underestimating their kid’s ability concerning school, sports or everyday life tasks. Moreover, informed parents know how to answer somewhat provoking questions from strangers, e.g. about stereotypes with red eyes, white hair and skin. Thus, they have to act as examples for their children as to their behavior in difficult situations. In summary, parents play a big role in building their child’s self-esteem and their capabilities. This knowledge is the most valuable and important part for a successful life with albinism and all the consequences.^{111,112}

Currently, the patient's right eye was undercorrected but she was using it as her preferred eye at near which creates a monovision situation. This strategy did not interfere with stereopsis (assessed with Lang Test) development since it was completely absent. The mother also reported reduced squinting and nystagmus with the CL that are worn 10 hours per day. Motility was normal, as well as confrontation visual fields. Nystagmus was measured by objective observation. Cover test revealed no phoria and tropia. In kindergarten, the patient started training with low vision aids and iPads. The mother reported that the new school will also have electronic textbooks. Due to reasons of time and moving away, a dilated fundus exam was not possible at this visit and clinic. A follow-up on the CL fitting and comprehensive pediatric examination was recommended in 6 months.

Chapter 8: Conclusion

In summary, research about albinism has progressed considerably in the last few decades but there are still unknown facts about certain aspects, especially about the exact etiology and the general management of visual consequences. However, research in the following areas show promise:

- High-tech low vision devices such as electronic tablets and visual stabilization devices for nystagmus
- Electronic tablets with games for the enhancement of visual skills and/or treatment of vergence range disorders, strabismus, amblyopia, binocularity and stereopsis
- Medication and supplementation for increased pigment production in children, adults and unborn affected individuals
- Vision therapy in order to enhance overall visual skills leading to a more stable visual system, such as accommodative facility/endurance and vergence ranges, leading to better near work skills. Also, as shown in section 7.1, vision therapy could be a helpful tool to increase binocularity and stereopsis. Further research with utilizing an appropriate sample size is required to show the benefits of these procedures.

To sum up, once albinism as a metabolic disease is diagnosed in early years, affected individuals can access a broad management spectrum; this is to be expanded in the future.

OCA	oculocutaneous albinism
OCT	optical coherence tomography
OD	right eye
OKN	optokinetic nystagmus
ONH	optic nerve head
ONL	outer nuclear layer
OPL	outer plexiform layer
OS	left eye
QOL	quality of life
R	rods
RGC	retinal ganglion cells
(R)PE	(retinal) pigment epithelium
TRP-1/TRP-2	tyrosinase-related protein 1 and 2
UV	ultraviolet
VEP	visually evoked potentials
VOR	vestibule-ocular reflex
VT	vision therapy

-
- Figure 19: Left photo shows a Grade 1, light ONH. The right fundus photograph presents a Grade 4, small ONH. Note the tilted optic disc syndrome with Grade 4.⁴⁰ 44
- Figure 20: Relationship of angle kappa with the displacement of the corneal reflex.⁸³ 46
- Figure 21: „The general course of a low-vision examination that forms the foundation of a low-vision rehabilitation model.“⁹³ 52
- Figure 22: Comparison between pre- and postoperative nystagmus. The foveation time increased from 16ms to 70ms whereas the frequency did not change.²⁰ 57
- Figure 23: OCT scans from right and left eye without foveal pit. A little change in the photoreceptor layer is notable where the pit should be. 62
- Figure 24: Grade 2, circular iris transillumination under slit lamp examination. 62
- Figure 25: Fundus photos from right and left eye. Note the absent fovea, the granular macular pigment and the slightly tilted, light ONH with vessel tortuosity. 62

List of tables

Table 1: Mutation overview of the most common types of ocular and oculocutaneous albinism. Adapted from: ²⁷	19
Table 2: Brief overview about the prevalence, location of appearance and clinical features of different forms of OCA. Adapted from: ³⁰	26
Table 3: Grading system for macular transparency. ¹⁷	34
Table 4: Grading system for foveal hypoplasia. Adapted from: ⁶⁷	36
Table 5: Grading system for iris transillumination. ¹⁷	39
Table 6: Grading system for ONH dysplasia in albinism. ⁴⁰	44
Table 7: Overview about VT procedures that can be used to improve visual skills in albinism. “P” indicates the primary aim of the exercise, whereas “S” indicates the secondary aim.	55
Table 8: Subjective and objective refraction results.	60
Table 9: Home-therapy for daily procedures.	63
Table 10: Pre- and post-treatment results of the evaluated visual functions after 5 weeks.	64
Table 11: CL parameters, dry over-refraction and aided distance VA.	66

