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Inherited retinal degenerations: clinical characterization on the road to therapy

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Citation

Talib, M. (2022, January 25). *Inherited retinal degenerations: clinical characterization on the road to therapy*. Retrieved from <https://hdl.handle.net/1887/3250802>

Version: Publisher's Version

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Note: To cite this publication please use the final published version (if applicable).

**INHERITED RETINAL DEGENERATIONS:
CLINICAL CHARACTERIZATION ON THE ROAD TO THERAPY**

**by
Mays Talib**

ISBN | 978-94-6423-566-1

Design Cover | Véronique Baur – www.veerillustratie.nl

Lay-out | Bregje Jaspers – www.ProefschriftOntwerp.nl

Printing | ProefschriftMaken, Vianen, The Netherlands

The research described in this thesis was financially supported by

Curing Retinal Blindness Foundation, Janivo Stichting, Bayer, Stichting Blindenhulp

Financial support for the printing of this thesis was kindly provided by

Stichting Leids Oogheelkundig Ondersteuningsfonds, Rotterdamse Stichting Blindenbelangen, Stichting Blindenhulp, Landelijke Stichting voor Blinden en Slechtzienden, Universitaire Bibliotheken Leiden, Vitaminen op Recept, Oculenti Contactlenspraktijken, Revoir/Ergra Low Vision, Laservision Instruments B.V., Medical Workshop B.V., Bayer, Theapharma, Santen, Synga Medical, Tramedico B.V., Santen Pharmaceutical, D.O.R.C., ChipSoft, Low Vision Totaal.

All funding organizations provided unrestricted grants and had no role in the design or conduct of this research described.

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**INHERITED RETINAL DEGENERATIONS:
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Proefschrift

ter verkrijging van
de graad van doctor aan de Universiteit Leiden
op gezag van rector magnificus prof. dr. ir. H. Bijl,
volgens besluit van het college voor promoties
te verdedigen op dinsdag 25 januari 2022
klokke 13:45
door

Mays Talib

Geboren te Al-Karg, Irak
In 1990

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Voor mijn ouders

LIST OF ABBREVIATIONS

ACD	anterior chamber depth
ADRP	autosomal dominant retinitis pigmentosa
AF	autofluorescence
ARRP	autosomal recessive retinitis pigmentosa
AZOR	acute zonal occult outer retinopathy
BCVA	best-corrected visual acuity
BCEA	bivariate contour ellipse area
CF	counting fingers
CFC	cystoid fluid collections
CFT	central foveal thickness
CHM	choroideremia
CI	confidence interval
CME	cystoid macular edema
COD	cone dystrophy
CORD	cone-rod dystrophy
CRB1	Crumbs 1
CRISPR	clustered regularly interspaced short palindromic repeats
CRT	central retinal thickness
CSL	cone sensitivity loss
DA	dark-adapted
dB	decibel
DNA	deoxyribonucleic acid
ELM	external limiting membrane
EMA	European Medicines Association
ERG	electroretinography
ERM	epiretinal membrane
ETDRS	Early Treatment Diabetic Retinopathy Study
EZ	ellipsoid zone
FAF	fundus autofluorescence
FAZ	foveal avascular zone
FD	flow density
FDA	Food and Drug Administration
FfERG	full-field electroretinography
FST	full-field stimulus testing
FTMH	full-thickness macular hole
GCL	ganglion cell layer
GI	genetic isolate
GVF	Goldmann visual field
HM	hand motion

ILM	inner limiting membrane
INL	inner nuclear layer
IPL	inner plexiform layer
iPSC	induced pluripotent stem cells
IQR	interquartile range
IRD	inherited retinal dystrophy
ISCEV	International Society for Clinical Electrophysiology of Vision
IZ	interdigitation zone
LA	light-adapted
LCA	Leber congenital amaurosis
LogMAR	logarithm of the minimal angle of resolution
LP	light perception
LRAT	Lecithin:retinol acetyltransferase
MAIA	macular integrity assessment
MD	mean deviation
MP	microperimetry
NLP	no light perception
OCT	optical coherence tomography
OCTA	optical coherence tomography angiography
ONL	outer nuclear layer
OPL	outer plexiform layer
ORF15	open reading frame 15
P-AI	participation and activity inventory
PPRPE	para-arteriolar preservation of the retinal pigment epithelium
PSD	pattern standard deviation
RD	retinal dystrophy
REP-1	Rab escort protein 1
RHO	rhodopsin
RP	retinitis pigmentosa
RPA	retinitis punctata albescens
RPE	retinal pigment epithelium
RPE65	RPE specific protein 65 kDa
RPGR	retinitis pigmentosa GTPase regulator
SD	standard deviation
SD-OCT	spectral domain optical coherence tomography
SE	standard error
SER	spherical equivalent of the refractive error
SPC	subcapsular posterior cataract
VA	visual acuity
VF	visual field
XLRP	X-linked retinitis pigmentosa

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