Incontinentia Pigmenti (IP) Conference: Translating Discovery to Therapy

February 20-22, 2025

nfed

NATIONAL FOUNDATION FOR ECTODERMAL DYSPLASIAS

Sponsored By: OHSU

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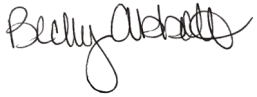
WELCOME!

Dear Colleague,

The conference will bring together clinicians, researchers, and young investigators to present and discuss current approaches and future directions for translating science to therapy for incontinentia pigmenti (IP). Our goal is to advance our ability to diagnose and manage this rare disease through the development of screening guidelines, effective diagnostics, and novel therapies.

Mary Fete, MSN, RN, CCM
Conference Co-Chair
Executive Director
National Foundation for Ectodermal Dysplasias



Becky Abbott, MPH
Conference Co-Chair
Director, Treatment and Research Advocacy
National Foundation for Ectodermal Dysplasias

J. Peter Campbell, MD, MPH
Conference Co-Chair
Professor of Ophthalmology, Casey Eye Institute, Oregon Health & Science University

Connie Chen, MD
Conference Co-Chair
Vitreoretinal specialist, Seattle, WA

Ian Han, MD
Conference Co-Chair
Director of the Retina Service and Associate Professor, Department of Ophthalmology and Visual Sciences, University of Iowa

ABOUT THE NFED

The National Foundation for Ectodermal Dysplasias (NFED) is the worldwide expert on ectodermal dysplasias and the only advocacy organization in the United States dedicated to those living with these disorders.

EDUCATION

We offer a range of educational resources and events to meet the changing needs of those affected by the different types of ectodermal dysplasias. And, by connecting you with other families like yours, we give you the opportunity to learn from one another.

SUPPORT

We provide lifelong support for individuals and families affected by ectodermal dysplasias.

RESEARCH

We encourage and support medical, dental, genetic and other ectodermal dysplasia–related research to improve the health and quality of life of everyone living with ectodermal dysplasia. Our ultimate goal is to find a cure.

AGENDA

THURSDAY, FEBRUARY 20

7:00 PM	**IP Patients and Families - Panel on the Patient Experience** - Mary Fete, Susanne Emmerich, Jacques Monnet (virtual), Becky Abbott
7:30 - 9:00 PM	**Welcome Dinner**

FRIDAY, FEBRUARY 21

7:00 AM	**Breakfast**
8:00 AM	**Welcome to Participants** - Mary Fete
8:05 AM	**Welcome, Goals and Expected Outcomes** - Peter Campbell
8:10 AM	**The genetic basis and inheritance of incontinentia pigmenti** - Kathy Grange
8:20 AM	**Ectopic cell senescence, defective epidermal stratification, and the protein PRMT5** - Eric Van Otterloo
8:30 AM	**Oral manifestations of IP** - Tim Wright
8:40 AM	**Q&A / Discussion**

AGENDA

FRIDAY, FEBRUARY 21 - CONTINUED

Translational Research / Neurology & Miscellaneous

8:50 AM	**Incontinentia pigmenti underlies thymic dysplasia, autoantibodies to type I IFNs, and viral diseases** - Jeremie Rosain (virtual)
9:00 AM	**Incontinentia Pigmenti - Translational Research: Neurology** - Markus Schwaniger (virtual)
9:10 AM	**Microglia-Endothelial Cell Interactions in an Incontinentia Pigmenti Mouse Model** - Phillip Ehrich
9:20 AM	**Can RIP kinase inhibition reduce the cerebral vascular phenotype of Incontinentia Pigmenti?** - Teresa Faupel
9:30 AM	**Q&A / Discussion / Open issues**
9:40 AM	**Break**

Clinical Research / Ophthalmology

10:00 AM	**Keynote Address** - Morton Goldberg (virtual)
10:20 AM	**IP spectrum of disease and longitudinal findings** - Connie Chen
10:30 AM	**FA findings in IP** - Audina Berrocal
10:40 AM	**OCT/OCTA findings in IP** - Xi Chen
10:50 AM	**Q&A / Discussion**

Translation Research / Ophthalmology

11:00 AM	**Retinal angiogenesis** - M.E. Hartnett
11:20 AM	**NFKB/Nemo** - Eric Nudleman
11:30 AM	**Landscape of ocular translational research** - Ian Han
11:40 - 12:00 PM	**Q&A / Discussion**
12:00 - 12:30 PM	**Lunch**

