STATE BOARD OF OPTOMETRY
2450 DEL PASO ROAD, SUITE 105, SACRAMENTO, CA 95834
P (916) 575-7170 F (916) 575-7292 www.optometry .ca.gov

Continuing Education Course Approval Checklist

Title: Macular Dystrophies with DRUSEN

Provider Name: NVISION Eye Centers c/o Ms. Leslie Kuhlman

☑Completed Application
Open to all Optometrists? ☑Yes ☐No
Maintain Record Agreement? ☑Yes ☐No

☑Correct Application Fee

☐Detailed Course Summary

☑Detailed Course Outline

☑PowerPoint and/or other Presentation Materials

☑Advertising (optional)

☑CV for EACH Course Instructor

☑License Verification for Each Course Instructor
Disciplinary History? ☐Yes ☑No

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CONTINUING EDUCATION COURSE APPROVAL APPLICATION

$50 Mandatory Fee

Pursuant to California Code of Regulations (CCR) § 1536, the Board will approve continuing education (CE) courses after receiving the applicable fee, the requested information below and it has been determined that the course meets criteria specified in CCR § 1536(g).

In addition to the information requested below, please attach a copy of the course schedule, a detailed course outline and presentation materials (e.g., PowerPoint presentation). Applications must be submitted 45 days prior to the course presentation date.

Please type or print clearly.

Course Title	**Course Presentation Date**
Macular Dystrophies with Drusen	03/05/2017

Course Provider Contact Information

Provider Name

Leslie (First) Kuhlman (Last) Ann (Middle)

Provider Mailing Address

Street 75 Enterprise **City** Aliso Viejo **State** CA **Zip** 92673

Provider Email Address Leslie.Kuhlman@nvisioncenters.com

Will the proposed course be open to all California licensed optometrists?	☑ YES ☐ NO
Do you agree to maintain and furnish to the Board and/or attending licensee such records of course content and attendance as the Board requires, for a period of at least three years from the date of course presentation?	☑ YES ☐ NO

Course Instructor Information

Please provide the information below and attach the curriculum vitae for each instructor or lecturer involved in the course. If there are more instructors in the course, please provide the requested information on a separate sheet of paper.

Instructor Name

Kent (First) Small (Last) W. (Middle)

License Number	**License Type** M.D.
Phone Number (310) 560-9121	**Email Address** Kentsmall@hotmail.com

I declare under penalty of perjury under the laws of the State of California that all the information submitted on this form and on any accompanying attachments submitted is true and correct.

Signature of Course Provider *Date* 3/1/17

OPTOMETRY

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For Office Use Only

Receipt No. _____

ATS No. _____

Date Rec'd _

Request for Approval of Continuing Education Course(s)

Leslie Kuhlman
NVISION Eye Centers
75 Enterprise, Suite 200
Aliso Viejo, CA 92656

Requests for approval of continuing optometric education (CE) courses should be submitted on this form. The California State Board of Optometry requires the following information in order to process a course approval request:

- **$50** processing fee
- Name of provider
- Course title(s)
- Date(s) the course is scheduled to be offered
- Topical outline of the course subject matter
- Any announcements, notices or advertisements of the course
- Curriculum vitae (CV) of all instructors and lecturers involved (NOTE: CVs should include every term of employment, academic credential, publication, contribution or significant achievement)

Requests for approval and the supplemental information should be submitted to the Board office at least 45 days prior to the first date that the course will be offered. Requests will be reviewed by staff and forwarded to the CE Committee for final review. If necessary, Board staff will contact the requestor for additional information. Course approvals are valid for 12 months or until the course is modified.

The CE Committee's decision(s) will be noted and a copy of this form will be returned to the provider to serve as official notification of approval and/or disapproval of the course(s). Please remember to include the contact person's name and mailing address in the space provided above.

_____

CE Committee Member

YOU'RE INVITED

ORANGE COUNTY REGIONAL
5-HOUR CE EVENT

Sunday, March 5, 2017 / 7:00 am - 1:30 pm
Improv Comedy, Irvine, CA

Join NVISION for an exciting continuing education event including networking, breakfast, lunch and raffles.

FEATURED EVENTS

Exciting Presentations, Fantastic Raffle Prizes,
Vendor Booths, Delicious Food & Drinks,
and Breakfast & Lunch

SPEAKERS

Tom Tooma, MD • Franklin Lusby, MD
Sheri Rowen, MD • John Nolan, MD
Jonathan Pirnazar, MD

TOPICS

LRS, Ocular Nutrition, Crosslinking, Corneal Inlay

Limited availability. Registration ends 3/3/17.
For more information and to RSVP, visit:
https://ocregional5hrce.eventbrite.com

NVISION EYE CENTERS

NVISION
EYE CENTERS

STATE BOARD OF OPTOMETRY
2450 Del Paso Road, Suite 105
Sacramento, CA 95834

On behalf of NVISION Eye Centers, we are writing to request approval of Continuing Education to California doctors of optometry. The education will be delivered by Board Certified Ophthalmologists, clinical investigators and experts in technology and patient consultation.

We are writing in response to your letter for information pursuant to CCR 1536 (g), to address why our application was submitted earlier than 45 days for course accreditation. As well as additional content requested.

The reason why our application was submitted earlier than 45 days for the course named "Macular Dystrophies with DRUSEN" g given March 5, 2017 access to the final presentation of the material being in development prior to the time period needed. Once information required, we moved quickly to process accreditation requests. Please accept our apologies and deepest regrets. Going forward, we will make every effort to process these applications in a timely manner.

Course Description: This course will introduce different types of macular dystrophies with drusen and cover treatment options.

Course Objective: The course will allow optometrists to better identify and properly diagnose macular dystrophies with drusen with the use of difference diagnostic images. It will educate optometrists with available treatments.

Conditions of Availability: This course will be open to all licensed ODs. They will be notified through flyers, Eventbrite, and fax by request.

Records: NVISION Eye Centers to maintain and furnish to the Board and/or attending licensee such records of course content and attendance as required for a minimum of three years.

Professional Advancement: NVISION Eye Centers seeks to offer professional education to local and regional optometrist. As a leading practice in the ophthalmology field, NVISION doctors are engaged in research and latest developments on procedures, technology, and clinical therapies. The field of optometry is constantly evolving at a rapid pace and optometrists need to keep up. All Things Refractive in an interactive presentation. This CE activity will help attending ODs learn a full understanding of refractive surgery technology, clinical treatments and procedures, candidates, post-op & pre-op care, cost, co-management, how it is performed, and benefits.

The contact person for this program is myself, and I can be reached at 949.234.8129 or Leslie.Kuhlman@nvisioncenters.com.

Sincerely,

Leslie Kuhlman
NVISION Laser Eye Centers
Continuing Education and Special Projects Coordinator

NVISION
EYE CENTERS

Presenter – Kent W. Small, M.D.

Course Title – Macular Dystrophies with DRUSEN

CV –

Kent W. Small, M.D.
training and experience
Residency: Duke University
Fellowship, Retina: Duke University
Fellowship, Molecular Genetics: Duke
Director: Retina, Univ of Florida
Professor: Retina, UCLA, Jules Stein
Chairman: Univ. Texas

- **Founder / President: Kent W. Small, M.D**
- **Founder / President: Macula & Retina Institute**
- **Founder / President: Molecular Insight Research Foundation**
- **Scientist: Regenerative Medicine Institute, Cedars-Sinai**

310-659-2200 818-552-5040
Molecular Insight Research Foundation

- **Kent W. Small, MD**
- **30 years of research and clinical experience**
  - **1st to use microsatelites DNA repeats in gene mapping in ophthalmology**
  - **1st to map a macular degeneration in the Human Genome Project**
  - **Found mutations in**
    - **North Carolina macular dystrophy (MCDR1)**
    - **cone-rod dystrophy (CORD5)**
    - **enhanced S-cone (Goldmann-Favre)**
    - **Reis-Buchler corneal dystrophy**
    - **Blepharophimosis syndrome (BPES)**
    - **Reigers Syndrome**
  - **Over 200 peer reviewed scientific publications**
  - **20 years of NIH support**
  - **15 years of Foundation Fighting Blindness support**
  - **CIRM (California Institute for Regenerative Medicine)**
  - **Trained over 100 ophthalmologists / optometrists**

Kent W. Small, M.D.
SOLO PRIVATE PRACTICE
Molecular Insight Research Foundation
Cedar-Sinai Regenerative Medicine Institute
Los Angeles, CA

NVISION
EYE CENTERS

Glendale, CA
KENT W SMALL MD
NO FINANCIAL DISCLOSURES
NO CONFLICTS OF INTEREST

Course Outline –

MACULAR DYSTROPHIES
with DRUSEN
A FEW CONCEPTS
LUMP vs SPLIT
Tremendous variable expression within single families shows the value of LUMPING
FAMILY CONTEXT
ALL DRUSEN ARE ALIKE
Splitting drusen into subcategories is worthless as are most genotype / phenotype studies
NORTH CAROLINA MACULAR DYSTROPHY / PRDM13
Are you a
LUMPER or a SPLITTER
Are you a
LUMPER or a SPLITTER
Lump or Split
Types of Macular Dystrophies
with drusen
Initial approach to a patient with macular dystrophy
bilateral and are usually extremely symmetrical
Most autosomal dominant
one or more living affected relatives
careful family history
Most over the age of 50 misdiagnosed as AMD
Careful medication history past and present / Drug toxicity
autosomal dominant macular dystrophies have visual acuities that are better than one might expect
autosomal recessive (Stargardt disease, ABCA4) acuities that are poorer than one might expect given the relatively mild abnormality of their fundus
Macular Dystrophies
THE CONTEXT OF THE FAMILY IS IMPORTANT
SOMETIMES CRITICAL
Only when other family members are examined can you appreciate the full spectrum of the disease and make the correct diagnosis
Drug Toxicity which can mimic macular / retinal degenerations
Macular Dystrophy?
Macular dystrophy?
"If your ship does not come in...
swim out to it..." Jonathan Winters
Lanai

Kent
Maui channel 10 miles Maui
(Lanai – Maui)
race 10.2 miles
"Treasure Your Exceptions"
William Bateson
8 August 1861 – 8 February 1926
English biologist
first person to use the term genetics to describe the study of heredity
the chief populariser of the ideas of Gregor Mendel
North Carolina Macular Dystrophy MCDR1 / NCMD
my 1st encounter
1987 at the Asheville VA hospital
DUKE rotation
20/50
Hallmark:
MARKED VARIABLE EXPRESSIVITY
bad macular lesion
good vision
1/3 of affected family
remainder asymptomatic
Confused with TOXO
Oteen VA,
Asheville, NC
proband encountered
Lefler Wadsworth Sidbury Syndrome
My initial years
redefined the phenotype

autosomal dominant
- completely penetrant
- congenital
- not progressive

NCMD / MCDR1
Grade 1 (not stages) drusen
NCMD / MCDR1
Grade 2 confluent drusen
Grade 3
coloboma-like defect in retina / choroid / scleral /
Toxo -like
Peripheral drusen only in some patients
ARVO / Sarasota 1989
1982
MCDR1 CHICAGO

Of these terms,
CALDERA MACULOPATHY
the least precise.

"Double, double,
toil and trouble,
Fire burn and caldron bubble."
Macbeth

Lets not stir the pot with yet more inaccurate names
The MANY NAMES of MCDR1
North Carolina Macular Dystrophy
TOO MUCH SPLITTING!
A SINGLE DISEASE IN A SINGLE FAMILY GIVEN ALL OF THESE DISEASE ENTITIES AND NAMES
Lefler Wadsworth Sidbury "Syndrome"
dominant macular degeneration and aminoaciduria
dominant progressive foveal dystrophy
central areolar pigment epithelial dystrophy
central pigment epithelial and choroidal degeneration
Caldera maculopathy
North Carolina Macular Dystrophy
SEEMS LIKE IT IS HUMAN NATURE TO WANT TO SPLIT
North Carolina Macular Dystrophy
Is a bad name
Is a misnomer
Given by Don Gass in his atlas ... and it stuck
Named for its FOUNDER EFFECT
large single family in a stable Southern society, very little inbreeding
found worldwide
Caucasian, Asian, Hispanic, Indigenous, African-American
MCDR1 in Nashville, TN
Cavitation not ideal descriptor
Implies tissue was present which later disappeared (sink hole)
This is a developmental abnormality...tissue never did develop / form
what happens to splitters and
Confederate spies
Extensive genealogical research looking for additional affected individuals
MCDR1: variable expressivity
clinical diagnosis
can be confused with
Dry AMD: drusen
Wet AMD: CNVM / sub-macular fibrosis
Bests macular dystrophy

Toxoplasmosis
RFLPs labor intensive, slow, bi-allelic systems, binary analysis
MFD97 ; D6S249 : Marshfield Marker
Jim Weber
LOD scores (40.03)
highest recorded in human genetics

positional cloning strategies paralleling the HUMAN GENOME PROJECT
FISH
YACS (yeast artificial chromosome)
BACs (bacterial artificial chromosomes)
PACs (P1 artificial chromosomes)
Pulse field gel electrophoresis
Created new polymorphic markers
Narrowed the genomic interval
Identified cDNAs in region
Sanger sequencing
By 2001, my lab:
Narrowed region to
CHR6:99,575,010- 100,455,412
883kb
Sanger Sequenced all exons in critical region
And promoter regions
Including a zinc finger (later named PRDM13)
expressed in the CNS ...
later found in embryonic retina
Targeted (883 kb)
NEXGEN SEQUENCING
MCDR1 bioinformatics
I was drowning in my own data
bioinformatics quagmire
Generating more and more data
Data analysis : bottleneck
Ed Stone with his team offered me a lifeline
OPENED A CAN OF BIOINFORMATICS "WHOOP-ASS"
"when a normal beating (analysis of the data) just won't do"
Initial publication of mutations
11 families

E-published October 26,2015
Dedicated to the memory of Maurice Rabb, MD (ascertained with KWS the Belize family)
Supported in part by
Stephen A. Wynn Institute for Vision Research
University of Iowa Carver College of Medicine

MCDR1 / mutations
10 MCDR1 families
1 MCDR3 family
SNP or mutation
NONE of the base pair changes found any of the public databases (1000 Genome Project) (dbSNP)
260 controls from Iowa
ONLY found in affected subjects
10 independent families initially
Chr6_100040906 G>T NC
Chr6_100040987 G>C France
Chr6_100041040 C>T Asia
Association between these variants and the disease phenotype is extraordinarily strong
Fisher exact test
All variants are in a DNAS I hypersensitivity binding site
Discovery of NCMD-causing variants in MCDR1
NCMD / MCDR1
all point mutations
in non-coding region
12KB from nearest gene (PRDM13)
In DNASE 1 hypersensitivity binding site (regulatory region)
Featured in OPHTHALMOLOGY lead article
on the cover
editorial
"The editorial staff of Ophthalmology is to be commended for the recognition and acceptance of one of the most important studies in the past several decades."
This report
12 new NCMD / MCDR1 families
To update the data of the initial (11) families
with the new (12) families with MCDR1
35 total families
Do new / additional data continue to support our initial publication results?
MCDR1: KWS: Riverside, CA:
French mutation 100040987
MCDR1:KWS:Riverside, CA:
French mutation 100040987
MCDR1: NY / Mexico / Dr Agemi
N Carolina mutation Chr6:100040906
MCDR1: NY / Mexico / Dr Agemi
N Carolina mutation Chr6:100040906
MCDR1: NY / Mexico / Dr Agemi
N Carolina mutation Chr6:100040906
35 total families available for study
Initial publication 11 families
NEW 12 more independent families

7 with the NC mutations (all USA)
5 with the French mutation (3 European, 2 USA)
6 no mutations found (? Diagnosis of some)
6 in progress
35 NCMD / MCDR1 families total being studied by KWS
DNASE1 hypersensitivity binding site
3 POINT MUTATIONS
ALL IN NON-CODING REGIONS
Why is North Carolina Macular Dystrophy Important?
MCDR1 / NCMD is due to overexpression of PRDM13
Expressed in many lower animals without maculae
New gene / new pathway in
macular disease
Understanding / manipulating
this gene could help us learn
to control / grow new maculae
Summary
Total 23 independent families
with MCDR1 / NCMD phenotype with documented mutations
4 mutations found (3 base pair changes and one duplication of PRDM13)
international collaboration needed to find and prove these mutations
if only one family (i.e NC) ... all changes found would be in linkage disequilibrium
Making it difficult to PROVE mutations
Functional data supports mutations affecting PRDM13 expression
New data continue to support that these DNA changes ARE causative mutations
Subsequently others have confirmed our findings:
MCDR1 / PRDM13 duplication independently confirmed in Texas
by Browne, Sullivan, Daiger et
Subsequently others have confirmed our findings:
MCDR1 / PRDM13 duplication independently confirmed in Texas
by Browne, Sullivan, Daiger et
Lessons learned from NCMD / MCDR1

1) Be a lumper not a splitter
2) Challenge the literature
3) Make friends / collaborations
4) Persevere

Great works are performed
not by strength,
but perseverance.
Samuel Johnson (1709 - 1784)
acknowledgements
NIH / NEI 1989-2004
2004- present Molecular Insight
Research Foundation (501c3)

