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The eyes in albinism: clinical characteristics and their contribution to visual function

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THE EYES IN ALBINISM

Clinical characteristics and their
contribution to visual function

Charlotte Corinne Kruijt

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LIST OF ABBREVIATIONS

CC	Chiasmal Coefficient
CHS	Chédiak-Higashi Syndrome
CSNB II	Congenital Stationary Night Blindness type 2
DCT	Dopachrome tautomerase
DHI	Dihydroxyindole
DHICA	Dihydroxyindole carboxylic acid
FAZ	Foveal Avascular Zone
FHONDA	Foveal Hypoplasia Optic Nerve Decussation defects and Anterior segment dysgenesis
fVEP	Flash Visually Evoked Potentials
GCL	Ganglion Cell Layer
HPS	Hermansky-Pudlak Syndrome
hh fVEP	handheld flash VEP, high frequency (2.5 Herz) flash VEP with a hand held device
IIN	diopathic Infantile Nystagmus
INL	Inner Nuclear Layer
IPL	Inner Plexiform Layer
MATP	Membrane-Associated Transporter Protein
NAF/NAFX	Nystagmus Acuity Function/Expanded Nystagmus Acuity Function
NGS	Next-Generation Sequencing
OA	Ocular Albinism
OCA	Oculocutaneous Albinism
OCT	Optical Coherence Tomography
ONL	Outer Nuclear Layer
OPL	Outer Plexiform Layer
PEDF	Pigment Epithelium Derived Factor
PME	Premelanosomes protein
poVEP	pattern-onset VEP
PWA	People or patients with albinism
RNFL	Retinal Nerve Fiber Layer
RPE	Retinal Pigment Epithelium
sfVEP	standard flash VEP
VA	Visual Acuity
VEP	Visually Evoked Potentials