AGENDA

FRIDAY, FEBRUARY 21 - CONTINUED

Clinical Practice Guidelines / Ophthalmology

12:30 PM	**Rationale for IP disease classification** - Sandra Montezuma
12:40 PM	**Evidence-based treatment guidelines (vis a vis classification)** - Philip Ferrone
12:50 PM	**PANEL DISCUSSION - CLASSIFICATION & TREATMENT** - ALL EYE MDs
1:30 - 1:50 PM	**Approaches to IP screening** - Peter Campbell
1:50 - 2:30 PM	**PANEL DISCUSSION / SCREENING** - ALL EYE MDs
2:30 - 2:50 PM	**Break**
2:50 - 4:00 PM	**Clinical Recommendations Working Groups**
4:00 - 4:30 PM	**Advocacy / Patient Experience** - Becky M. Abbott
4:30 - 5:30 PM	**Poster Presentations** - 5-7 minute presentations
5:30 - 6:30 PM	**Break / Evening Reception**
6:30 - 8:30 PM	**Dinner**

SATURDAY, FEBRUARY 22

7:00 AM	**Breakfast**
8:00 - 10:00 AM	**Action Items and Deliverables - Nomenclature, Screening Guidelines, Registry/Database, Multidisciplinary and Eye Management Recommendations**
10:00 - 11:00 AM	**Writing Committees Meet**
11:00 AM	**Wrap-up and Closing**

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NFED Family Member

OUR RESEARCH IMPACT

The National Foundation for Ectodermal Dysplasias (NFED) has been the driving force behind ectodermal dysplasias research for more than 30 years. Very little was known about the ectodermal dysplasias when we formed in 1981.

We knew from the beginning that only research was going to hold the answers to our families' questions and provide hope for possible cures. Like many rare disease organizations, the NFED stepped up to serve as a catalyst for research that is otherwise likely to be neglected by mainstream science.

$3.6M
Invested in Research

118
Invested In Different Research Studies & Trials

12
Research Conferences

1,000+
Participants from NFED Families

We have made enormous strides in the understanding and treatment of ectodermal dysplasias. The NFED's research impact spans decades, millions of dollars in funding, and our community of families around the world. We serve as a catalyst for research that is otherwise likely to be neglected by mainstream science.

Now, with more than 30 years of leadership, the NFED is driving the charge to develop effective treatments and—most importantly—cures.

GOALS

Short Term

- Establish research priorities aimed at accelerating incontinentia pigmenti research and therapeutic development.
- Develop and implement an incontinentia pigmenti ophthalmology referral network.
- Disseminate conference proceedings and ophthalmology protocols through publication.

Long Term

- Support and sustain long-term collaborations directed towards incontinentia pigmenti research and developing therapeutics.
- Develop effective protocols and therapeutics to address the ectodermal defects related to incontinentia pigmenti.

CONFERENCE CO-CHAIRS

Becky M. Abbott, MPH
Director, Research and Treatment Advocacy
National Foundation for Ectodermal Dysplasias

Becky Abbott is the Director of Treatment and Research Advocacy for the National Foundation of Ectodermal Dysplasias and Co-Chair of the NFED Family Advocacy Committee where she leads efforts to move the Ensuring Lasting Smiles Act (ELSA) through Congress. She received her undergraduate degree in biology from Mount Mary College in Milwaukee, WI and her Masters in Public Health at Benedictine University in Lisle, Illinois. She resides in Slinger, Wisconsin with her husband, Tom, and three sons: Lachlen, Ryder, and Aidan, who has been diagnosed with x-linked hypohidrotic ectodermal dysplasia (XLHED).

Mary Fete, MSN, RN, CCN
Executive Director
National Foundation for Ectodermal Dysplasias

Mary Fete is the Executive Director of the National Foundation for Ectodermal Dysplasias (NFED), a leading voice on ectodermal dysplasias and the only U.S.-based advocacy organization dedicated to individuals affected by these rare disorders.

With 23 years of experience in nonprofit leadership, program development, and fundraising, Mary is deeply committed to advancing patient advocacy, support, and groundbreaking research. Under her leadership, the NFED has expanded services, increased funding, and launched transformative initiatives that empower individuals and families.

Passionate about creating sustainable change, Mary collaborates with stakeholders, donors, and the community to ensure that those affected by ectodermal dysplasias receive the resources, care, and support they need to thrive.