2004- present:
KWS American Express Plum Card
6/2015 – present:
Stephen A. Wynn Institute for
Vision Research at Univ. of Iowa
(Ed Stone)
North Carolina Macular Dystrophy
treatments
For rare CNVM: anti-VEGF
Because of relative stability and is congenital
... no treatment
Future: CRISPR-CAS9 ... ?
Best Macular Dystrophy
autosomal dominant
mutations in the BEST1 gene
"classic" form symmetric egg-yolk-like lesion centered
20/20 or better in eyes with undisturbed vitelliform lesions
surprising considering the substantial physical separation of the photoreceptor outer segments and the RPE that exists for decades in some individuals
Optical Coherence Tomography (OCT), Fluorescein Angiograms (FA)/Autoflourescence, and Electroretinography
Normal ERG, with abnormal EOG
increased RPE lipofuscin, loss of photoreceptors, sub-RPE drusenoid material, and accumulation of cells and material in the subretinal space
Best Macular Dystrophy
Best Macular Dystrophy
Best Macular Dystrophy
Best Macular Dystrophy
Lump or Split
Best Macular Dystrophy
Treatment for BEST's
primarily of recognizing choroidal neovascularization :
anti-VEGF therapy
elongating the intervals between anti-VEGF injections, discontinuing them altogether,
once the visual acuity is stabilized and all subretinal blood has been resorbed.
AREDS?
FUTURE: CRISPR-CAS9
Stargardts Disease
Most of the differences in clinical findings in patients with ABCA4 disease can be explained by the interplay of three factors that vary among patients:
(1) the severity of their ABCA4 genotype (and hence the rate at which toxic bisretinoids form in the photoreceptors);
(2) the relative sensitivity of the foveal cones to the genotype
(3) the relative sensitivity of the retinal pigment epithelium to the genotype

loss of visual acuity, which can be as mild as 20/30 or as severe as 20/200
abnormal fundus appearance that is incidentally discovered during a routine eye examination
light-colored flecks at the level of the retinal pigment epithelium
The RPE itself responds to ABCA4 mutations quite differently in different patients
Stargardts Disease
Treatment
There is currently no proven treatment for ABCA4 disease
extensive ongoing research in genetics, disease mechanisms, gene therapy, and cell replacement.
drugs that modulate the visual cycle
isoretinoin and fenretinide
slow the formation of these toxic products in Abca4 knockout mice
Autosomal recessive disease more difficult to treat with CRISPR-CAS9
AREDS MINUS beta-carotene
Stargardt's...
Stargardts Disease
Stargardts Disease
Stargardt-like dominant macular dystrophy (SLDMD)
Zhang et al. 5 base pair deletion in the gene ELOVL4 in the affected members of five families
despite 1 of these families being mapped by him to the wrong chromosome
90% of cases in North America
progressive central vision loss
symptoms in the first decade of life
20/200 or worse by 30 years of age
Similarly to the autosomal recessive Stargardt disease
fundus of this disease show pisciform flecks, peripapillary sparing, and macular atrophy
Mutations cause mis-trafficking of the mutant protein in vitro
results in cell death
Stargardt-like dominant macular dystrophy (SLDMD)
61-year-old male
common Leu263 del5tttCTTAA mutation in ELOVL4
20/100
53-year-old male
common Leu263 del5tttCTTAA mutation in ELOVL4
20/100
Pattern Dystrophy
Pattern dystrophy
a group of inherited retinal dystrophies
pigment changes at the level of the RPE
The most common cause: single gene, PRPH2
originally RDS – OMIM #179605
Mutations in this gene also cause some cases of
central areolar choroidal dystrophy,
retinitis pigmentosa
fundus flavimaculatus variant of Stargardt disease

experience macular photostress in their daily life
18% risk of CNVM
normal cone and rod amplitudes and implicit times on the full-field ERG
reduction can be seen when there are more extensive changes
EOG light-peak to dark-trough ratios normal - modestly subnormal
Pattern Dystrophy
SAME MUTATION: MULTIPLE PHENOTYPES
Butterfly-shaped pigment dystrophy
Butterfly-shaped pigmentation
yellow, white, or black and accumulate configuration of three to five "arms" or "wings" that resemble the wings of a butterfly.
pigment deposits that look like drusen or flecks can be seen peripheral to the central lesion
Adult-onset foveomacular vitelliform pattern dystrophy
asymptomatically or with mild blurring
symmetric, solitary, autofluorescent vitelliform lesions in the macula with a central pigmented spot
Sjögren reticular dystrophy of the RPE
clearly defined network of black-pigmented lines
resembles a fishnet with knots or chicken wire
Some patients in the same family as those with reticular dystrophy also have vitelliform and/or butterfly-shaped pigment changes
Central areolar choroidal dystrophy (central areolar retinochoroidal dystrophy)
The earliest change is a fine, mottled depigmentation in the macula of both eyes that appears between the second and fourth decades and gradually evolves into symmetric, sharply outlined, bull's-eye oval or round areas of geographic atrophy of the RPE
Treatment for Pattern Dystrophy
A significant risk for vision loss exists from CNVM
Although CNV less frequent than in AMD
anti-VEGF injections limits vision loss
delayed recovery from exposure to bright light
wearing dark glasses and a hat when outside can allow them to adapt more readily when coming inside
AREDS?
CRISPR-CAS9?
Pattern Dystrophy
Pattern Dystrophy
54-year-old male
    Thr146 ins1aC mutation in PRPH2
    20/40
    nummular atrophy in the macula
spared areas of RPE block the fluorescence from the underlying choroidal vessels
Sorsby Fundus Dystrophy
earliest symptoms NYCTALOPIA
Visible yellow-to-gray material at Bruch's membrane
drusen / pigment
coalesces into a fairly uniform yellowish-gray sheet

more prominent with increasing age
untreated CNV often results in extensive disciform scarring
severely reduces visual acuity
mutations create a new cysteine residue in the mutant protein TIMP3
perturbation of tertiary structure through altered disulfide bonding
TIMP3
component of Bruch's membrane
component of drusen
abnormal deposits in SFD
Treatment aimed at CNV control
laser therapy and PDT are not effective
Sorsby Fundus Dystrophy
36-year-old female
Ser181Cys TIMP-3
20/15
61-year-old female
Trp175Cys TIMP-3
20/125
Autosomal dominant radial drusen
Doyne honeycomb retinal dystrophy
malattia leventinese
Drusen 2nd decade to 7th decade
drusen in the center of the macula and on the nasal edge of the optic disc
tend to be large and round while those at the temporal margin of the macula tend to be smaller, elongated, and radial
drusen abutting the nasal aspect of the nerve
Later may develop central atrophy, scarring, and pigment proliferation that can look similar to SFD
visual acuity is usually excellent until central atrophy
drusen hyperautofluorescent
There is currently no treatment
anti-VEGF therapy
Candidate for CRISPR-CAS9?
Autosomal dominant radial drusen
Doyne honeycomb retinal dystrophy
malattia leventinese
Small 3 exon gene
EFEMP1
ALL affected have the same mutation
Arg345Trp
Most descendants for the
Leventine Valley in Swizerland
Autosomal dominant radial drusen
Doyne honeycomb retinal dystrophy
malattia leventinese

46-year-old female
Arg345Trp mutation in EFEMP1
20/40
large drusen abutting the optic disc
radially oriented drusen
drusen in the central lesion have fused into a honeycomb configuration
57-year-old female
Arg345Trp mutation in EFEMP1
counting fingers
atrophic changes in the central macula
large drusen are present nasal to the nerve head

Enhanced S Cone Syndrome
Goldmann-Favre
rare non syndromic autosomal recessive
absence of rod function
large-amplitude S cone-mediated electrical responses
psychophysical hypersensitivity to blue light
a cone dominant retina
ERG's: greater amplitudes to short-wavelength
gain in function of photoreceptors
NR2E3 mutations for ESCS
retinal nuclear receptor, a ligand dependent transcription factor
expression limited to the outer nuclear layer of the human retina
ESCS
RESULTS
Case 1:23 year old female
THE ORIGINAL CASE DEFINING ESCS
phenotype PREVIOUSLY reported in detail
19 year old female
unusual ERG responses led to the recognition of the ESCS syndrome
Night blind all of her life
otherwise asymptomatic
20/25 RT, 20/20 LT
fields and color vision are normal
exam shows no abnormalities other than some very sparse yellowish spots in the arcade regions
ESCS #1
ESCS case 1
Glomerulonephritis Type II and Drusen
The majority of patients with membranproliferative glomerulonephritis (MPGN) type II
basal laminar drusen
visual acuity tends to be preserved unless CNV
exudative drusen
serous detachment complicate the disease

abnormal EOG with a relatively normal ERG in some patients
suggesting a more global retinal dysfunction than the visible drusen would suggest
morphologically and compositionally similar to the drusen found in AMD
Glomerulonephritis Type II and Drusen
In Vivo CRISPR/Cas9 Gene Editing Corrects Retinal Dystrophy in the
S334ter-3 Rat Model of Autosomal Dominant Retinitis Pigmentosa
CRISPR/Cas9 Gene Editing in RP
GENE EDITING
CRISPR/Cas9 Gene Editing in Dmd
The Carver Genetic Testing Laboratory
The Carver Genetic Testing Laboratory
The Carver Genetic Testing Laboratory
TAKE HOME POINTS
LUMP
FULL DISCERNMENT ONLY WITH USING FAMILY DATA / MEMBERS / MOLECULAR GENETICS
FAMILY CONTEXT
ALL DRUSEN ARE CLINICALLY ALIKE
NORTH CAROLINA MACULAR DYSTROPHY / MCDR1 mutations affect PRDM13 expression
PERSEVERANCE
50 meter butterfly 30.02 sec

Macular Dystrophies with DRUSEN

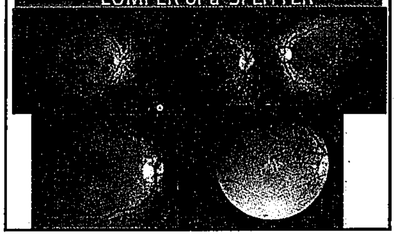
Kent W. Small

MACULA & RETINA INSTITUTE
KENT W. SMALL, MD
Innovative, experienced and highly personalized retinal care

Kent W. Small, M.D.

SOLO PRIVATE PRACTICE
Molecular Insight Research Foundation
Cedar-Sinai Regenerative Medicine Institute
Los Angeles, CA
Glendale, CA

Are you a LUMPER or a SPLITTER



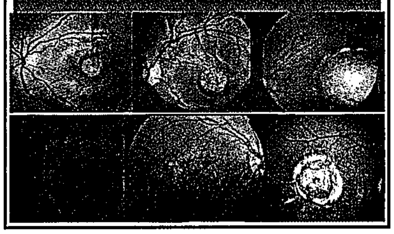
Kent W. Small, M.D. training and experience

* Residency: Duke University
* Fellowship, Retina: Duke University
* Fellowship, Molecular Genetics: Duke
* Director: Retina, Univ of Florida
* Professor: Retina, UCLA, Jules Stein
* Chairman: Univ. Texas
* Founder / President: Kent W. Small, M.D
* Founder / President: Macula & Retina Institute
* Founder / President: Molecular Insight Research Foundation
* Scientist: Regenerative Medicine Institute, Cedars-Sinai
* 310-659-2200 818-552-5040

KENT W SMALL MD

* NO FINANCIAL DISCLOSURES
* NO CONFLICTS OF INTEREST
* (MAY BE THE ONLY TALK HERE WITH NO FINANCIAL INTERESTS / DISCLOSURE)

Are you a LUMPER or a SPLITTER



Molecular Insight Research Foundation

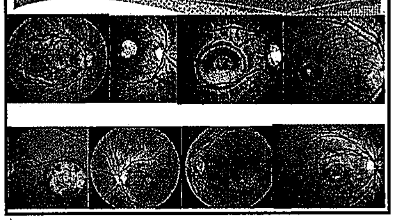
* Kent W Small MD
* 30 years of research and clinical experience
  * 1st to use microsatellites DNA repeats in gene mapping in ophthalmology
  * 1st to map a macular degeneration in the Human Genome Project
  * Found mutations in
    * North Carolina macular dystrophy (MCDR1)
    * cone-rod dystrophy (CORD5)
    * enhanced S-cone (Goldmann-Favre)
    * Reis-Buchler corneal dystrophy
    * Blepharophimosis syndrome (BPES)
    * Reigers Syndrome
  * Over 200 peer reviewed scientific publications
  * 20 years of NIH support
  * 15 years of Foundation Fighting Blindness support
  * CIRM (California Institute for Regenerative Medicine)
  * Trained over 100 ophthalmologists / optometrists

310-659-2200 818-552-5040

MACULAR DYSTROPHIES with DRUSEN

* A FEW CONCEPTS
  * LUMP vs SPLIT
    * Tremendous variable expression within single families shows the value of LUMPING
  * FAMILY CONTEXT
  * ALL DRUSEN ARE ALIKE
    * Splitting drusen into subcategories is worthless as are most genotype / phenotype studies
  * NORTH CAROLINA MACULAR DYSTROPHY / PRDM13

Lump or Split

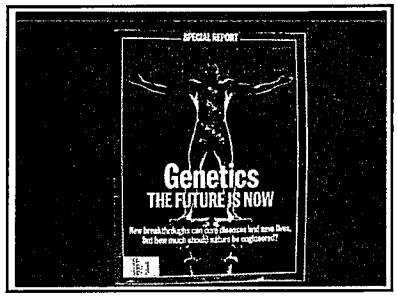


Types of Macular Dystrophies with drusen

DISEASE NAME	GENE	CHROMOSOME	INHERITANCE
BEST5	BEST1	11	AD
STARGARDT	ABCA4	1	AR
STARGARDT-LIKE MACULAR DYSTROPHY	ELOVL4	6	AD
PATTERN DYSTROPHY	PRPH2	6	AD
SORSBY FUNDUS DYSTROPHY	TIMP3	22	AD
DOMINANT DRUSEN	EFEMP1	2	AD
NORTH CAROLINA MACULAR DYSTROPHY	PRDM13	6	AD
ENHANCED S-CONE (GOLDMANN-FAVRE)	NR2E3	15	AR
Glomerulonephritis Type			

Drug Toxicity which can mimic macular / retinal degenerations

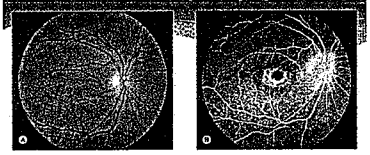
Agent	Reference(s)
Chloroquine	Hobbs et al., 1959; Marmor et al., 2011
Hydroxychloroquine	Shearer et al., 1965; Marmor et al., 2011;
Thioridazine (mellaril)	Weekley et al., 1960
Chlorpromazine (thorazine)	Delong et al., 1965
Clofazimine	Craythorn et al., 1986
Tamoxifen	Kaiser-Kupfer and Lippman, 1978
Oxalosis/methoxyfluorane	Bullock and Albert, 1975; Albert et al., 1975
Canthoxanthine	Boudreault et al., 1983; Ros et al., 1985
Nitrofurantoin	Ibanez et al., 1994
Talc	ALLee, 1972
Deferoxamine	Halmovici et al., 2002; Gonzales et al., 2004



Initial approach to a patient with macular dystrophy

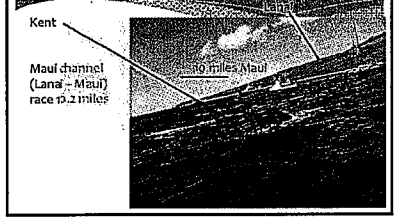
- bilateral and are usually extremely symmetrical
- Most autosomal dominant
- one or more living affected relatives
- careful family history
- Most over the age of 50 misdiagnosed as AMD
- Careful medication history past and present / Drug toxicity
- autosomal dominant macular dystrophies have visual acuities that are better than one might expect
- autosomal recessive (Stargardt disease, ABCA4) acuities that are poorer than one might expect given the relatively mild abnormality of their fundus

Macular Dystrophy?



50-year-old female , visual acuity was 20/30
plaquenil toxicity
bull's-eye fundus appearance
easily be mistaken for Stargardt disease without flecks

"If your ship does not come in... swim out to it..." Jonathan Winters

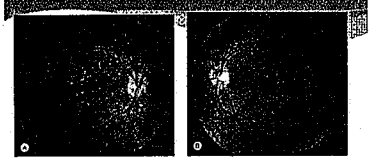


Macular Dystrophies

*THE CONTEXT OF THE FAMILY IS IMPORTANT

* SOMETIMES CRITICAL
* Only when other family members are examined can you appreciate the full spectrum of the disease and make the correct diagnosis

Macular dystrophy?



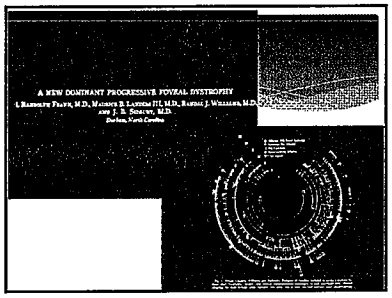
A 33-year-old female with 20/20 visual acuity
asymmetry is more suggestive of inflammatory disease
multifocal choroiditis

"Treasure Your Exceptions"

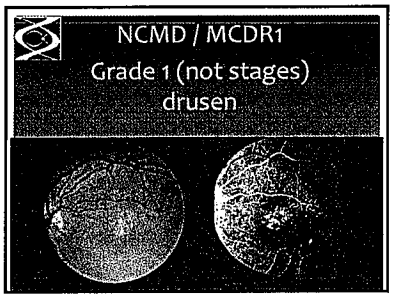
William Bateson
8 August 1861 – 8 February 1926

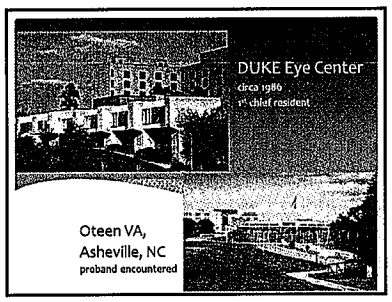
- English biologist
- first person to use the term genetics to describe the study of heredity
- the chief populariser of the ideas of Gregor Mendel

North Carolina Macular Dystrophy
MCDR1 / NCMD
my 1st encounter
1987 at the Asheville VA hospital
DUKE rotation
20/50
Hallmark:
MARKED VARIABLE EXPRESSIVITY
bad macular lesion
good vision
1/3 of affected family
remainder asymptomatic
Confused with TOXO

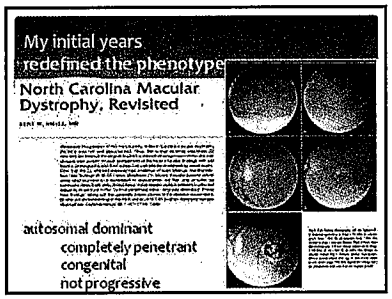


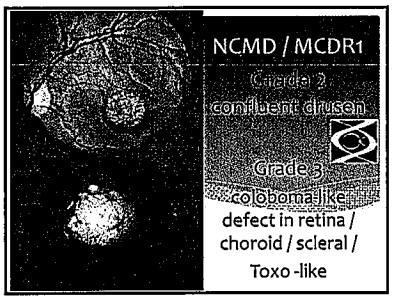
NCMD / MCDR1
Grade 1 (not stages)
drusen



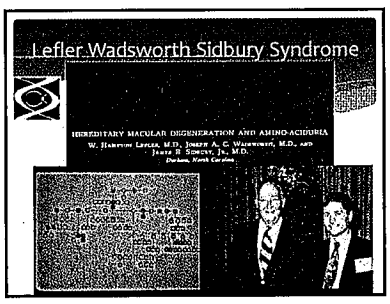


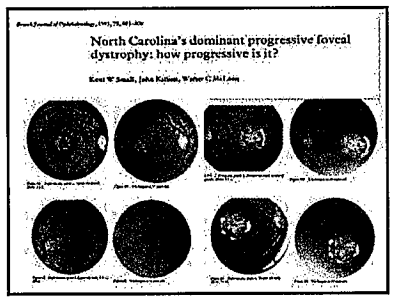
My initial years
redefined the phenotype
North Carolina Macular Dystrophy, Revisited
autosomal dominant
completely penetrant
congenital
not progressive



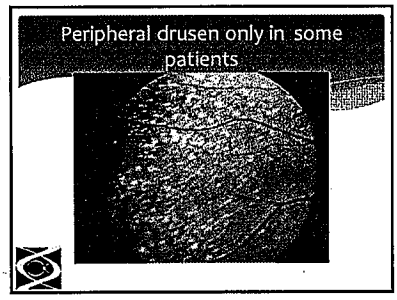


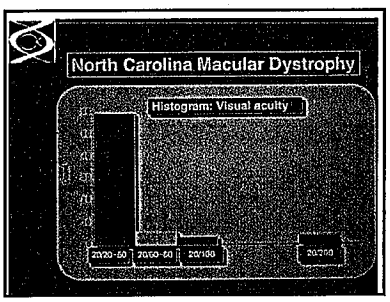
Lefler Wadsworth Sidbury Syndrome

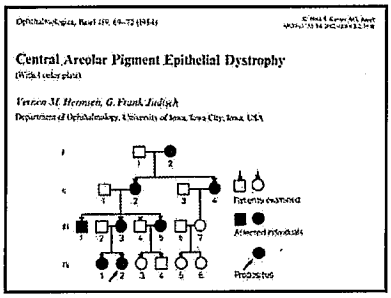


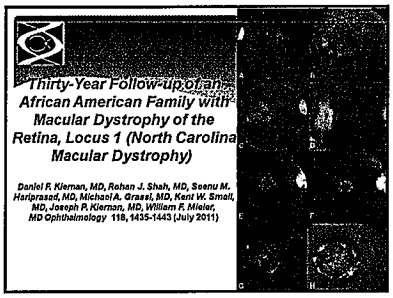


Peripheral drusen only in some patients

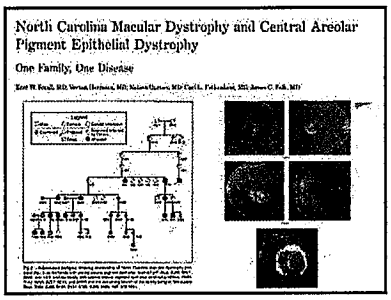


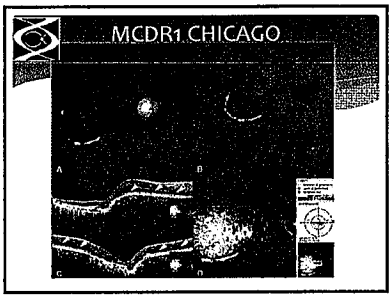

North Carolina Macular Dystrophy
Histogram: Visual acuity
20/20-50
20/60-80
20/100
20/200

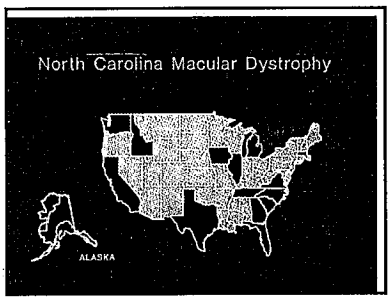

Central Areolar Pigment Epithelial Dystrophy
Affected individuals
Probands

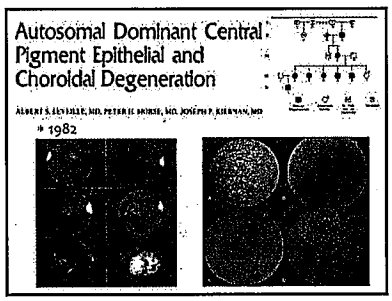

Thirty-Year Follow-up of an African American Family with Macular Dystrophy of the Retina, Locus 1 (North Carolina Macular Dystrophy)
Daniel F. Kiernan, MD, Rohan J. Shah, MD, Soonu M. Hariprasad, MD, Michael A. Grassi, MD, Kent W. Small, MD, Joseph P. Kiernan, MD, William F. Mieler, MD Ophthalmology 118, 1435-1443 (July 2011)

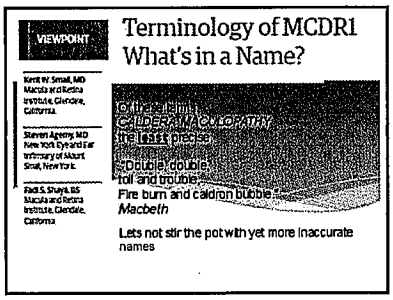

ARVO / Sarasota 1989


North Carolina Macular Dystrophy and Central Areolar Pigment Epithelial Dystrophy
One Family, One Disease


MCDR1 CHICAGO


North Carolina Macular Dystrophy
ALASKA


Autosomal Dominant Central Pigment Epithelial and Choroidal Degeneration
1982


VIEWPOINT
Terminology of MCDR1
What's in a Name?
Fire burn and caldron bubble
Macbeth
Lets not stir the pot with yet more inaccurate names

The MANY NAMES of MCDR1

North Carolina Macular Dystrophy

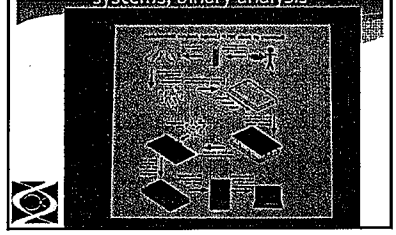
TOO MUCH SPLITTING!

- A SINGLE DISEASE IN A SINGLE FAMILY-GIVEN ALL OF THESE DISEASE ENTITIES AND NAMES
- Lefler Wadsworth Sidbury "Syndrome"
- dominant macular degeneration and aminoaciduria
- dominant progressive foveal dystrophy
- central areolar pigment epithelial dystrophy
- central pigment epithelial and choroidal degeneration
- Caldera maculopathy
- North Carolina Macular Dystrophy
- SEEMS LIKE IT IS HUMAN NATURE TO WANT TO SPLIT

what happens to splitters and Confederate spies

Extensive genealogical research looking for additional affected individuals

RFLPs labor intensive, slow, bi-allelic systems, binary analysis



VIEWPOINT

Terminology of MCDR1 What's in a Name?

Kent W. Small, MD · Macula and Retina Institute, Glendale, California.

Steven Agemy, MD · New York Eye and Ear Infirmary of Mount Sinai, New York.

Paul S. Sinyk, BS · Macula and Retina Institute, Glendale, California.

North Carolina Macular Dystrophy

Is a bad name

Is a misnomer

Given by Don Gass in his atlas - and it stuck

Named for its FOUNDER EFFECT

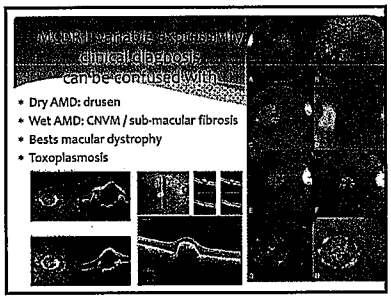
large single family in a stable Southern society, very little inbreeding

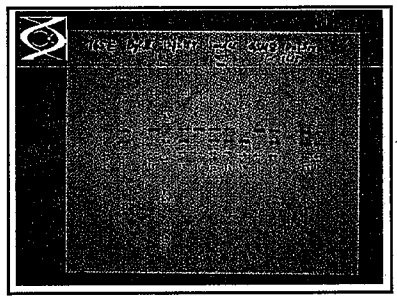
found worldwide

Caucasian, Asian, Hispanic, Indigenous, African-American

MCDR1 variable clinical phenotype can be confused with

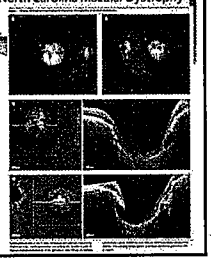
- Dry AMD: drusen
- Wet AMD: CNVM / sub-macular fibrosis
- Bests macular dystrophy
- Toxoplasmosis

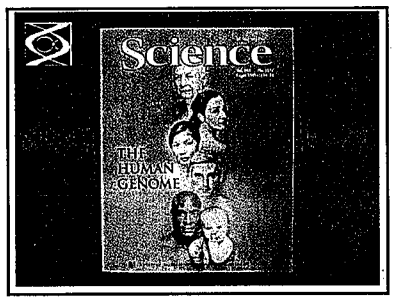




Intrachoroidal Cavitation in North Carolina Macular Dystrophy

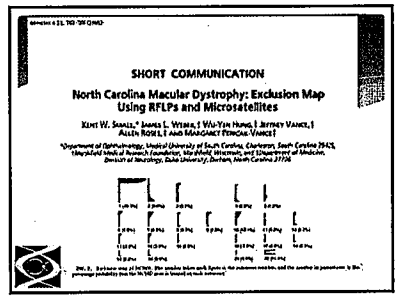
- MCDR1 in Nashville, TN
- Cavitation not ideal descriptor
- implies tissue was present which later disappeared (sink hole)
- This is a developmental abnormality... tissue never did develop / form

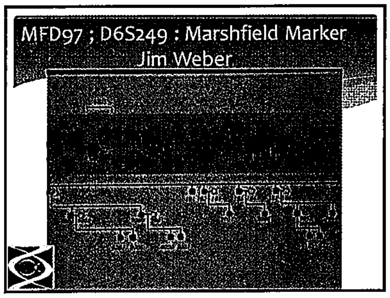


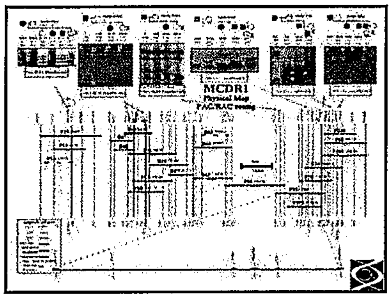


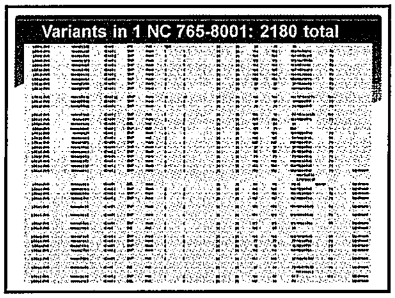
SHORT COMMUNICATION

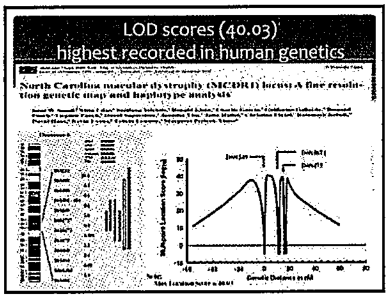
North Carolina Macular Dystrophy: Exclusion Map Using RFLPs and Microsatellites

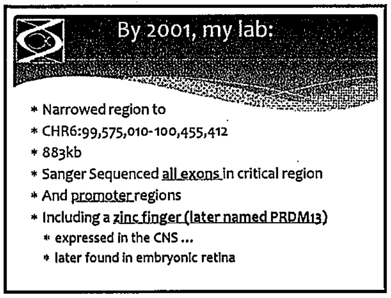


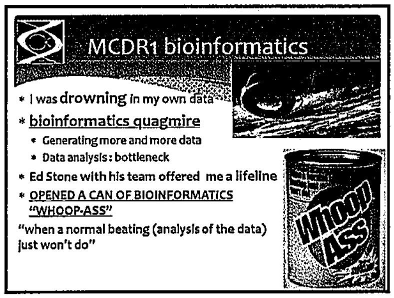

MFD97 ; D6S249 : Marshfield Marker
Jim Weber

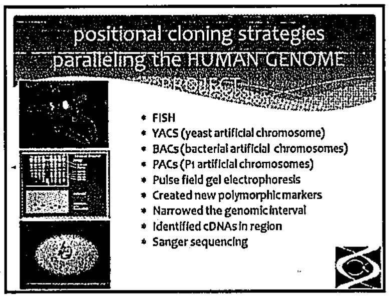

MCDR1
Physical Map

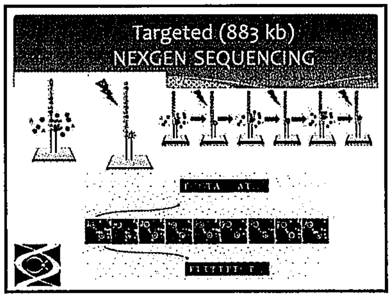

Variants in 1 NC 765-8001: 2180 total

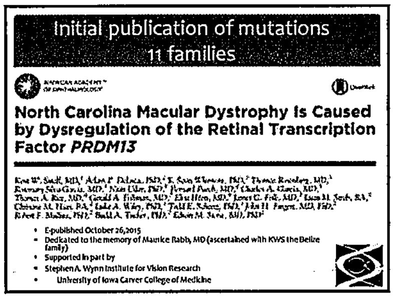

LOD scores (40.03)
highest recorded in human genetics
North Carolina macular dystrophy (MCDR1) locus: A fine resolution genetic map and haplotype analysis


By 2001, my lab:
Narrowed region to
CHR6:99,575,010-100,455,412
883kb
Sanger Sequenced all exons in critical region
And promoter regions
Including a zinc finger (later named PRDM13)
expressed in the CNS …
later found in embryonic retina


MCDR1 bioinformatics
I was drowning in my own data
bioinformatics quagmire
Generating more and more data
Data analysis: bottleneck
Ed Stone with his team offered me a lifeline
OPENED A CAN OF BIOINFORMATICS "WHOOP-ASS"
"when a normal beating (analysis of the data) just won't do"
Whoop Ass


positional cloning strategies paralleling the HUMAN GENOME PROJECT
FISH
YACS (yeast artificial chromosome)
BACs (bacterial artificial chromosomes)
PACs (P1 artificial chromosomes)
Pulse field gel electrophoresis
Created new polymorphic markers
Narrowed the genomic interval
Identified cDNAs in region
Sanger sequencing


Targeted (883 kb)
NEXGEN SEQUENCING


Initial publication of mutations
11 families
North Carolina Macular Dystrophy Is Caused by Dysregulation of the Retinal Transcription Factor PRDM13
Epublished October 26,2015
Dedicated to the memory of Maurice Rabb, MD (ascertained with KWS the Belize family)
Supported in part by
Stephen A. Wynn Institute for Vision Research
University of Iowa Carver College of Medicine

MCDR1 / mutations
10 MCDR1 families
1 MCDR3 family

Table 1

Family	Previously Published Family ID (reference)	Haplotype	Variant #	HG19 Chromosomal Location	Nucleotide Change	Affected	Unaffected	Total Samples
A	786	North Carolina	V1	Chr6_100040906	G>T	61	22	73
B	703	North Carolina	V1	Chr6_100040906	G>T	4	5	9
C	764	North Carolina	V1	Chr6_100040906	G>T	1	1	2
D	772	North Carolina	V1	Chr6_100040906	G>T	4	0	4
E	1193	North Carolina	V1	Chr6_100040906	G>T	3	0	3
F	1292	North Carolina	V1	Chr6_100040906	G>T	2	1	3
G	780	French	V2	Chr6_100040987	G>C	1	0	1
H	718	French / German	V2	Chr6_100040987	G>C	1	0	1
I	709	Asian	V3	Chr6_100041040	C>T	2	0	2
J	1463	Belize	V4	Chr6_100020306-100143306	123101bp Tandem Duplication	11	4	15
K	MCDR3 (Rosenberg, et al)	Danish	V5	Chr5_3647801-4499077	851356bp Tandem Duplication	11	1	12
						81	34	115

NCMD / MCDR1 all point mutations

* in non-coding region
* 12KB from nearest gene (PRDM13)
* In DNASE 1 hypersensitivity binding site (regulatory region)

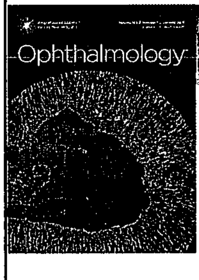
This report 12 new NCMD / MCDR1 families

* To update the data of the initial (11) families
* with the new (12) families with MCDR1
* 35 total families
* Do new / additional data continue to support our initial publication results?

SNP or mutation

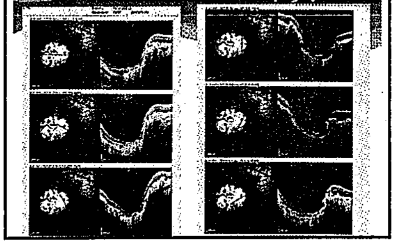
* NONE of the base pair changes found any of the public databases (1000 Genome Project) (dbSNP)
* 260 controls from Iowa
* ONLY found in affected subjects

10 independent families initially
* Chr6_100040906 G>T NC
* Chr6_100040987 G>C France
* Chr6_100041040 C>T Asia
* Association between these variants and the disease phenotype is extraordinarily strong $P < 10^{-291}$
* Fisher exact test
* All variants are in a DNAS I hypersensitivity binding site



Featured in OPHTHALMOLOGY lead article on the cover editorial

MCDR1: KWS: Riverside, CA: French mutation 100040987 OD



Discovery of NCMD-causing variants in MCDR1

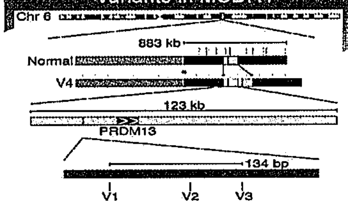


Figure 2

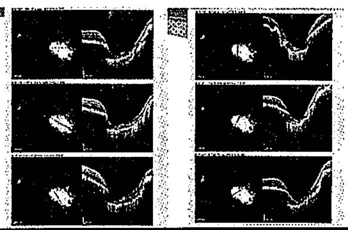
Editorial

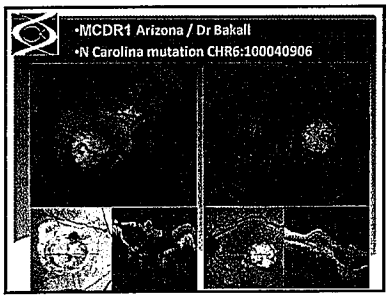
Dysregulation of Retinal Transcription Factor PRDM13 and North Carolina Macular Dystrophy
[illegible]

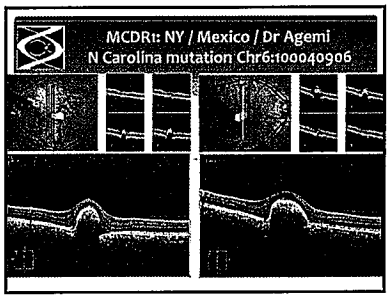
* "The editorial staff of Ophthalmology is to be commended for the recognition and acceptance of

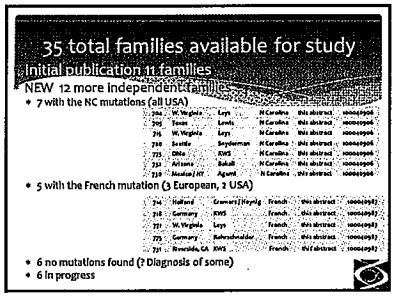
one of the most important studies in the past several decades."

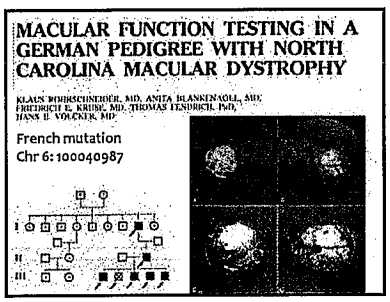
MCDR1:KWS:Riverside, CA: French mutation 100040987 OS

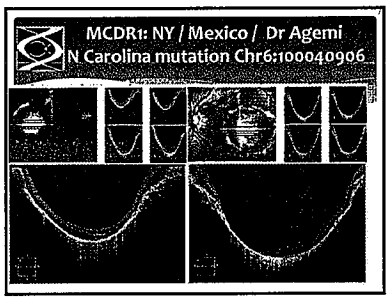


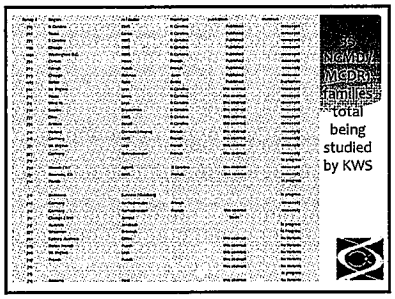

•MCDR1 Arizona / Dr Bakall
•N Carolina mutation CHR6:100040906

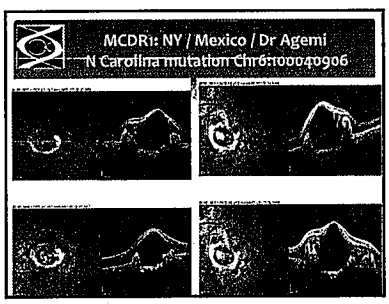

MCDR1: NY / Mexico / Dr Agemi
N Carolina mutation Chr6:100040906

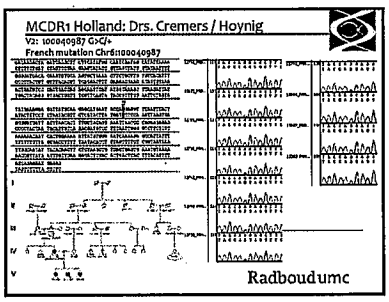

35 total families available for study
Initial publication 11 families
NEW 12 more independent families
• 7 with the NC mutations (all USA)
• 5 with the French mutation (3 European, 2 USA)
• 6 no mutations found (? Diagnosis of some)
• 6 in progress

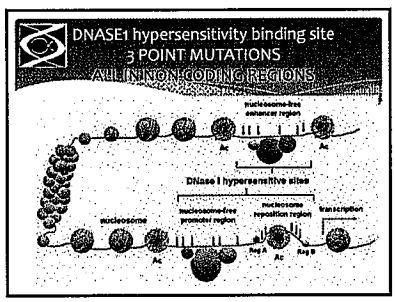

MACULAR FUNCTION TESTING IN A GERMAN PEDIGREE WITH NORTH CAROLINA MACULAR DYSTROPHY
French mutation
Chr 6: 100040987


MCDR1: NY / Mexico / Dr Agemi
N Carolina mutation Chr6:100040906


35 NCMD/MCDR1 families total being studied by KWS

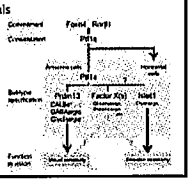

MCDR1: NY / Mexico / Dr Agemi
N Carolina mutation Chr6:100040906


MCDR1 Holland: Drs. Cremers / Hoynig
Radboudumc


DNASE1 hypersensitivity binding site
3 POINT MUTATIONS
ALL IN NON-CODING REGIONS
Dnase I hypersensitive sites
nucleosome
nucleosome-free promoter region
nucleosome repositon region
transcription

Why is North Carolina Macular Dystrophy Important?