CONFERENCE CO-CHAIRS

J. Peter Campbell, MD, MPH

Dr. Campbell is the Edwin and Josephine Knowles Professor of Ophthalmology at the Casey Eye Institute, Oregon Health & Science University. His clinical training and practice is focused on the care of adult and pediatric patients with vitreoretinal diseases. In addition, he is a translational clinician scientist primarily focused on two main research areas: the development of artificial intelligence (AI) algorithms in retinopathy of prematurity (ROP) with the Imaging and Informatics in ROP (i-ROP) research consortium, and optical coherence tomography (OCT) for pediatric retina with the Center for Ophthalmic Optics & Lasers [COOL Lab] headed by David Huang, MD at OHSU. He was the recipient of a Career Development Award from Research to Prevent Blindness, and has published more than 180 peer-reviewed articles, He is on the steering committee for the International Classification of Retinopathy of Prematurity, and is co-Chair for the Stop Infant Blindness in Africa committee, and is the Chair of the American Academy of Ophthalmology Committee on Artificial Intelligence.

Connie Chen, MD

Dr. Chen is a vitreoretinal specialist practicing in Seattle, Washington with a focus on adult vitreoretinal diseases and inherited retinal conditions. She received her medical degree from Weill Cornell Medical School. She is a graduate of the Johns Hopkins Ophthalmology residency program and former Stephen J. Ryan Assistant Chief of Service at Johns Hopkins Hospital. During her time at Johns Hopkins, she came to know many families with incontinentia pigmenti. In collaboration with colleagues Drs. Morton Goldberg and Ian Han, she helped characterize natural history and complications arising from incontinentia pigmenti-related retinopathy.

Ian Han, MD

Dr. Han is Director of the Retina Service and Associate Professor in the Department of Ophthalmology and Visual Sciences at the University of Iowa. He received his M.D. from the Duke University School of Medicine and completed ophthalmology residency and vitreoretinal surgery fellowship at the Wilmer Eye Institute at Johns Hopkins Hospital, where he also served as the Stephen J. Ryan, M.D. Assistant Chief of Service. As an active clinician-scientist, Dr. Han leads multidisciplinary teams in the Retinal Imaging and Modeling Lab and the Retinal Transplant Surgery Lab at the Institute for Vision Research at the University of Iowa.

PARTICIPANTS

Mary Elizabeth Hartnett, MD

Mary Elizabeth Hartnett, MD, is the Michael F. Marmor, M.D. Professor in Retinal Science and Diseases and is a Professor of Ophthalmology at Stanford University. Dr. Hartnett is the director of Pediatric Retina at Stanford University and principal investigator of a retinal angiogenesis laboratory, in which she studies causes and treatments for diseases including retinopathy of prematurity and age-related macular degeneration. She created the first-ever academic textbook on the subject, Pediatric Retina, in its third edition, which has proven to be an invaluable resource for residents and ophthalmologists internationally.

Sandra Montezuma, MD

Sandra Montezuma, M.D, is a Professor in Ophthalmology at the University of Minnesota (UMN). She earned her medical degree from the Universidad Nacional de Colombia. She did her first residency training in ophthalmology at the Fundacion Oftalmologica de Santander, Colombia in 2001. She was a fully trained ophthalmologist before coming to the United states in 2001. She completed 10 more years of training at Harvard Medical School. In 2001she joined the Boston Retinal Implant Project at Massachusetts Eye and Ear Infirmary/ Harvard Medical School, where she performed basic science research for nearly 4 years. After her Research fellowship, she completed a second surgical internship at Brigham and Women's hospital in 2005, a second residency in ophthalmology in 2008 and a Surgical Retina fellowship in 2010 at Massachusetts Eye and Ear infirmary/Harvard Medical School. She joined the UMN in 2010. She was awarded a Distinguished Teaching and Service Award at Massachusetts Eye and Ear infirmary/Harvard Medical School, she was honor the 2010 Gilligham Fellowship Award at The Pan-American Association of Ophthalmology, the 13th Fellowship Research Award at the Retina Society, the Distinguished Mentoring Award at the University of Minnesota Medical School in 2017 and The Knobloch Chair Professorship in 2018. She is the director of the retina service and electrophysiology service at the UMN. Her research interest includes translational research in Inherited Retinal Diseases (incontinentia Pigmenti; retinitis pigmentosa), retinal implants, retinal prosthesis, gene therap, Age-Related Macula Degeneration, ocular trauma and surgical retina. Being a part of discovering cures for blinding conditions is Dr. Montezuma's passion. She believes the ability to apply her clinical and surgical knowledge to help patients with all types of blinding conditions is truly a privilege. Dr. Montezuma finds interacting with patients and the ability to improve patients' vision the most rewarding aspect of her job. She appreciates the long-term relationships she has with her patients and being able to meet their needs with the highest quality of eye care available