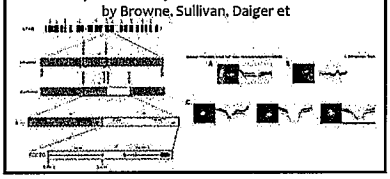
- MCDR1 / NCMD is due to overexpression of PRDM13
- Expressed in many lower animals without maculae
- New gene / new pathway in macular disease
- Understanding / manipulating this gene could help us learn to control / grow new maculae



Subsequently others have confirmed our findings:

MCDR1 / PRDM13 duplication independently confirmed in Texas

by Browne, Sullivan, Daiger et



North Carolina Macular Dystrophy treatments

- For rare CNVM: anti-VEGF
- Because of relative stability and is congenital
- ... no treatment
- Future: CRISPR-CAS9 ... ?

Summary

***Total 23 independent families**

- with MCDR1 / NCMD phenotype with documented mutations
- 4 mutations found (3 base pair changes and one duplication of PRDM13)
- international collaboration needed to find and prove these mutations
- If only one family (i.e NC) ... all changes found would be in linkage disequilibrium
- Making it difficult to PROVE mutations
- Functional data supports mutations affecting PRDM13 expression

*** New data continue to support that these DNA changes ARE causative mutations**

Lessons learned from NCMD / MCDR1

1) Be a lumper not a splitter
2) Challenge the literature
3) Make friends / collaborations
4) Persevere

Great works are performed not by strength, but perseverance.

Samuel Johnson (1709 - 1784)

Johnson (1775) showing his intense concentration and the weakness of his eyes; he did not want to be depicted as "Blinking Sam"

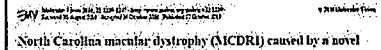
Best Macular Dystrophy

- autosomal dominant
- mutations in the BEST1 gene
- "classic" form symmetric egg-yolk-like lesion centered
- 20/20 or better in eyes with undisturbed vitelliform lesions
- surprising considering the substantial physical separation of the photoreceptor outer segments and the RPE that exists for decades in some individuals
- Optical Coherence Tomography (OCT), Fluorescein Angiograms (FA)/Autofluorescence, and Electroretinography
- Normal ERG, with abnormal EOG
- Increased RPE lipofuscin, loss of photoreceptors, sub-RPE drusenoid material, and accumulation of cells and material in the subretinal space

Subsequently others have confirmed our findings:

MCDR1 / PRDM13 duplication independently confirmed in Texas

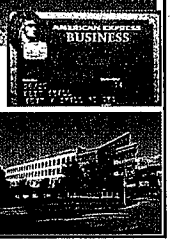
by Browne, Sullivan, Daiger et



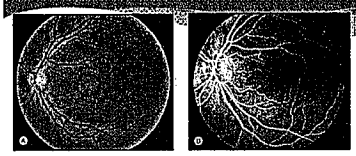
North Carolina macular dystrophy (MCDR1) caused by a novel tandem duplication of the *PRDM13* gene

acknowledgements

- NIH / NEI 1989-2004
- 2004- present Molecular Insight Research Foundation (501c3)
- 2004- present: KWS American Express Plum Card
- 6/2015 – present:
- Stephen A. Wynn Institute for Vision Research at Univ. of Iowa (Ed Stone)

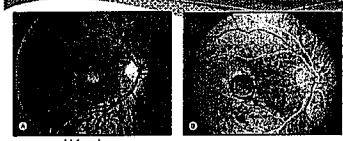


Best Macular Dystrophy



51-year-old male with a 3 nucleotide deletion in BEST1 (Leu294 del3cTCA). 20/30 in this eye
classic vitelliform lesion remains completely homogeneous after more than five decades of life.
almost complete masking of the normal choroidal circulation underlying the lesion.

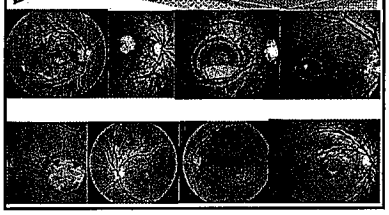
Best Macular Dystrophy



23-year-old female
evolving subretinal fibrotic nodule.
subretinal blood can be seen at the inferior margin
20/100.
classic choroidal neovascular membrane

Lump or Split

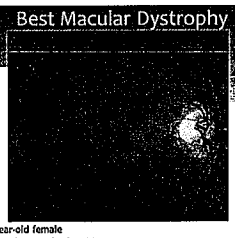
Best Macular Dystrophy



Stargardts Disease

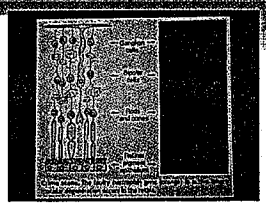
- Most of the differences in clinical phenotype in patients with ABCA4 disease can be explained by the interplay of three factors that vary among patients:
  - (1) the severity of their ABCA4 genotype (and hence the rate at which toxic bisretinoids form in the photoreceptors);
  - (2) the relative sensitivity of the foveal cones to the genotype
  - (3) the relative sensitivity of the retinal pigment epithelium to the genotype
- loss of visual acuity, which can be as mild as 20/30 or as severe as 20/200
- abnormal fundus appearance that is incidentally discovered during a routine eye examination
- light-colored flecks at the level of the retinal pigment epithelium
- The RPE itself responds to ABCA4 mutations quite differently in different patients

Best Macular Dystrophy



72-year-old female
Thr307Ile mutation in BEST1
20/80
circular zone of geographic atrophy centered on fixation
flanked superiorly by a few drusen-like deposits

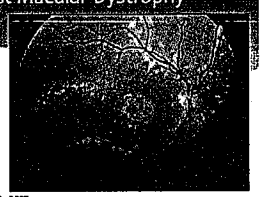
Best Macular Dystrophy



Stargardts Disease Treatment

- There is currently no proven treatment for ABCA4 disease
- extensive ongoing research in genetics, disease mechanisms, gene therapy, and cell replacement.
- drugs that modulate the visual cycle
  - Isoretinoin and fenretinide
  - slow the formation of these toxic products in Abca4 knockout mice
- Autosomal recessive disease more difficult to treat with CRISPR-CAS9
- AREDS MINUS beta-carotene

Best Macular Dystrophy

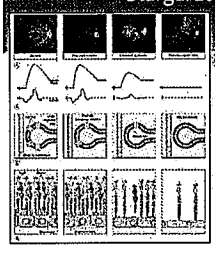


42-year-old male
Tyr227Asn mutation in BEST1
20/100
two large areas of subretinal fluid
subretinal deposits of lipofuscin along the edges of both lesions.

Treatment for BEST's

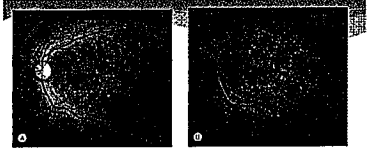
- primarily of recognizing choroidal neovascularization
- anti-VEGF therapy
- elongating the intervals between anti-VEGF injections, discontinuing them altogether,
- once the visual acuity is stabilized and all subretinal blood has been resorbed.
- AREDS?
- FUTURE: CRISPR-CAS9

Stargardt's...



- effects of reduced ABCA4 function on full-field electroretinograms.
- reduced ABCA4 function on the accumulation of bisretinoid (yellow symbols) on the inner leaflet of the photoreceptor outer-segment disc membranes.
- histopathological effects of reduced ABCA4 activity. In patients with Stargardt disease, the rate of bisretinoid formation in the outer segments is relatively slow and the photoreceptors are not directly injured.

Stargardts Disease

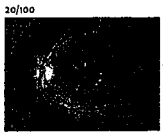


25-year-old female with compound heterozygous mutations in ABCA4 (Thr1019Met/Lys583Asn)
20/80 despite extensive injury to the RPE
relative preservation of the foveal and peripapillary retina and RPE.

Stargardt-like dominant macular dystrophy (SLDMD)

61-year-old male
common Leu263 delsttCTTAA mutation in ELOVL4
20/100

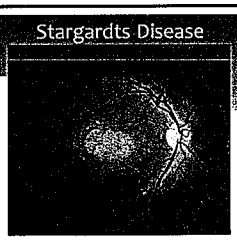
53?-year-old female
common Leu263 delsttCTTAA mutation in ELOVL4
20/100



Treatment for Pattern Dystrophy

- A significant risk for vision loss exists from CNVM
- Although CNV less frequent than in AMD
- anti-VEGF injections limits vision loss
- delayed recovery from exposure to bright light
- wearing dark glasses and a hat when outside can allow them to adapt more readily when coming inside
- AREDS?
- CRISPR-CAS9?

Stargardts Disease

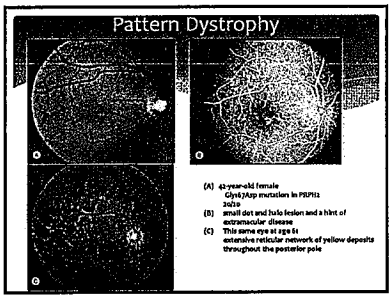


36-year-old female
compound heterozygous mutations in ABCA4 (Phe608Ile/Gly1961Glu)
20/100
pisciform flecks ring the central lesion

Pattern Dystrophy

- Pattern dystrophy
- a group of inherited retinal dystrophies
- pigment changes at the level of the RPE
- The most common cause: single gene, PRPH2
- originally RDS – OMIM #179605
- Mutations in this gene also cause some cases of
  - central areolar choroidal dystrophy,
  - retinitis pigmentosa
  - fundus flavimaculatus variant of Stargardt disease
- experience macular photostress in their daily life
- 18% risk of CNVM
- normal cone and rod amplitudes and implicit times on the full-field ERG
- reduction can be seen when there are more extensive changes
- EOG light-peak to dark-trough ratios normal - modestly subnormal

Pattern Dystrophy



(A) 42-year-old female
Gly167Asp mutation in PRPH2
20/20
(B) small dot and halo lesion and a hint of extramacular disease
(C) This same eye at age 61
extensive reticular network of yellow deposits throughout the posterior pole

Stargardt-like dominant macular dystrophy (SLDMD)

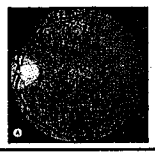
- Zhang et al. 5 base pair deletion in the gene ELOVL4 in the affected members of five families
- despite 1 of these families being mapped by him to the wrong chromosome
- 90% of cases in North America
- progressive central vision loss
- symptoms in the first decade of life
- 20/200 or worse by 30 years of age
- Similarly to the autosomal recessive Stargardt disease
  - fundus of this disease show pisciform flecks, peripapillary sparing, and macular atrophy
- Mutations cause mis-trafficking of the mutant protein in vitro
- results in cell death

Pattern Dystrophy

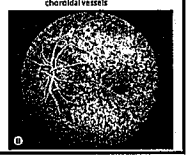
- SAME MUTATION: MULTIPLE PHENOTYPES
- Butterfly-shaped pigment dystrophy
  - Butterfly-shaped pigmentation
  - yellow, white, or black and accumulate configuration of three to five "arms" or "wings" that resemble the wings of a butterfly.
  - pigment deposits that look like dozen or flecks can be seen peripheral to the central lesion
- Adult-onset foveomacular vitelliform pattern dystrophy
  - asymptomatically or with mild blurring
  - symmetric, solitary, autofluorescent vitelliform lesions in the macula with a central pigmented spot
- Sjögren reticular dystrophy of the RPE
  - clearly defined network of black pigmented lines
  - resembles a fishnet with knots or chicken wire
  - Some patients in the same family as those with reticular dystrophy also have vitelliform and/or butterfly-shaped pigment changes
- Central areolar choroidal dystrophy (central areolar retinochoroidal dystrophy)
  - The earliest change is a fine, mottled depigmentation in the macula of both eyes that appears between the second and fourth decades and gradually evolves into a symmetric, sharply outlined, bull's-eye oval or round area of geographic atrophy of the RPE

Pattern Dystrophy

54-year-old male
Thr46InsaC mutation in PRPH2
20/40
nummular atrophy in the macula



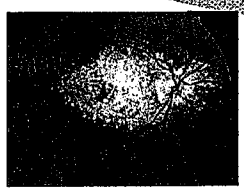
spared areas of RPE block the fluorescence from the underlying choroidal vessels



Sorsby Fundus Dystrophy

- Visible yellow-to-gray material (drusen)
- drusen / pigment
- coalesces into a fairly uniform yellowish-gray sheet
- more prominent with increasing age
- untreated CNV often results in extensive disciform scarring
- severely reduces visual acuity
- mutations create a new cysteine residue in the mutant protein TIMP3
- perturbation of tertiary structure through altered disulfide bonding
- TIMP3
- component of Bruch's membrane
- component of drusen
- abnormal deposits in SFD
- Treatment aimed at CNV control
- laser therapy and PDT are not effective

Autosomal dominant radial drusen Doyne honeycomb retinal dystrophy malattia leventinese



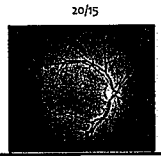
- Small 3 exon gene
- EFEMP1
- ALL affected have the same mutation
- Arg345Trp
- Most descendants for the
- Leventine Valley in Switzerland

ESCS

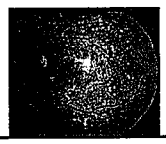
- RESULTS
- Case 1: 23 year old female
- THE ORIGINAL CASE DEFINING ESCS
- phenotype PREVIOUSLY reported in detail
- 19 year old female
- unusual ERG responses led to the recognition of the ESCS syndrome
- Night blind all of her life
- otherwise asymptomatic
- 20/25 RT, 20/20 LT
- fields and color vision are normal
- exam shows no abnormalities other than some very sparse yellowish spots in the arcade regions

Sorsby Fundus Dystrophy

36-year-old female
Ser181Cys TIMP-3
20/15



61-year-old female
Trp175Cys TIMP-3
20/125

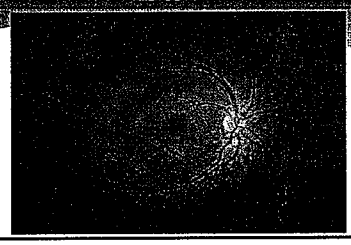


Autosomal dominant radial drusen Doyne honeycomb retinal dystrophy malattia leventinese

46-year-old female
Arg345Trp mutation in *EFEMP1*
20/40
large drusen abutting the optic disc
radially oriented drusen
drusen in the central fovea have fused into a honeycomb configuration

37-year-old female
Arg345Trp mutation in EFEMP1
counting fingers
atrophic changes in the central macula
large drusen are present nasal to the nerve head

ESCS #1



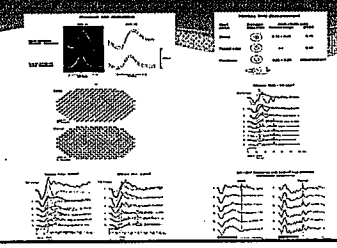
Autosomal dominant radial drusen Doyne honeycomb retinal dystrophy malattia leventinese

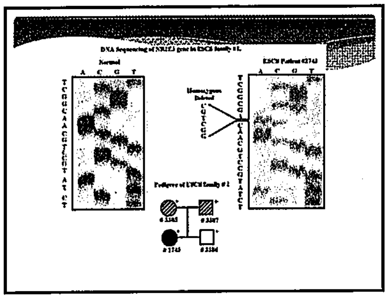
- Drusen 2nd decade to 7th decade
- drusen in the center of the macula and on the nasal edge of the optic disc
- tend to be large and round while those at the temporal margin of the macula tend to be smaller, elongated, and radial
- drusen abutting the nasal aspect of the nerve
- Later may develop central atrophy, scarring, and pigment proliferation that can look similar to SFD
- visual acuity is usually excellent until central atrophy
- drusen hyperautofluorescent
- There is currently no treatment
- anti-VEGF therapy
- Candidate for CRISPR-CAS9?

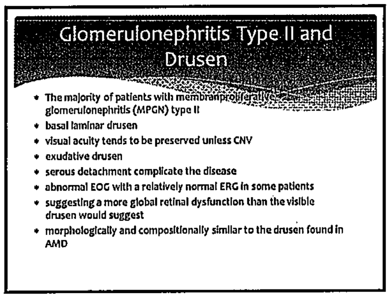
Enhanced S Cone Syndrome Goldmann-Favre

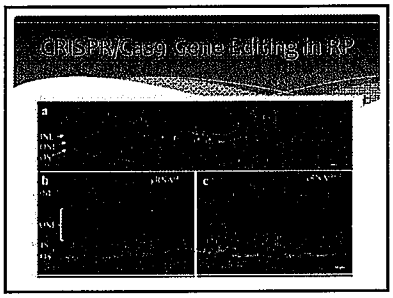
* rare non syndromic autosomal recessive
* absence of rod function
* large-amplitude S cone-mediated electrical responses
* psychophysical hypersensitivity to blue light
* a cone dominant retina
* ERG's: greater amplitudes to short-wavelength
* gain in function of photoreceptors
* NR2E3 mutations for ESCS
* retinal nuclear receptor, a ligand dependent transcription factor
* expression limited to the outer nuclear layer of the human retina

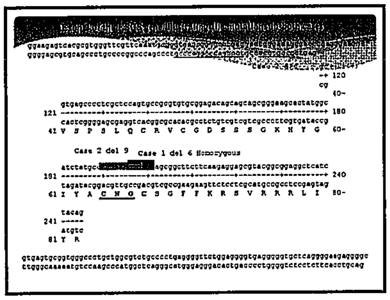
ESCS case 1

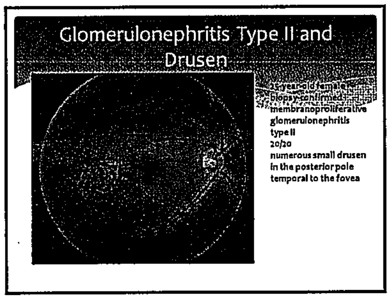


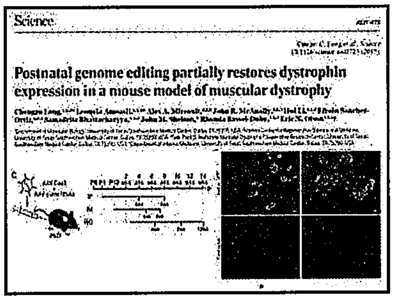


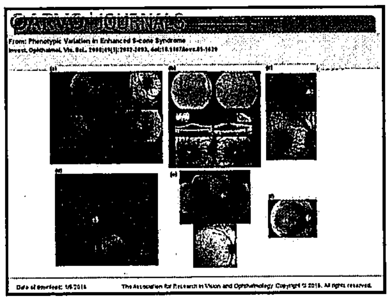

Glomerulonephritis Type II and Drusen
The majority of patients with membranoproliferative glomerulonephritis (MPGN) type II
basal laminar drusen
visual acuity tends to be preserved unless CNV
exudative drusen
serous detachment complicate the disease
abnormal EOG with a relatively normal ERG in some patients
suggesting a more global retinal dysfunction than the visible drusen would suggest
morphologically and compositionally similar to the drusen found in AMD

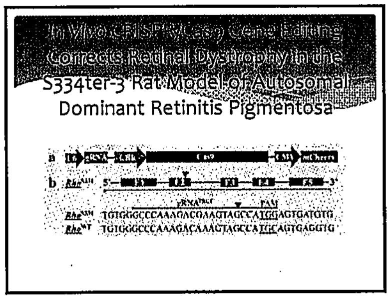

CRISPR/Cas9 Gene Editing in RP

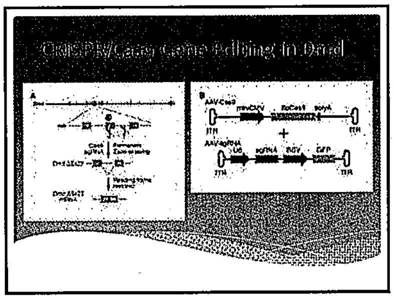



Glomerulonephritis Type II and Drusen
membranoproliferative glomerulonephritis type II
20/20
numerous small drusen in the posterior pole temporal to the fovea


Science
Postnatal genome editing partially restores dystrophin expression in a mouse model of muscular dystrophy

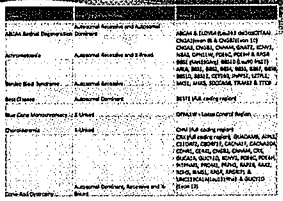

From: Phenotypic Variation in Enhanced S-cone Syndrome


In Vivo CRISPR/Cas9 Gene Editing Corrects Retinal Dystrophy in the S334ter-3 Rat Model of Autosomal-Dominant Retinitis Pigmentosa



The Carver Genetic Testing Laboratory

THE UNIVERSITY OF IOWA

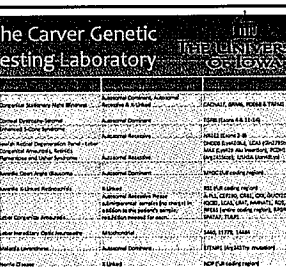


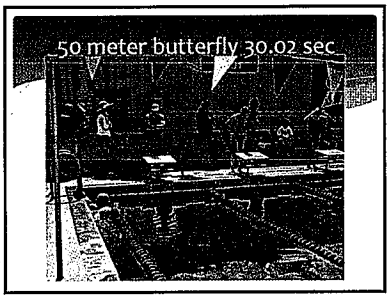
TAKE HOME POINTS

- LUMP
- FULL DISCERNMENT ONLY WITH USING FAMILY DATA / MEMBERS / MOLECULAR GENETICS
- FAMILY CONTEXT
- ALL DRUSEN ARE CLINICALLY ALIKE
- NORTH CAROLINA MACULAR DYSTROPHY / MCDR1 mutations affect PRDM13 expression
- PERSEVERANCE

The Carver Genetic Testing Laboratory

THE UNIVERSITY OF IOWA





The Carver Genetic Testing Laboratory

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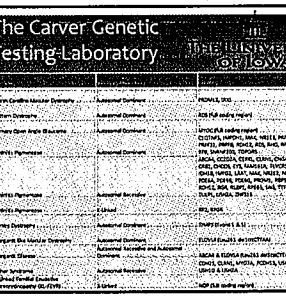

CURRICULUM VITAE

NAME: Kent W. Small, M.D. DATE: 12/2014

PRESENT POSITION AND ADDRESS:

Scientist: Cedars-Sinai Medical Center, Regenerative Medicine Institute
President / Founder Kent W. Small, M.D. A Medical Corporation
President / Founder Macula and Retina Institute
President / Founder: Molecular Insight LLC, A Research Corporation
President / Founder: Molecular Insight Research Foundation (501C3)
8635 W. 3rd Street , Suite 395 W, Los Angeles, CA 90049 310-659-2200
501 N Orange St. Suite 250, Glendale, CA 91203 818-552-5040

BIOGRAPHICAL: Date and place of birth: 19 October 1956, New Orleans, LA
Citizenship: US
Home address: 3134 Corda Dr. , Los Angeles, CA 90049

EDUCATION:

Aug 1974 – Aug 1977, Zoology, None, Louisiana State University
Aug 1977 – June 1981, Medicine, M.D. , Tulane University
May 1979 – Aug 1979, Cancer Research Externship during medical school, None, Tulane University
Jul 1981 – Jul 1985, Resident in General and Cardiothoracic Surgery, None, Duke University
Jul 1983 – Jul 1985, Research Fellow in Surgical Cardiac Electrophysiology, None, Duke University
Jul 1985 – Jul 1988, Resident in Ophthalmology, None, Duke University
Jul 1988 – Jan 1991, Fellow in Vitreoretinal Surgery, None, Duke University
Jan 1989 – Jan 1991, Research Associate Neurogenetics

PROFESSIONAL WORK HISTORY AND TEACHING EXPERIENCE:

Jul 2004 – Sept. 2004 Professor and Chair, Department of Ophthalmology and Visual Sciences, UTMB, Galveston, Texas
Jul 1998 – Jun 2004, Professor w/tenure, Jules Stein Eye Institute, UCLA, Los Angeles, CA
Aug 1994 – Jun 1998, Associate Professor w/tenure, Jules Stein Eye Institute, UCLA, Los Angeles, CA
Jun 1994 – Aug 1994, Associate Professor w/tenure, Department of Ophthalmology, Biochemistry, and Molecular Biology, University of Florida, Gainesville, FL
Aug 1992 – Jun 1994, Assistant Professor tenure track, Department of Ophthalmology, Biochemistry and Molecular Biology, University of Florida, Gainesville, FL
Jan 1992 – Aug 1992, Research Associate, Neurogenetics, Department of Medicine, Division of Neurogenetics, Duke University, Durham, NC
Jan 1991 – Jan 1992, Assistant Professor tenure track, Department of Ophthalmology School of Graduate Studies, Medical University of South Carolina, Charleston, SC

RESEARCH ACTIVITIES:

A. Area of research: Molecular genetics of macular and retinal degenerations as

well as inherited corneal diseases, inherited eyelid dysmorphology, and inherited optic nerve abnormalities. Stem cells fetal cortical neural progenitor cells to treat macular and retinal degenerations.

B. Grant support:

Contracts and Grants:
Previously Funded

1. NRSA (National Research Service Award / NEI) Amount: $40,000. Duration: 2 years. Title: The effect of total body irradiation on corneal neovascularization. Principal Investigator.: Kent W. Small, M.D. 1985, was asked to decline by Duke Eye Center to begin residency in ophthalmology one year earlier.
2. Jacob Javits Fellow: National Institutes of Health/NINDS. amount $60,000, duration: 1988-1989. Title: Genetics mapping of inherited diseases. Principal Investigator.: Kent W. Small, M.D.
3. K11 Physician Scientist Award, National Institutes of Health, National Eye Institute, EY00313. Principal Investigator.: Kent W. Small, M.D. Amount/Years Duration: 5 years, $360,000; Title: Genetics Studies of North Carolina Macular Dystrophy. 1989-1994.
4. Jules and Doris Stein Research to Prevent Blindness Professorship. Amount: $350,000; Duration: 5 years. Additional $100,000 for equipment. Title: Molecular Genetics Studies of Ocular Diseases. 1993-1994. Principal Investigator: Kent W. Small, M.D.
5. Retinitis Pigmentosa Foundation.; Amount: $50,000. Title: Cloning the Gene for Dominant Cone Degeneration. 1993-1995. Principal Investigator: Kent W. Small, M.D.
6. Division of Sponsored Research for University of Florida. Inherited Ocular Disease-gene mapping. Duration: 1 year, 1993, $15,000, Principal Investigator: Kent W. Small, M.D.
7. National Institutes of Health, National Eye Institute RO-1 - EY10239 Duration: 5 years; Amount: $987,000 Direct Cost. Title: Hereditary Macular Degenerations. 8/93-8/98. Principal Investigator: Kent W. Small, M.D.
8. The Foundation Fighting Blindness: 9/96-8/99, 180,000, Cloning the gene for autosomal dominant cone degeneration. Principal Investigator: Kent W. Small, M.D.
9. Ciba-Giegy Photodynamic Therapy for Age-related macular degeneration., $10,000. 11/1/99-3/2000
Participating surgeon: Kent W. Small, M.D
10. National Institutes of Health, National Eye Institute, Submacular Surgery Trials (SST): age-related macular degeneration. participating surgeon 7/1998 – 6/2000.
11. Agouron Pharmaceuticals, Oral AG3340 for the treatment of choroidal neovascularization in age-related macular degeneration, PI at UCLA center, Principal -Investigator: Kent W. Small, M.D. $100,000, 1998-2001
12. National Institutes of Health, National Eye Institute RO-1 – EY11645: Duration: 3 years; Amount: $700,000 Direct Cost. Title: Cloning the blepharophimosis gene. 1998-2001. Principal Investigator: Kent W. Small, M.D
13. Lu-Tex Photodynamic Therapy for AMD, (Alcon Laboratories). A phase I trial, a dose – ranging study of Lutetium Texaphyrin injectable formulation in the photodynamic treatment of subfoveal choroidal neovascularization. Participating / treating physician: Kent W. Small, M.D.
14. Alcon Laboratories, Inc. An evaluation study on the safety and efficacy of Anecortave Acetate Sterile Suspension vs. placebo following sub-tenon's injections for the inhibition of Neovascularization in patients. C-98-03 (Schwartz, PI) 12/01/2000 – 12/01/2003 $225,000 Kent W. Small, M.D. Role: Co-Investigator
15. ROI EY11645 (Kent W. Small, PI) 08/01/2001 – 07/31/2004
NIH/NEI $200,000
Cloning the gene defect causing blepharophimosis syndrome
The major goals of this project are to identify the mutations causing the blepharophimosis syndrome
16. Muscular Dystrophy Association (Kent W. Small, PI) 07/01/2003 – 06/30/2006
Hereditary and Motor and Sensory Neuropathy Type (CMT) IV, $63,410
Gene Mapping and Positional Cloning
This project aims to identify the gene responsible for a specific neuromuscular disease that causes muscle weakness and otpic nerve atrophy, Charcot-Marie-Tooth type 6 (CMT6).

17. The Foundation Fighting Blindness (Bok, Center Coordinator) 07/01/00 – 06/30/05
JSEI Center Core Grant $121,137 (KWS Portion)
Module V: Cloning the gene for autosomal dominant cone degeneration (CORD5)
The major goals of this project are to identify the genetic basis of an inherited retinal degeneration, CORD5.
Role: Kent W. Small, M.D.: Principal Investigator of Module V

18. P 30 EY00331 (Hubbell, PI) 03/01/99-02/28/04
NIH/NEI $291,029 (Core)
Core Center Grant for Vision Research
The major goal of this grant is to provide shared use of core facilities among 17 investigators.
Role: Kent W. Small, M.D.: Investigator

19. T32 EYE07026 (Farber, PI) 09/30/00-09/29/05
NIH/NEI $248,248
Vision Research Training Grant, Biochemistry of Cone Visual Cells
This project funds predoctoral and postdoctoral fellows in vision science (fourteen mentors in Ophthalmology).
Role: Kent W. Small, M.D. Investigator

20. R01 MH63764-01A1 (Kumar, PI) 5/1/2002-4/30/2006
NIH/National Institute of Mental Health
Cerebrovascular Basis of Depression in Diabetes-NIDDM
Role: Kent W. Small, M.D.: Co-Investigator

21. CIRM (California Institute for Regenerative Medicine) Neural progenitor 1/2015-1/2018
cells for the treatment of retinitis pigmentosa. $15,000,000
Role: Kent W. Small, MD Co- Investigator accepted

CLINICAL TRIALS

Amicus (Small sub-investigator)
Protocol GAU-CL-202 for the treatment of Gaucher's Disease 4/09-1/10
$300

SRFR-001 (Small PI) 9/07-9/09
Sirion Therapeutics Inc $179,445
A Phase II Multicenter, Randomized, Double-Masked, Placibo-Controlled, Dose Comparison Study of the Safety and Efficacy of Fenretinide in the Treatment of Geographic Atrophy in Subjects with Age Related Macular Degeneration

VGFT-OD-0605 (Small PI) 9/07-12/08
REGENERON INC $272,890
A PHASE III Multicenter, Randomized, Double-Masked, Controlled, Dose Comparison Study of the Safety and Efficacy VEGF-TRAP (VGFT-OD-0605) in Subjects with Exudative Age related Macular Degeneration

RADICAL (Small PI) 10/07-10/09
QLT INC $287,500
A Phase III, Multicenter, Randomized, Double-Masked, Controlled, Dose Comparison Study of the Safety and Efficacy of Photodynamic Therapy in Combination with Intravitreal Lucentis Injections Compared to Lucentis Injections as Monotherapy.

Brinzolamide / Timolol Study Group (Small PI) 10/06-10/07
ALCON RESEARCH LTD $25,000

Efficacy of Brinzolamide 1% / Timolol 0.5% Fixed Combination Compared to Brinzolamide 1% and Timolol0.5%.

FVF2508g (Small PI) 05/15/2003 – 12/31/2004
Genetech, Inc. $15,429
Protocol FVF2508g, "An Extension Study to Evaluate the Safety and Tolerability of Multiple-Dose Intravitreal Injections of rhuFabV2 in Subjects with Neovascular Age-Related Macular Degeneration Who Have Completed the Treatment Phase of a Genetech-Sponsored Phase I or Phase I/II rhuFab V2 Study"

SMS995 804 (Small, PI) 11/13/2000 – 01/01/2006
Novartis Pharmaceutical $359,562
Protocol SMS9950804, "A Randomized, Controlled Study on the Efficacy and Safety of Sandostatin LAR in the Therapy of Patients with Moderately severe or non-proliferate retinopathy (NPDR) or low risk proliferate diabetic retinopathy (PDR).

EOP1005C (Gonzales, PI) 9/6/2002-9/5/2004
EyeTech Pharmaceuticals, Inc. $147,459.38
"A Phase II Randomized, Controlled, Double-Masked, Dose-Finding, Multi-Center, Comparative Trial, in Parallel Groups, to Establish the Safety and Preliminary Efficacy of Intravitreal Injections of EYE001 (Anti-VEGF Pegylated Aptamer), Given Every 6 Weeks for 12 to 30 Weeks to Patients with Clinically Significant Diabetic Macular Edema (CSME) Involving the Center of the Macula".
Kent W. Small, M.D. Role: Co-Investigator

EOP1006B (Gonzales, PI) 2/6/2003-2/11/2004
EyeTech Pharmaceuticals, Inc. $159,336.00
A Randomized, Double-Masked, Multi-Center Trial of the Safety, Tolerability and Pharmacokinetics of 1 Mg/Eye and 3 Mg/Eye Intravitreal Injections of Pegaptanib Sodium (Anti-VEGF Pegylated Aptamer) Given Every 6 Weeks for 54 Weeks in Patients with Exudative Age-Related Macular Degeneration (AMD). Kent W. Small, M.D. Role: Co-Investigator

EOP1004 (Gonzales, PI) 10/16/2001-10/15/2003 (?)
EyeTech Pharmaceuticals, Inc. $171,529.00
A phase II/III randomized, double-masked, controlled, dose-ranging Multicenter comparative trial, in parallel groups to establish the safety and efficacy of intravitreal injections of EYE-001 (anti-VEGF pegylated aptamer) given every six weeks for 54 weeks in patients with exudative Age-Related Macular Degeneration. Kent W. Small, M.D. Role: Co-Investigator

Submacular Surgery Trials (Schwartz, PI) 05/01/2003 – 04/30/2004
Johns Hopkins University/NIH/NEI $56, 180
A randomized, multicenter trial to evaluate submacular surgery for removal of subfoveal choroidal neovascular lesions in selected patients with Age-Related Macular Degeneration (AMD) compared to observation, patients with new, large and /or poorly demarcated subfoveal lesions.
KWS Role: Co-Investigator

B7A-MC-MBCM (Schwartz, PI) 08/27/2001 – 03/01/2005
Lily Research Laboratories $167,565
Protein Kinase C B Inhibitor – Diabetic Retinopathy Study 2(PKC-RS2), A Phase III Clinical Trial.
Kent W. Small, M.D. Role: Co-Investigator

FVF2508g (Kent W. Small, M.D. PI) 05/15/2003 – 12/31/2004
Genentech, Inc. $15,429
"An Extension Study to Evaluate the Safety and Tolerability of Multiple-Dose Intravitreal Injections of rhuFabV2 in Subjects with Neovascular Age-Related Macular Degeneration Who Have Completed the Treatment Phase of a Genentech-Sponsored Phase I or Phase I/II rhuFab V2 Study"

SMS995 804 (Kent W. Small, M.D. PI) 11/13/2000 – 01/01/2006
Novartis Pharmaceutical $359,562

Protocol SMS9950804, "A Randomized, Controlled Study on the Efficacy and Safety of Sandostatin LAR in the Therapy of Patients with Moderately severe or non-proliferate retinopathy (NPDR) or low risk proliferate diabetic retinopathy (PDR).

FVF2587g (Kent W. Small, M.D. PI) 02/10/2004 – 03/31/2006
Genentech, Inc. $840,292.00
"A Phase III, Multicenter, Randomized, Double-Masked, Active Treatment-Controlled Study of the Efficacy and Safety of rhuFab Vs (Ranibizumab) Compared to Verteporfin (Visudyne) Photodynamic Therapy in Subjects with Predominantly Classic Subfoveal Neovascular Age-Related Macular Degeneration"

Iridex (PI: Kent W. Small, MD) 10/20/2000-4/20/2005
Transpupillary Thermotherapy of Occult Subfoveal Choroidal Neovascular Membranes in Patients with Age-Related Macular Degeneration (The TTT4CNV Clinical Trial)

Valeant, (PI Kent W. Small, MD) 2011-2014
Long lasting effects of photodynamic therapy as combination therapy with anti-VEGFs. $10,000
Investigator Initiated Trial

CIRM (California Institute for Regenerative Medicine) (PI Shaomei Wang PhD) 2015-2018
IND-enabling study of subretinal delivery of human neural progenitor cells $5,000,000
for the treatment of retinitis pigmentosa

Patents: U.S. Patent Application Serial No. 12/044,833 For: Suspended Slit Lamp Perkins Coie Ref. No.: 65677.8001.US00

Other Research Interests: (NOT FUNDED)

1. Silicone oil clinical trial. Principal Investigator: Kent W. Small, M.D. 1992-93
2. Vitrectomy endophthalmitis clinical trial. Co-Investigator: Kent W. Small, M.D. 1992- 1993
3. Richard James Silicone Oil Investigation. Co-Investigator: Kent W. Small, M.D. 1994-1996
4. Intraoperative Use of Perfluorocarbon Liquids: Co-Investigator: Kent W. Small, M.D. 1994-1996
5. Gilead Sciences, duration 1 year. Intravitreal injection of cidifovir for recurrent CMV retinitis in AIDS patients. Principal Investigator: Kent W. Small, M.D. 1995-1996
7. Laser to Drusen Trial. Principal Investigator for the Jules Stein Eye Institute: Kent W. Small, M.D. 1994-1996
8. Surgical Repair of Macular Holes Research Study. Co-Investigator: Kent.W. Small, M.D. 1994-present
9. The artificial retina. Principal Investigator: Kent W. Small, M.D., Co-Investigators William Dawson, Ph.D., and Kieth Rambo, Ph.D. (University of Florida) 1992- 1997
10. Surgical Repair of Macular Holes with Silicone Oil. Principal -Investigator: Kent W. Small, M.D. 1997-1999
11. Treatment of choroidal neovascularization from age-related macular degeneration with low dose external beam irradiation. AMDRT 1997-present. Principal Investigator: Kent W. Small, M.D.
12. The Effectiveness of a Reading Training Program in Low Vision Patients with Age-related Macular Degeneration. Principal Investigator: Melissa Chun, O.D., Co-Investigator Kent W. Small, M.D. 1/2000-12/20001, no funding
13. Vervet Research Colony as a Biomedical Resource , P40 Core support grant to NCRR, Lynn Fairbanks, PI, Kent Small, M.D., collaborator 2001- 2004

COMMITTEE RESPONSIBILITIES:

A. National
B. UTMB
C. Departmental

D. Other

Administrative duties, University Governance and Service (Florida):
1. Search Committee for Department of Ophthalmology Chairman -1993-94.
2. Strategic Planning Committee for Center for Mammalian Genetics - 1992 - 94.
3. Residency Selection Committee, Department of Ophthalmology -1992-94.
4. Assistant Director of M.D./Ph.D. program at the University of Florida - 1993 - 94.
5. Member C.Q.I. (continuing quality improvement) Physician Facilitation team Shands Hospital (one of 12 selected physicians at U.F. to train other U.F. physicians in the C.Q.I. process - a new technique in assessing systems efficiencies) 1993- 94

Administrative duties, University Governance and Service (UCLA) Jul 1998 – Jun 2004:
1. Director of the Macula Center at the Jules Stein Eye Institute
2. Advisor: Low Vision Center at The Jules Stein Eye Institute
3. Dept. of Ophthalmology Clinical Committee: member
4. Dept. of Ophthalmology Research Committee: member
5. Dept. of Ophthalmology Committee (provides policies and procedures for the academic Senate): member
6. Dept. of Ophthalmology Appointment, Academic Advancement and Promotion Committee: member
7. Jules Stein Eye Institute Committee (for organized research units)
8. Quality Assurance Committee of the Ophthalmology Service, UCLA Medical Center Medical Staff: member
9. Dept. of Ophthalmology Quality Assurance Committee: member
10. Dept. of Ophthalmology, Ophthalmic Genetics Center: member
11. Jules Stein Eye STAR Program Committee: member
12. Vision Genetics Center: member

TEACHING RESPONSIBILITIES AT UTMB:
A. Medical School
B. GSBS, SAHS and SON as applicable
C. Other

Presentations (abstracts presented at meetings)
Regional:

Annual Spring Cardiovascular Symposium, Duke University Medical Center "Changes in myocardial resistivity during global ischemia: On-line identification of the onset of severe but reversible ischemic injury." 10/85

North Carolina Medical Society Meeting 132nd Annual Scientific Session, Asheville, NC,"Vitamin A deficiency in American adults." 6/86

McPherson Hospital Scientific Presentations, Durham, NC "Pseudotumor oculi". 5/85

McPherson Hospital Scientific Presentations, Durham, NC "Fundus findings in primary oxalosis." 5/87

Jackson Mutant Mouse Lab, Bar Harbor, ME, "Genetics of macular degeneration" 7/89

North and South Carolina Ophthalmological Society, Charleston, SC "Electrophysiological Tests" 5/91

Duke University Alumni Meeting, Durham, N.C., "North Carolina Macular Dystrophy in Belize" 4/93
San Antonio Ophthalmological Society, "Ocular Genetics for the Clinician" 9/93

National

Christian Ophthalmology Society, Hilton Head, SC "Vitamin A and the eye" 7/88

Combined Retina Society and Macular Society Meeting, Boston, MA "North Carolina macular dystrophy: update" 5/89

Ophthalmologic Genetics Study Club, New Orleans, LA "North Carolina macular dystrophy: genetics studies." 10/89

International Society of Genetics Eye Diseases, Atlanta, GA, "Genetics exclusion map of North Carolina macular dystrophy" 10/90

Retina Society, Key Largo, FL. "Pigmented paravenous retinochoroidal atrophy, discordant expression in monozygotic twins." 10/90

Christian Ophthalmology Society Meeting. Callaway Gardens, GA "Ocular Genetics Update." 6/91

Ophthalmologic Genetics Study Club, Anaheim, CA, Oct. "Alstrom's, a case misdiagnosed as Bardet-Biedel's". 10/91

Ophthalmologic Genetics Study Club, Anaheim, CA, "Autosomal dominant cone degeneration, a large single family study" 10/91

Association of University Radiologists, Chicago, IL, Keuthe DO, Small KW, Blinder RA: Are large magnetic fields safe for patients with metallic retinal tacks? 5/89.

FASEB Summer Symposium, Copper Mountain, CO, 7/95 Small KW. High resolution genetic map of the MCDR1 locus

International
International Society of Ocular Pathology, New Orleans, LA "Primary oxalosis: a clinicopathologic study" 10/89

International Pediatric Nephrology Association, Paris, France: Scheinman JI, Fallon MD, Small KW, Mahan JD, Letson RD, Klintworth GK: Primary hyperoxaluria (PHO): Common mitogenic effects on bone and retina. 6/89

Canadian Association of Physicists, Division of Medical and Biological Physics. Keuthe DO, Small KW, Blinder RA: Dynamic similarity to the Maxwell equations to determine if patients with metallic tacks in their eyes are safe in large magnets. 6/89

International Society of Genetics Eye Disease, Atlanta, GA. Small KW, Vance JM, Jones, MA, Hung W-Y, Yamaoka L, Roses AD, Pericak-Vance MA: Genetics linkage analysis in North Carolina macular dystrophy. 9/90

Walsh Neuroophthalmology Society Small KW, Buckley EG: Recurrent unilateral ptosis. Vancouver, British Columbia, CAN 8/88

European Society of Human Genetics "Mutation screening of the BIGH3 gene in patients with Keratoconus".Strasbourg, France, May 25-28th 2002.

Human Genome Organization, HUGO "Characterization of mutations within the FOXL2 gene in Blepharophimosis Ptosis Epicanthus Inversus patients and its evolutionary conservation in Fugu". 2002, Shanghai, China April 2002.

International Congress Of Human Genetics "Identification and Characterization of mutations in families affected with Corneal dystrophy". 10th 2001 Vienna, Austria, May 15 - 19, 2001

American Society of Human genetics "Characterization of mutations in families affected with Corneal dystrophy"., 50th ASHG, Pennsylvania, Philadelphia, 3-7 Oct 2000.

Human Genome Organization,"Physical map of North Carolina Macular degeneration – MCDR1 locus". HUGO'99, Brisbane, Australia 1999.

HUGO (Human Genome Organization) 2002, Shanghai, China April 2002., N. Udar , V. S. Yellore, M. Chalukya, S. Yelchits, R. Silva-Garcia, K. Small "Characterization of mutations within the FOXL2 gene in Blepharophimosis Ptosis Epicanthus Inversus patients and its evolutionary conservation in Fugu".

INTERNATIONAL CONGRESS OF HUMAN GENETICS 2001 Vienna, Austria, May 15 - 19, 2001, N. Udar , V. S. Yellore, M. Chalukya, S. Yelchits, R. Silva-Garcia, K. Small "Identification and Characterization of mutations in families affected with Corneal dystrophy".

10th HUGO '99, Brisbane, Australia 1999. N. Udar , V. S. Yellore, M. Chalukya, S. Yelchits, R. Silva-Garcia, K. Small "Physical map of North Carolina Macular degeneration – MCDR1 locus".

Invited Lectures:
Regional:

Braille Institute, Los Angeles, CA "Macular degeneration: there is hope", 3/97

Midway Hospital Medical Center, Los Angeles, CA, New Advances in Retina: "New techniques in the management of diabetic retinopathy and venous occlusive disease. 1/97

Braille Institute, Los Angeles, CA "Macular degeneration: new therapies", 3/98

Southern California College of Optometry, Fullerton, CA, Retina and Low Vision Symposium, 1/99, "Macular diseases" University of California , Berkeley School of Optometry. The Sixth Annual Continuing Education in Southern California Lectureship and Symposium. 2/99

Cedars-Sinai Medical Center, Genetic Disease and the Eye. "Genetics of Macular Degeneration" Los Angeles, CA 1/99

Foundation Fighting Blindness, Symposium on new research in macular degeneration. Invited Speaker, Los Angeles, CA 4/99

Visions 99'. Sponsored by The Foundation Fighting Blindness. "Age-related macular degeneration, Stargardt's disease and Best's disease." July 1999.

Los Angeles Research Study Club, Universal City, CA, invited speaker, "Genetics of age-related macular degeneration" 1/2000

Greater Los Angeles Coding Network (GLACN) and Southern California Health Information Association (SCHIA). Ophthalmology and coding issues. Los Angeles, CA 2/2001

Greater Los Angeles Coding Network (GLACN) and Southern California Health Information Association (SCHIA). Ophthalmology and coding issues. Los Angeles, CA 3/2002

California Academy of Ophthalmology: Age-related macular degeneration, update. Santa Monica, CA, 9/2002

Nebraska Academy of Ophthalmology: Age-related macular degeneration, update. Omaha, Nebraska 9/2002

Nebraska Academy of Ophthalmology: The macular dystrophies. Omaha, Nebraska 9/2002

Braille Institute, Los Angeles, CA "Macular degeneration: new therapies", 4/03
R and R: Retina and Rehabilitation. Invited keynote speaker "From Molecules to Magnifiers" 10/17/03 Point Clear AL

8th Annual UCLA Research Conference on Aging, "Age-related macular degeneration, from molecules to magnifiers" Los Angeles, 6/03

Braille Institute, Los Angeles, CA "Macular degeneration: update", 3/04

Braille Institute, Los Angeles, CA "Macular degeneration: update", 3/05

Discovery Eye Foundation: "Macular Degeneration: update" 10/09 Skirball Center, Los Angeles, CA

Blinded Veterans Association, Wadsworth VA Hospital, "Macular Degeneration: update" 3/14/2010

National

Association of Ophthalmic Photographers, Anaheim, CA "Using the hand held fundus camera". 10/91

University of Texas Southwestern, Dept. of Ophthalmology Dallas, TX, 1990, Hereditary macular degeneration.

University of Louisville, Dept. of Ophthalmology, Louisville, KY, 1990, Hereditary macular degeneration.

Washington University, Dept. of Ophthalmology, St. Louis, MO, 1990, Hereditary macular degeneration.

University of Texas, Dept. of Ophthalmology, San Antonio, TX, 1990, Hereditary macular degeneration.

Medical University of South Carolina, Dept. of Ophthalmology, Charleston, SC, 1990, Hereditary macular degeneration.

Medical College of Georgia, Dept. of Ophthalmology, Augusta, GA, 1990, Hereditary macular degeneration.

Hershey Medical Center, Dept. of Ophthalmology, Hershey, PA, 1990, Hereditary macular degeneration.

Johns Hopkins, Wilmer Eye Institute, Dept. of Ophthalmology, Baltimore, MD, 1991, Hereditary macular degeneration.

National Institutes of Health, National Eye Institute, Bethesda, MD, 1991, Hereditary macular degeneration.

University of West Virginia, Dept. of Ophthalmology, Morganton, W.Va., 1991, Hereditary macular degeneration.

Harvard Medical School, Massachusetts Eye and Ear Infirmary, Dept. of Ophthalmology, Boston, MA, 1991, Hereditary macular degeneration.

Pacific Medical Center, Dept. of Ophthalmology, Smith-Ketterwell Eye Research Institute, San Francisco, CA, 1991, Hereditary macular degeneration.

University of Nebraska, Dept. of Ophthalmology, Omaha, NE, 1991, Hereditary macular degeneration.

University of Minnesota, Dept. of Ophthalmology, Minneapolis, MN, 1991, Hereditary macular degeneration.

University of Cincinnati, Dept. of Ophthalmology, Cincinnati, OH, 1992, Hereditary macular degeneration.

University of South Florida, Dept. of Ophthalmology, Tampa, FL, 1992, Hereditary macular degeneration.

University of Florida, Dept. of Ophthalmology, Gainesville, FL, 1992, Hereditary macular degeneration.

University of Wisconsin, Dept. of Ophthalmology, Madison, WI, 1992, Hereditary macular degeneration.

Yale University, Dept. of Ophthalmology, New Haven, CT, 1992, Hereditary macular degeneration.

University of Texas, Houston, Dept. of Ophthalmology, Houston, TX, 1992, Hereditary macular degeneration.

Geisinger Medical Center, Dept. of Ophthalmology, Danville, PA, 1992, Hereditary macular degeneration.

University of Michigan, Dept. of Genetics, Ann Arbor, MI, 1992, Hereditary macular degeneration.

University of South Carolina, Dept. of Ophthalmology, Columbia, SC, 1992, Hereditary macular degeneration.

Texas Tech, Dept. of Ophthalmology, Lubbock, TX, 1992, Hereditary macular degeneration.

University of Minnesota, Dept. of Ophthalmology, Minneapolis, MN, 1992, Hereditary macular degeneration.

Tulane University School of Medicine, Dept. of Ophthalmology, New Orleans, LA, 1993, Hereditary macular degeneration.

Oschner Medical Center, Dept. of Ophthalmology, New Orleans, LA, 1993, Hereditary macular degeneration.

University of California, Los Angles (UCLA), Jules Stein Eye Institute, Dept. of Ophthalmology. 1994, Hereditary macular degeneration.

University of South Carolina, Dept. of Ophthalmology, Columbia, SC, 1994, Hereditary macular degeneration, B. cereus endophthalmitis

University of Pennsylvania, Dept. of Ophthalmology, Philadelphia, PA, 1994, Hereditary macular degeneration.

University of Wisconsin, Dept. of Ophthalmology, Madison, Wisconsin, 1994, Hereditary macular degeneration.

University of California, Berkeley, Berkeley, CA, 1995, Genetics of Myopia

University of Alabama, Birmingham, AL, Dept. of Ophthalmology, 1996, Hereditary macular degeneration.

Vanderbilt University, Nashville, TN, 1997, Hereditary macular degeneration.

University of California, Davis, 1998, Hereditary macular degeneration.

Mid Winter Vitreo-retinal Surgery Course, Sarasota, FL, "Should we treat retinitis pigmentosa with vitamins?" 2/94

Mid Winter Vitreo-retinal Surgery Course, Sarasota, FL, "The future of molecular genetics in ophthalmology" 2/94

Retina Society, Santa Fe, NM. "Autosomal Dominant Cone Degeneration Maps to Chromosome 17p" 10/95

Macula Society, Tucson, Arizona "North Carolina macular dystrophy in Central America" 2/96

Western Retina Study Club, Yosemite, CA. "Autosomal Dominant Cone Degeneration Maps to Chromosome 17p" 3/96

WAVE (Western Association of Vitreo-retinal Education), Maui, Hawaii, "Wide angel viewing using the AVI contact lens system" 7/96

WAVE (Western Association of Vitreo-retinal Education), Maui, Hawaii, "Perfluorocarbon liquids: indications, techniques and complications" 7/96

WAVE (Western Association of Vitreo-retinal Education), Maui, Hawaii , "ICG and Digital Imaging Experience at the Jules Stein Eye Institute/UCLA" 7/96

Joint Commissions of Allied Health Professional Organization. Chicago, IL "Genetics of Eye Diseases" 10/96

Aspen Retinal Detachment Society, Aspen, CO, "Hereditary macular degenerations" 3/97

Aspen Retinal Detachment Society, Aspen, CO, "The artificial retina", 3/97

University of California at Davis, Sacramento, CA. 1/98 "Age-related macular degeneration and other inherited macular degenerations", grand rounds visiting professor

University of California, San Francisco, Dept. of Ophthalmology. 1998, Hereditary macular degeneration. grand rounds visiting professor

Medical University of South Carolina, Dept. of Ophthalmology, Charleston, SC, 4/99, "Hereditary macular degenerations" grand rounds visiting professor

Medical College of Virginia, Dept. of Ophthalmology, Richmond, VA. 3/2000 "Hereditary macular degenerations" grand rounds visiting professor

Small KW, Vu I, Glasgow B, Flannary J. Histopathologic study of North Carolina macular dystrophy. American Ophthalmological Society, Hot Springs, VA 5/2001 Abstract #24

University of Miami, Bascom Palmer Eye Institute, "North Carolina macular dystrophy". 3/2003

Louisiana State University Health Science Center. GUCY2D Gene Mutations in CORD5 Families and Evidence of Incomplete Penetrance 4 / 03

University of California, Irvine, GUCY2D Gene Mutations in CORD5 Families and Evidence of Incomplete Penetrance 5 03

University of California Irvine, "North Carolina macular dystrophy" 5/03

University of California Los Angeles, Center on Aging, invited Plenary speaker, "Macular Degeneration, from Molecules to Magnifiers" 6/03

International

First International Workshop of Human Chromosome 6, sponsored by HUGO (Human GenomeOrganization). Ann Arbor, Michigan, "North Carolina macular dystrophy maps to 6q14-q16.2". 6/92

University of Lille, France, 1998, North Carolina macular degeneration and the genetics of age-related macular degeneration

Hospital Evitar La Cieguería, Mexico City, Mexico, 1996, Hereditary macular degeneration.

University of Iceland, Reykjavik, Iceland, 1998, Genetics of age-related macular degeneration.

ARVO (Association for Research in Vision and Ophthalmology) SIG (special interest group) Ft. Lauderdale, FL, 1995: "Hereditary macular degenerations" 5/95

American Academy of Ophthalmology, Atlanta, GA, ARVO Sponsored Special Symposium on "Advances in Molecular Genetics and Their Clinical Impact on Retinal and Choroidal Diseases" 10/95

Coloquio Manejo Medico Quirurgico del Trauma de Segmento Posterior sponsored by the La Sociedad Mexicana de Oftalmogia y La Association Mexicana de Retina, "Masa inflamatoria retineana subsecuente a trauma" 5/96

International European Union Meeting on "Genetics of Macular Degeneration", Amsterdam, Netherlands, "North Carolina macular dystrophy" 6/97

Macula Society, Florence, Italy, "North Carolina macular dystrophy in France" 6/97

Macula Society, Florence, Italy, Moderator: Inherited retinal and macular degenerations, 6/97

American Academy of Ophthalmology, 1997 Subspecialty Day, San Francisco, CA, "What to tell patients about the genetics of age-related macular degeneration" 10/97

American Academy of Ophthalmology, 1997 Subspecialty Day, San Francisco, CA, "Hereditary macular dystrophies" 10/97

The Retina Society Vancouver, British Columbia, CAN "North Carolina macular dystrophy (MCDR1) family in Texas maps to Chromosome 6q16" 9/97

American Academy of Ophthalmology, Vitreoretinal Update 1997. Invited Speaker "What do we tell our patients about the genetics of age-related macular degeneration?" San Francisco, CA, 10/97

American Academy of Ophthalmology, invited discussant, "Phenotype-genotype correlations of the keratoepithelin gene. 10/97

ARVO SIG (special interest group) Ft. Lauderdale, FL, "Hereditary macular degenerations", 5/97

American Academy of Ophthalmology, 1998, New Orleans, LA. Invited Speaker for ARVO symposium, "Inherited macular diseases."

Macula Society, San Diego, CA , Chair: Inherited retinal and macular degenerations, 2/99

Icelandic Ophthalmologic Society, invited speaker, "The genetics of age-related macular degeneration", Reykjavek, Iceland 3/99

Icelandic Ophthalmologic Society, invited speaker, "Inherited macular dystrophies", Reykjavek, Iceland 3/99

American Academy of Ophthalmology and the Pan-American Association of Ophthalmology. Course #602 "Surgical Management of Intraocular Infections" 1999, Orlando, Fl

Macula Society, Puerto Rico, "Histopathologic studies of dominant cone degeneration (CORD5)" 2/2000

33rd Panhellenic Ophthalmologic Congress; Thessaloniki, Greece, 5/24-27, 2000. "The genetics of age-related macular degeneration"

33rd Panhellenic Ophthalmologic Congress, Thessaloniki, Greece, 5/24-27, 2000. "Round table symposium new surgical methods in macular disease"

33rd Panhellenic Ophthalmologic Congress, Thessaloniki, Greece, 5/24-27, 2000. "Round table symposium photodynamic therapy in age related macular degeneration"

Retina Society, Coral Gables, FL, Silicon oil loss. 11/2000

Invited Speaker: Braille Institute, Los Angeles, CA 3/2001, "Update of macular degeneration"

American Ophthalmological Society : Small KW, Vu I, Glasgow B, Flannary J. Histopathologic study of North Carolina macular dystrophy. Hot Springs, VA 5/2001 Abstract #24

Volunteer Physician Lectures: Rotary Humanitarian Projects Sponsored Eye Clinic in Depensar, Indonesia 8/3-8/2001

2nd Updates in Ophthalmology, Singapore National Eye Institute, "Submacular Surgery for choroidal neovascularization" 8/12/2001

2nd Updates in Ophthalmology, Singapore National Eye Institute, Chaired session on Age-related macular degeneration. 8/12/2001

2nd Updates in Ophthalmology, Singapore National Eye Institute, "Clinical approach to macular dystrophies" 8/12/2001

Naples, Fl , Macula Society, 3/2003. GUCY2D Gene Mutations in CORD5 Families and Evidence of Incomplete Penetrance

Queen's University, Belfast Ireland, invited speaker, "Age-Related Macular Degeneration, from Molecules to Magnifiers." 9/03

Anaheim, CA , Subspecialty Day, American Academy of Ophthalmology, invited speaker, "Genetic Testing in the Clinic" 11/03

Boulder, CO , SiRNA Therapeutics, "Therapy of ocular diseases." 1/8/04 SIRNA,

Barcelona, Spain, invited guest lecturer by Prof. Borja Cocostegui MD, "North Carolina macular dystrophy" 6/05

Cape Town, South Africa, The Retina Society "Avastin (bevazizumab) for wet AMD, 6 month data" 10/06

San Diego, CA, The Retina Society, " New mutations in enhanced s-cone / Goldmann-Favre" 10/05

London, UK The Macula Society, "Combination therapy (anti-VEGFs with PDT) for wet AMD" 6/07

Oahu, Hawaii, PCOOS Pacific Coast Ophthalmology and Oto-Laryngology Society. "Combination therapy (anti-VEGFs with PDT) for wet AMD" 7/07

Marbella, Spain, 7th International Age-Related Macular Degeneration (AMD) Congress, , "Combination therapy (anti-VEGFs with PDT) for wet AMD" 10/07

Indian Wells, CA, The Macula Society, "Combination therapy (anti-VEGFs with PDT) for wet AMD" 12/07

Hue, Viet Nam. ICEM (2nd Imperial City Eye Meeting) 2008. Macular Degeneration, examination techniques 6/08

Hue, Viet Nam. ICEM (Imperial City Eye Meeting) 2008. Macular Degeneration, special testing 6/08

Hue, Viet Nam. ICEM (Imperial City Eye Meeting) 2008. Macular Degeneration, medical treatments 6/08

Hue, Viet Nam. ICEM (Imperial City Eye Meeting) 2008. Macular Degeneration, surgical treatments 6/08

Nuku' Olafa, Tonga: Diabetic Retinopathy update 9/09

Kona, Hawaii, Hawaiian Eye Meeting, 2013 Presentation of fungal endophthalmitis outbreak following intravitreal injections of triamcinolone contaminated by a compounding pharmacy

Dana Point, CA, Macula Society, 2013, Presentation of fungal endophthalmitis outbreak following intravitreal injections of triamcinolone contaminated by a compounding pharmacy

La Jolla, CA, American Ophthalmological Society, 2013, Presentation of fungal endophthalmitis outbreak following intravitreal injections of triamcinolone contaminated by a compounding pharmacy

Toronto, Canada, American Society of Retinal Specialist, 2013, Presentation of fungal endophthalmitis outbreak following intravitreal injections of triamcinolone contaminated by a compounding pharmacy

Vienna, Austria. American Society of Retina Specialists. Aspirin use in age-related macular degeneration. 7/2015

Philadelphia, PA, Temple University Alumni Day: Invited Grand Rounds Speaker: Dealing with an outbreak of fungal endophthalmitis due to contaminated triamcinolone from a compounding pharmacy.

Philadelphia, PA: Temple University: Invited speaker for resident's day: North Carolina Macular Dystrophy: gene found!

MEMBERSHIP IN SCIENTIFIC SOCIETIES:

American Medical Informatics Association 12/03 - present
The American Academy of Ophthalmology: 1985 - present.
American Medical Association: 1985 - present
The Association for Research in Vision and Ophthalmology (ARVO): 1985 - present.
American Society of Human Genetics: 1988 - present.
American Association for the Advancement of Science: 1988 -present.
Ophthalmic Genetics Study Club: 1990 - present.
Christian Ophthalmological Society: 1990 - present.
The Macula Society: 1994- present
The Los Angeles County Medical Society: 1994- present
California Ophthalmological Association: 1994-present
California Medical Association, 1995 - present
Western Retina Study Club, 1996 - present
The Retina Society, 1996 - present
Internet Ophthalmology Society, 1995-present
The American Ophthalmological Society Associate (AOS) 1998 – present www.aosonline.org
The American College of Physician Executives 1997 – present
Pan-American Association of Ophthalmology 1999 – present
National Association for the Visually Handicapped (NAVH http://www.navh.org), member Medical Advisory Board, 2000- present
Member program committee for The American Ophthalmological Society (AOS) 2003-2006

BOARD CERTIFICATION: American Board of Ophthalmology, 1989

LICENSURE INFORMATION:

Louisiana Board of Medical Examiners, license #16124 6/18/81 – 10/04
North Carolina Board of Medical Examiners License, license #28088 6/12/84 - present
South Carolina Board of Medical Examiners license #15175 10/5/90 - present
Massachusetts Board of Registration in Medicine license #75230 12/91-12/92
Maryland Board of Medical Examiners license #D42715 12/91 - 12/93
Florida, Department of Professional Regulation, license # ME 0065156 10/93-present
California Board of Medical Examiners license # A053173 issued 6/8/94- 10/31/05
Tennessee Board of Medical Examiners license # 30080 issued 4/98-4/99
Mississippi State Board of Medical Examiners License # 16564 issued 10/4/99

HONORS:

Top Doctors in Southern California: 2009 (LA Times)
Outstanding Scientist of the 21st Century, elected 11/14/2001
Life member of Kingston's National Registry of Who's Who in the 2002 edition:

American Ophthalmological Society, elected full member 6/2001
American Ophthalmological Society, elected associate 6/98
Honor Award from the American Academy of Ophthalmology, received 1998
National Institutes of Health, National Eye Institute Visual Science C Study Section, temporary member. 10/97
National Institutes of Health, National Eye Institute Visual Science B Study Section, Ad Hoc member. 8/97
Who's Who in Diabetes Treatment, Education, and Research named 1996
Best Doctors in Los Angeles. named by the Los Angeles Magazine, 1996
American Men and Women of Science: named 1997
Best Doctors in America, named in 1996 - present
Best Doctors in America, Pacific Region. named in 1996
Attending of the Year, 1992-93, elected by the residents, University of Florida
Jacob Javits Fellow 12/88 - 11/90
Diplomat, American Board of Ophthalmology: 11/14/1989 - present.
The First Chief Resident in Ophthalmology at the Duke Eye Center 5/87 - 12/87
Cabarrus Surgical Fellowship - 1984 - 1985
Alpha Omega Alpha Honor Medical Society: initiated 1980 - present.
American Cancer Association Research Externship 1979
Alpha Omega Alpha: Elected 1980 Tulane University Medical School
Phi Eta Sigma Honor Society - elected 1976 L.S.U.
Phi Kappa Phi Honor Society - elected 1976 L.S.U.
Alpha Epsilon Delta Honor Society - elected 1976 L.S.U.
Dean's List L.S.U. - all seven semesters, 8/74 - 8/77

ADDITIONAL INFORMATION: Include pertinent information concerning your educational and scientific background, and activities that do not fit into the categories previously listed.

Consultations Outside the University (U.F. and UCLA):
Retina consultation at Veterans Administration Hospital, Gainesville, FL: 1992 - 94.
Retina consultation at North Florida Eye Center in Lake City, FL. July 1993 - 94
Retina consultations at The Valley Eye Center in Van Nuys, CA 4/95-5/97
Retina consultations at Wadsworth Veterans Administration Hospital, Los Angeles, CA 6/95- present
Agouron Pharmaceuticals, LaJolla, CA, Scientific Advisory Board - member, 1998-2000
DeCode, Reykjavik, Iceland, Scientific Advisor, 1998

Editor of Scholarly Journal, Service on an Editorial Advisory Board, or Reviewer of a Scholarly Journal:
Editorial Board: Molecular Vision 2000-present
Guest Editor: Investigative Ophthalmology and Visual Sciences 2001
Guest Editor: Investigative Ophthalmology and Visual Sciences 1999
Editorial Board: Ophthalmic Genetics 1995-2000
Archives of Ophthalmology: Reviewer 1989 – present
Investigative Ophthalmology: Reviewer 1989 - present
Ophthalmology: Reviewer 1991 – present
Canadian Research Council: Grant reviewer - 1991 present; reviews one grant per year.
Canadian Retinitis Pigmentosa Foundation: Grant reviewer - 1991- present; reviews one grant per year.
Foundation Fighting Blindness, grant reviewer, 1994-present
Ophthalmic Genetics: Reviewer - 1992 - present
Retina: 1995-present
ARVO moderator 1994, Molecular genetics
ARVO moderator 1995, Molecular genetics
ARVO moderator 1996, Molecular genetics
American Journal of Ophthalmology 95-present
Genomics, 1996-present
Retina, 1997-present
Human Molecular Genetics 1998-present

American Journal of Human Genetics 1998-present
Molecular Vision, 1998 – present
Editorial Board member Molecular Vision 2000-present
Journal of the American Association of Pediatric Ophthalmology and Strabismus: reviewer 2000- present
Welcome Foundation: grant reviewer 2001- present
Cornea: 2000 – present
Health Psychology: 2004

International Activities:
Activity: moderator at ARVO (Association for research in vision and ophthalmology), June 1994,
Service: moderator: retinal genetics section

Activity: moderator at ARVO (Association for research in vision and ophthalmology), June 1995,
Service: moderator: retinal genetics section

Activity: moderator at ARVO (Association for research in vision and ophthalmology), June 1996,
Service: moderator: retinal genetics section

Activity: Studied families in Belize, Central America with inherited retinal degenerations.
Service: Performed eye examinations on indigent population in Belize, Central America.
Research: Ascertaining families with rare inherited eye diseases and performed molecular genetic studies.

Activity: Participation at the First International Workshop on Human Chromosome 6
Research: Develop genetics linkage map of chromosome 6 for HUGO (Human Genome Organization).

Activity: Studied families in northern France with inherited macular degeneration in collaboration with Professor Bernard Puech in Lille, France.
Service: Performed eye examinations on families
Research: Ascertained families with rare inherited eye diseases and performed molecular genetics studies

Activity: Representative of the Jules Stein Eye Institute and the Paul Kaiser Foundation of the Pan-American Ophthalmological Association to Mexico City, May 13-18, 1996.
Service: examined indigent patients with retinal diseases at The Hospital Evitar La Ciegeuria in Mexico City
Research: Set up collaborations for studying inherited ocular diseases in Mexican families.

Activity: WAVE (Western Association of Vitreo-retinal Education), Maui, Hawaii WAVE (Western Association of Vitreo-retinal Education). Moderator Wide Angle Viewing Session, Maui, Hawaii. 7/96

Activity: Established the international consortium to map and clone macular degeneration genes. 1997

Activity For Last 10 Years:

	1999-94	1994-1993	1993-1992	1991-1990	1990-1989	1988-1989
Teaching	5 %	10%	10%	5%	5%	10%
Research	10%	10%	10%	75%	95%	10%
Service	85%	80%	80%	20%	0%	0%
Extension	0%	0%	0%	0%	0%	80%
	100%	100%	100%	100%	100%	100%

Teaching, Advising, and/or Instructional Accomplishments (UF):

Dr. Small actively participates in the advising of residents for post-graduate training and practice of ophthalmology and retinal/vitreous diseases. 1990 - Present

Dr. Small has participated in the teaching of a laser photocoagulation course (Skills Transfer Course) at the American Academy of Ophthalmology for the past 3 years as well. 1989 - 1992

1. Lecture to Residents - Monthly One Hour Lectures:

1991	Jan:	General Genetics 1
	Feb.	Ocular Genetics 1
	Mar:	Ocular Genetics 2
	April:	Electro-retinography 1
	May	Electro- retinography 2
	June	Inherited Retinal Degenerations 1
	July	Inherited Retinal Degenerations 2
	August:	Inherited macular degeneration
	Sept.:	Uveitis 1
	Oct.:	Uveitis 2
1992:	Aug.:	Retinal Detachment 1
	Sept.:	Retinal Detachment 2
	Oct.:	Diabetic Retinopathy 1
	Nov.:	Diabetic Retinopathy 2
	Dec.:	Diabetic Retinopathy 3
1993:	Jan.:	Age-Related Macular Degeneration 1
	Feb.:	Age-Related Macular Degeneration 2
	Mar:	Inherited Retinal Degenerations
	April:	Electro-retinopathy
	May:	Uveitis 1
	June:	Uveitis 2
	Aug.:	Retinal Detachment 1
	Sept.:	Retinal Detachment 2
	Oct.:	Diabetic Retinopathy 1
	Nov.:	Diabetic Retinopathy 2
	Dec.:	Diabetic Retinopathy 3
1994:	Jan.:	Age-Related Macular Degeneration 1
	Feb.:	Age-Related Macular Degeneration 2
	Mar:	Inherited Retinal Degenerations
	April:	Electro-retinopathy
	May:	Uveitis 1

2. Lectures to Medical Students: 1992-present on:
   - Central Retinal Artery Occlusion
   - Hypertensive Retinopathy
   - Diabetic Retinopathy
   - Ocular Sarcoidosis
   - AIDS - Retinopathy
   - Macular Degeneration
   - Workshop on Molecular Genetics
3. Clinical teaching during retina clinic to 1st, 2nd, and 3rd year residents - every Monday, 8:00 am - 7:00 PM.
4. Clinical teaching during VA Retina Clinic to 1st, 2nd, and 3rd year residents - every Wednesday, 2:00 PM - 4:00 PM.
5. Surgical teaching to 2nd year residents 3-4 times per week and once every other week at the VA.
6. Lecture seminar to Center for Mammalian Genetics: "Hereditary Macular Degeneration" - 1992-93.

Teaching Evaluation

RESIDENT'S TEACHING EVALUATIONS (UF)
(18 Residents/Year)
Kent Wilson Small

	1992-93	1993-94
	Dr. Small Dept. Avg.	Dr. Small/ Dept. Avg
Didactic Teaching: lectures & conferences		
Punctuality & dependability	1.3/1.9	1.8/2.2
Teaching Skills	1.5/1.7	1.3/2.0
Provides useful clinical information	1.4/1.8	1.5/1.9
Gives sufficient lectures	1.5/2.1	1.3/1.9
Encourages questions/comments	1.5/2.1	1.7/2.1
Clinical Duties		
Time spent teaching (in clinic)	1.7/2.2	2.1/2.3
Clinic run in orderly manner	2.3/2.3	1.2/1.8
Approachable for questions	1.2/2.0	1.3/2.0
Establishes patient rapport	1.6/1.9	1.3/1.9
Treats clinic staff respectfully	1.4/2.0	1.2/2.2
Avoids outbursts of anger	1.2/2.0	1.2/1.9
Allows resident participation	1.5/2.0	1.6/2.1
Fully available for call	1.3/2.0	1.3/2.0
Administrative/Research		
Active in own research	1.2/1.8	1.0/2.2
Departmental duties	1.9/1.9	1.5/2.3
Active in political issues	2.0/1.9	1.6/2.2
Enhances Dept.	1.2/2.1	

Rating Scale:(1) Outstanding; (2) Above Average; (3) Average; (4) Unsatisfactory; (5) Totally Inadequate
No comparable college mean available This was required

Teaching Evaluation

RESIDENT'S TEACHING EVALUATIONS (UCLA)
(21 Residents/Year) score =1 is poor, 10.0= best; note this is opposite scoring from UF
Dr. Small's score/dept. average
Kent Wilson Small

	94-95	95-96	96-97	97-98	98-99
Has command of the subject	7.0/6.5	6.3/6.3	7.0/6.6	6.8/6.4	6.5/6.6
presents material in organized, clear manner	7.0/6.3	6.0/6.2	6.7/6.5	6.8/6.3	6.5/6.0
Sensitive to response of students, house-staff	7.0/5.9	6.4/5.9	6.5/6.4	6.7/5.9	6.1/5.8
Available to and friendly towards house-staff	7.0/6.1	6.7/6.0	6.7/6.3	6.8/5.9	6.4/5.9
Enjoys teaching and is enthusiastic about the subject	7.0/6.2	6.5/6.1	6.5/6.5	6.8/6.1	6.4/6.0
deeply interested in patient care; often makes contributions	6.5/6.1	6.4/6.1	7.0/6.6	6.7/6.1	6.5/6.1
Meets appointments; punctual	7.0/6.1	5.3/6.0	7.0/6.3	6.5/6.1	6.0/6.0
How does the instructor compare with other clinical teachers you have had at UCLA	6.5/6.1	6.1/5.9	6.7/6.5	6.8/6.0	6.1/5.8

2nd Year Medical Student Evaluations
Teaching Evaluations (Approximately 100 Students)
(Evaluations based 3 Lectures)

	1993
Department Average	1.84
Dr. Kent Small	1.80

Note: The Department of Ophthalmology concentrates its formal teaching of medical students into two weeks. The evaluation of our faculty occurs only for that lecture series. Their evaluations of Dr. Small's lectures are shown above.

Teaching, Advising, and/or Instructional Accomplishments (at UCLA):
Jules Stein Eye Institute, Clinical Teaching Conference, lecture: "vitrectomy for ocular toxocariasis" 1 hour, 11/94
Jules Stein Eye Institute, Vision Genetic Meeting, lecture: "molecular genetics of macular degeneration" 1 hour, 11/94
Jules Stein Eye Institute, Clinical Science Series Seminar, administered the JSEI weekly quiz, 10/94
Jules Stein Eye Institute, 1 hour discussion of retinal research activity with residents, 12/94
Jules Stein Eye Institute, CMV retinopathy study group, presentation: "use of intravitreal cidofovir" 1 hour lecture, 1/95
Jules Stein Eye Institute, Vision Genetics Meeting, lecture: molecular genetics of autosomal dominant cone degeneration maps to chromosome 17p" 1 hour lecture, 1/95
Jules Stein Eye Institute, Retinitis Pigmentosa Study Group, presentation "positional cloning of the North Carolina macular dystrophy gene" 1 hour lecture, 4/95
Jules Stein Eye Institute, Clinical Seminars Series, Moderator of Grand Rounds. 4/95
Jules Stein Eye Institute, Basic Science Series Seminar, "selective hybridization cloning of the North Carolina macular dystrophy gene" 1 hour lecture, 6/95
Jules Stein Eye Institute, Clinical Science Series Seminar, Moderator of Grand Rounds, 6/95
Jules Stein Eye Institute, Clinical Science Series Seminar, Moderator of Grand Rounds, 9/95
Jules Stein Eye Institute, Clinical Science Series Seminar, administered the JSEI weekly quiz, 10/95 Jules Stein Eye Institute, Clinical Science Series Seminar, administered the JSEI weekly quiz, 10/96
Jules Stein Eye Institute, Clinical Science Series Seminar, Moderator of Grand Rounds, 11/9 Jules Stein Eye Institute, Clinical Science Series Seminar, administered the JSEI weekly quiz, 10/97
Jules Stein Eye Institute, Clinical Science Series Seminar, Moderator of Grand Rounds, 11/97
Jules Stein Eye Institute, Clinical Science Series Seminar, administered the JSEI weekly quiz, 10/98
Jules Stein Eye Institute, Clinical Science Series Seminar, Moderator of Grand Rounds, 11/98
Jules Stein Eye Institute, Clinical Science Series Seminar, administered the JSEI weekly quiz, 10/99
Jules Stein Eye Institute, Clinical Science Series Seminar, Moderator of Grand Rounds, 11/99
Jules Stein Eye Institute, Clinical Science Series Seminar, administered the JSEI weekly quiz, 10/00
Jules Stein Eye Institute, Clinical Science Series Seminar, Moderator of Grand Rounds, 11/00
Jules Stein Eye Institute, Basic Science Series Seminar, "Ocular Genetics" 10/01
Jules Stein Eye Institute, "The genetics of myopia" Myopia for the Year 2000: clinical and research perspectives, 2/96
Jules Stein Eye Institute, Clinical Fundamentals Course M201 for Medial Students - Instructor 3 hours, 9/94
Jules Stein Eye Institute, Clinical Fundamentals Course M201 for Medial Students - Instructor 3 hours, 2/95
Jules Stein Eye Institute, Clinical Fundamentals Course M201 for Medial Students - Instructor 3 hours, 9/95
Jules Stein Eye Institute, Clinical Fundamentals Course M201 for Medial Students - Instructor 3 hours, 2/96
Jules Stein Eye Institute, Clinical Fundamentals Course M201 for Medial Students - Instructor 3 hours, 9/96
Jules Stein Eye Institute, Clinical Fundamentals Course M201 for Medial Students - Instructor 3 hours, 2/97
Jules Stein Eye Institute, Clinical Fundamentals Course M201 for Medial Students - Instructor 3 hours, 10/97
Jules Stein Eye Institute, Clinical Fundamentals Course M201 for Medial Students - Instructor 3 hours, 4/98
Jules Stein Eye Institute, Clinical Fundamentals Course M201 for Medial Students - Instructor 3 hours, 4/99
Jules Stein Eye Institute, Clinical Fundamentals Course M201 for Medial Students - Instructor 3 hours, 11/2000
Jules Stein Eye Institute, Clinical Fundamentals Course M201 for Medial Students - Instructor 3 hours, 11/2001
Jules Stein Eye Institute, Clinical Fundamentals Course M201 for Medial Students - Instructor 3 hours, 11/2002
"Hereditary Macular Disease" Jules Stein / UCLA Post Graduate Seminar, Los Angeles, CA , 3/96
"Laser to Drusen Trial" Jules Stein / UCLA Post Graduate Seminar, Los Angeles, CA, 3/96
"Silicon Retina / Retinal Transplants" Jules Stein / UCLA Post Graduate Seminar, Los Angeles, CA, 3/96
"Macular Hole Surgery" Jules Stein / UCLA Post Graduate Seminar, Los Angeles, CA, 3/96
"Phototoxicity" Jules Stein / UCLA Post Graduate Seminar, Los Angeles, CA, 3/96
Co-Moderator of The Jules Stein / UCLA Post Graduate Seminar, Los Angeles, CA , 3/96
"Complications of perfluorocarbon liquids" The New Age of Perfluoron Liquids Course by Infinitec, Los Angeles, CA, 5/96

"New Age of Perfluoron Liquids" Course by Infinitec, Co-Moderator, 5/96
"Hereditary retinal degenerations", Jules Stein Eye Institute Clinical Science Lectures Series: 3 hours, 10/96
"Macular degeneration", Jules Stein Eye Institute Clinical Science Lectures Series: 3 hours, 10/96
Initiated and maintained the macular degeneration support group at UCLA, 2/97- present
UCLA Intercampus Medical Genetics Training Program, Graduate Course in Genetics: 3 hours lecture: "review of ophthalmic genetics" 4/97
"Surgical macular diseases" Jules Stein / UCLA Post Graduate Seminar, Comprehensive Eye Care Update for the Practitioner, Los Angeles, CA, 4/98

"The Aging Eye" Biology of Aging course (MCD Biology CM 149/Pathology M262) 4/98 2 hour lecture for senior undergraduates
"Hereditary retinal degenerations", Jules Stein Eye Institute Clinical Science Lectures Series: 4/99
"What's new in age-related macular degeneration" Jules Stein Eye Institute Clinical Science Lectures Series: 1/2001
Moderator: Jules Stein Eye Institute Clinical Science Lectures Series: 1/2001
"Update on macular degeneration" Jules Stein Eye Institute Clinical Science Lectures Series: 1/2001
"Age-related macular degeneration: update" 10/2002 Invited Speaker Braille Institute, Los Angeles, CA 3/2001
"Update on macular degeneration" Jules Stein Eye Institute Clinical Science Lectures Series: 10/2002
Moderator: Jules Stein Eye Institute Clinical Science Lectures Series: 10/2002
"Finding the genes of AMD" Jules Stein / UCLA Post Graduate Seminar, Los Angeles, CA , 4/2003
"Cone-Rod Dystrophies" Jules Stein / UCLA Post Graduate Seminar, Los Angeles, CA , 34/2003
"AREDS, new findings" Jules Stein / UCLA Post Graduate Seminar, Los Angeles, CA , 4/2003
Moderator: Jules Stein / UCLA Post Graduate Seminar, Los Angeles, CA , 4/2003

Teaching Accomplishments in Laboratory Molecular Genetics

name	year	status
Kay Kelley, Ph.D.,	1992-1994	Biological Scientist UF
Anthony Sanchez, M.S.	1992-1994	Biological Scientist UF
Svetlana Yelchits, Ph.D.	1992-1997	Biological Scientist UF and UCLA
Lynne Mullen, BS	1992-1997	Biological Scientist UF and UCLA
Lacretia Fisher, BS	1992-1994	Medical Student UF
Mike Stalvey, BS	1992-1994	Graduate Student UF
Nitin Udar, Ph.D.,	1996-1997	UCLA post-doc
James Fink, BS	1995	UCLA medical student
Shiang Do	1995-96	UCLA undergraduate
Christine Chin	1996	UCLA undergraduate
Pamela Golchet	1996-97	UCLA undergraduate
Steven Mellul, BS	1996	Hanneman University medical student
Sara Gislison	1998	UCLA undergraduate
Gisitzman Medical Student Geriatric Scholars Program: took one student each year in the lab 1995-1999		
Nancy Padilla	1998-2000	UCLA undergraduate
Jessica Felipe	1998-2000	UCLA undergraduate
Tara Anderson	1998-2001	UCLA undergraduate
Allen Shirvanian	1998-2000	UCLA undergraduate
Liezel Morales	1998-present	UCLA undergraduate
Pranav Vyas	1998-2000	UCLA post-doc
Vivek Yellore	2000-present	UCLA post-doc

Graduate Faculty Status:
Department of Biochemistry and Molecular Biochemistry and Molecular Biology, University of Florida - Affiliate member: 1992 - 94.
Faculty of School of Graduate Studies, Medical University of South Carolina, Charleston: 1991.

Graduate Committee Activities:
Member, School of Graduate Education, Medical University of South Carolina, Charleston ,SC 1991
Assistant Director of M.D./ Ph.D. program, University of Florida: 1993 - 94

Courses Attended (attended without giving a presentation)
Course in Mammalian Genetics, Bar Harbor, Maine, 1989
Genetic Linkage Analysis, Basic Course, Columbia University, by Jurg Ott, Ph.D. 1994
Leadership and Decision Making in Organizations, Anderson School of Business, UCLA, Los Angeles CA, 11/12-15/96

CPT Coding Course sponsored by The California Association of Ophthalmology 1995
CPT Coding Course sponsored by The California Association of Ophthalmology 1996
CPT Coding Course sponsored by The California Association of Ophthalmology 1997
UCLA Teaching Physician Regulations for Medicare and Medicaid compliance, 1997
UCLA CPT coding course, 1997
UCLA Anderson School of Business and Management 295A: New Venture Initiation, audited and completed, 1997

UCLA Anderson School of Business and Management 298D15: Business Plan Development and Writing, audited and completed 1997
The AUPO (Association of University Professors of Ophthalmology) Management Program, 1997
Advanced Genetic Linkage Analysis Course of Complex Traits, Rockefeller University, by Jurg Ott, Ph.D. 1997
UCLA Anderson School of Business and Management: Executive business course in finances 6/98
S.A.G.E. Course in genetic segregation analysis and non-parametric genetic studies. Given by Robert Elston. Costa Mesa, CA 1999

Summit Technology, Photorefractive Keratectomy and Toric PRK. 11/14/99

Summit Technology, Microkeratome Training. 11/14/99

Summit Technology, Photorefractive Keratectomy and Toric PRK. 11/14/99

Summit Technology, Photorefractive Keratectomy . 11/14/99

Ophthalmology Coding, sponsored by the California Ophthalmology Association, 6/2000
Ophthalmology Coding, sponsored by the California Ophthalmology Association, 6/1998
Ophthalmology Coding, sponsored by the California Ophthalmology Association, 6/1997
Ophthalmology Coding, sponsored by the California Ophthalmology Association, 6/1996
Ophthalmology Coding, sponsored by the California Ophthalmology Association, 1/2003
Publicity / Public Interviews
1992:
Associated Press release of mapping macular degeneration gene, published in many newspapers across the country.
Science News, The weekly Newsmagazine of Science, (International publication) July 18, 1992, vol. 142, pg. 37. Featured section on Dr. Small's mapping of the macular degeneration gene.

1993:
Gainesville Sun announcing Dr. Small's Jules and Doris Stein Research to Prevent Blindness Professorship.
WTVD TV, channel 11, "Health Spot" interviewed Dr. Small on genetics of macular degeneration.
Friday Evening Post (University of Florida paper) announcing the Jules and Doris Stein RPB Award.
The Tulane Medicine, New Orleans, LA announcing Dr. Small's mapping of the macular degeneration gene
Duke University Eye Center Alumni 1992 Report; one page feature of Dr. Small's mapping of the macular degeneration gene
The University of Florida Health Science Center Annual Report 1992-93. Featured article on Dr. Small and The Center for Mammalian Genetics.
Visions, News and Views from the College of Medicine featured section on Dr. Small and his research. May 1993
The Research to Prevent Blindness Progress Report 1993 section on Dr. Small and his research.
The Research to Prevent Blindness Annual Report 1992, Featured section on Dr. Small and his research. pg. 10.
The Florida Physicain Fall 1993, pg. 15, section on Dr. Small and his research

Associated Press release: Nov. 22, 1993, TV appearance describing research of locating the macular degeneration gene- this release was aired by many local and national news stations across the country (judging by the phone calls received) CNBC national cable news network "Medical Beat", WTVD 11 Gainesville and Ocala are some examples,
The Friday Evening Post Nov. 19, 1993, announcing the NIH grant (RO-1) $1,000,000 which Dr. Small is PI on
The Gainesville Sun, Nov. 16, 1993, full page featured article on Dr. Small and his research
1994
KCAL radio station, San Francisco, CA., interview, Macular Degeneration
1995
Ophthalmology World News, The Independent Newspaper of the American Academy of Ophthalmology. volume 1, number 12, 1995, pg. 14-16 "The Genetics Key to AMD"
1996
KCAL-9 TV station 1/19/96. Macular Degeneration interview
"Researchers find an easy switch from manual to automated genetics analysis" Biosystems Reporter volume 1, 1996.

Asheville Citizen Times, Aug. 1996. "Researcher works with family with rare eye disorder".
Los Angeles Magazine, Kent Small, M.D. named as one of LA's best doctors Nov. 1996
1997
Senior World Newsmagazine vol. 11, May 1997, page 30. "Preventive care critical for aging eyes"
1999
"Genetic Eye Disease: Will you be prepared for the future?" in Ophthalmology Management April, 1999, pp105-108,

MDForum: Internet Macular degeneration forum 10/1999 http://members.aol.com/danlrob/MDpeople/clinic.html
DNA.com internet chat interview "The genetics and treatment of degenerative retinal diseases" 8/17/200
www.dna.com/eventTranscript
KNBC: television interview about Stargardt disease and the American Olympic runner Marla Runyon. 9/29/2000

OCT (Orange County Times TV) Age-related macular degeneration and genetics of macular diseases 3/2001

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Publications:

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2. Ellenby MI, Small KW, Wells RM, Hoyt DJ, Lowe JE: Changes in myocardial resistivity during global ischemia: On-line identification of the onset of severe but reversible ischemic injury. Surgical Forum 36:1985,198-200.
3. Damiano RJ Jr, Smith PK, Tripp HF Jr. Asano T, Small KW, Lowe JE, Ideker RE, Cox JL: The effect of chemical ablation of the endocardium on ventricular fibrillation threshold. Circulation 74:1986,645-652.
4. Damiano RJ Jr, Tripp HF, Asano T, Small KW, Jones RH, Lowe JE: Left ventricular dysfunction and dilation resulting from chronic supraventricular tachycardia. Journal Thoracic and Cardiovascular Surgery 94:1987,135-143.
5. Small KW, Stefansson E, Hatchell DL: Retinal blood flow in normal and diabetic dogs. Investigative Ophthalmology and Visual Science 28:1987,672-675.
6. Small KW, Williams Sr. J, McCuen II BW, deJuan Jr E, Machemer RM: Vitrectomy in ocular toxocariasis. American College of Surgeons, Surgical Forum 38:1987,526-528.
7. Ellenby MI, Small KW, Wells RM, Hoyt DJ, Lowe JE: On-line identification of the reversible ischemic injury by measurement of myocardial electrical impedance. Annals of Thoracic Surgery 44:1987,587-597.
8. Pericak-Vance MA, Yamaoka LH, Vance JM, Small KW, Rosenwasser GOD, Hung WY, Alberts MJ, Hayes CS, Speer MC, Gilbert JR, Herbstreith M, Aylsworth A, Roses AD: Genetics linkage studies of chromosomes 17 RFLPs in Von Recklinghausen neurofibromatosis. Genomics 1:1987,349-352.

9. Small KW, Pollock SC, Scheinman J: Optic atrophy in primary oxalosis. American Journal of Ophthalmology 106:1988,96-97.
10. Small KW, Buckley EG: Recurrent ptosis secondary to a pituitary tumor. American Journal of Ophthalmology 106:1988,760-761.
11. Small KW, Buckley EG: Can cardiac surgery provoke vitreous hemorrhage in proliferative diabetic retinopathy? Surgical Forum 39:1988,517-519.
12. Alexander III E, Rossitch Jr E, Small K, Abson P: Merkel cell tumor metastatic to brain and choroid: Evidence for hematogenous spread and the implications for therapy. Clinical Neurology and Neurosurgery 91:1989,307-310.
13. Vance JM, Pericak-Vance MA, Yamaoka LH, Speer MC, Rosenwasser GOD, Small KW, Gaskel PC, Hung WY, Alberts MJ, Hayes CS, Gilbert JR, Aylsworth A, Roses AD: Genetics linkage mapping of chromosome 17 markers and peripheral neurofibromatosis (NF1). American Journal of Human Genetics 44:1989,25-29.
14. Small KW, Stefansson E, Hatchell DL: Coronary blood flow in insulin dependent diabetic dogs. Acta Diabetol. Lat. 26:1989,275-278.
15. Small KW, McCuen II BW, de Juan III E, Machemer R: The surgical management of retinal traction caused by toxocariasis. American Journal of Ophthalmology 108:1989,10-14.
16. Small KW: North Carolina macular dystrophy: revisited. Ophthalmology 96:1989,1747-1754.
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18. Small KW, Rosenwasser GO, Alexander III E, Dutton JJ: Presumed choroidal metastasis of Merkel cell carcinoma. Annals of Ophthalmology 22:1990,187-190.
19. Small KW, Letson R, Scheinman J: Ocular findings in primary hyperoxaluria. Archives of Ophthalmology 108:1990,89-93.
20. Vance JM, Small KW, Jones MA, Stajich JM, Yamaoka LH, Roses AD, Hung W, Pericak-Vance MA: Confirmation of linkage in Von Hippel-Lindau Disease. Genomics 6:1990,565-567.
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22. Small KW, Killian J, McLean W: North Carolina's dominant progressive foveal dystrophy. How progressive is it? British Journal of Ophthalmology 75:1991,401-406.
23. Small KW, Anderson WB: Pigmented paravenous retinochoroidal atrophy discordant expression in monozygotic twins. Archives of Ophthalmology 109:1991,1408-1410.
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78. Parwar B, Small KW. Silicone oil through an Ahmed valve. Retina 22: 657-658, 2002
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MISC INFO.

Current Medicaid #: GR0058400 / California
UPIN # C86490
Current Malpractice Insurance: Medical Protective $2,000,000 / 4,000,000
Medicare California #: 18491A
DEA # BS 2498548

Other Activities:
Blacknall Memorial Presbyterian Church, Durham,NC: member 7/82-10/95
Bel Air Presbyterian Church, Los Angeles, CA: member 10/95-present
Steering Committee member of Christian Family Fellowship Sunday School Class at Bel Air Presbyterian Church: 10/95-present

Deacon: Bel Air Presbyterian Church, Los Angeles, CA: 1/09- 12/12

UCLA Master's Swim Team: member May 1996-present
Medaled in 200 m butterfly in Nationals Meet 2009 Fresno, CA

Extra in feature film "Naturally Native", a film about modern Native American issues
Volunteer: Los Angeles Mission soup kitchen, 1998-present
Volunteer Physician: Rotary Humanitarian Projects Sponsored Eye Clinic in Denpensar, Indonesia
8/3-8/2001
Steering Committee member of Youth Ministry at Bel Air Presbyterian Church: 10/01-present

Medical Mission trip to Port Villa, Vanuatu through S.E.E. International to study families with inherited eye disease and perform surgery 8/2002

Medical Mission trip: Volunteer Ophthalmologist / Retinal Surgeon, Nuku' Olafa, Tonga and Va'Vau, Tonga, 9 09, with Surgical Eye Expedition and The Hawaiian Eye Foundation

Medical Service: Volunteer Ophthalmologist, Rescati Family Rescue Mission, San Fernando Valley, CA 2009-2012