PARTICIPANTS

Aliyah King, BA

Aliyah King is a third-year medical student at the University of Ottawa Faculty of Medicine and a Canadian finalist for the 2024 Rhodes Scholarship. As the first author, she will present the findings of a systematic review that identifies cutaneous squamous cell carcinoma as a late and underrecognized manifestation of Incontinentia Pigmenti. This work has been published in The Journal of the American Academy of Dermatology Reviews, one of the world's leading peer-reviewed dermatology journals. Outside of medicine, Aliyah enjoys figure skating and represented Canada at the 2016 International Children's Winter Games in Innsbruck, Austria.

Reid Wilson, MD, PhD

Reid Wilson, MD PhD is a vitreoretinal surgery fellow at the Casey Eye Institute at Oregon Health and Science University, where he also completed his residency training in ophthalmology. He graduated from Amherst College with a degree in chemistry cum laude and was a member of the varsity soccer team. He earned his MD from Baylor College of Medicine and his PhD in bioengineering from Rice University as part of their combined Medical Scientist Training Program.

His prior research focused on engineering microenvironments for stem cell-derived organoids and using computational modeling to improve the efficacy of corneal crosslinking. These efforts resulted in a US patent and numerous publications in respected journals. He is a passionate educator and has served in several teaching positions in gross anatomy, neuroanatomy, and bioengineering. He is the recipient of a NIH Ruth L. Kirschstein National Research Service Award Individual Predoctoral MD/PhD Fellowship (F30) and a Heed Fellowship. He is also a member of Alpha Omega Alpha.

He is interested in applying his bioengineering background to develop new imaging technologies to study pediatric and adult retinal diseases, and to improve the ability of cell and gene therapies to treat them. After fellowship, he plans to apply for a K award and pursue a career as a clinician-scientist.

PARTICIPANTS

Nancy Arias, MD

Dr. Nancy Arias is a Research Fellow in Pediatric Retina at the Bascom Palmer Eye Institute, University of Miami, under the mentorship of Dr. Audina Berrocal. She completed a Retina Specialist Fellowship at the Autonomous University of Mexico and an Ophthalmology Residency at the University of Guadalajara.

Dr. Arias is a co-investigator in the study "Macular Thickness and Treatment Response in a Pediatric Sample with Incontinentia Pigmenti: A Cohort Study." She has contributed to several publications and participated in meeting presentations. Her research interests focus on pediatric retina and infrequent diseases.

She has received several prestigious awards, including the Gillingham Pan American Fellowship Program (2024), the Jose Berrocal Award at the Panamerican Retina Society (2024), and First Place in the Clinical Cases Competition at the Challenging Cases: Young Retina Specialists (2022). Dr. Arias is committed to advancing the field of pediatric retina care through clinical excellence and groundbreaking research.

Phillip Ehrich

Phillip Ehrich is a PhD candidate researching the brain involvement of Incontinentia pigmenti. His work involves studying a genetic model of Incontinentia pigmenti using an NF-κB essential modulator (NEMO) knockout mouse line. Utilizing high-resolution imaging techniques and single-cell transcriptomics he investigates cellular interactions and gene expression changes that contribute to inflammation, vascular rarefaction and blood-brain barrier impairment. A particular focus of his research is on dynamic interactions between microglia and cerebral endothelial cells within the neurovascular unit.

PARTICIPANTS

Nicole Somani, MD, MPH

Nicole Somani, MD, MPH is a first-year surgical retina fellow at the University of Iowa. She completed her medical school training at the University of Texas Health Science Center at Houston and ophthalmology residency at Baylor College of Medicine.

Eric Van Otterloo, PhD

Eric Van Otterloo is an accomplished Associate Professor at the University of Iowa's College of Dentistry and the Iowa Institute for Oral Health Research. With over 15 years of expertise in developmental and stem cell biology, his research focuses on the molecular mechanisms of embryonic development, particularly gene-regulatory networks governing craniofacial, dental, and epithelial development. Dr. Van Otterloo has authored over 25 peer-reviewed publications, secured significant NIH and private funding, and earned numerous honors, including the H.W. Mossman Award in Developmental Biology. His work integrates advanced techniques like next-generation sequencing and bioinformatics to explore pathways linked to human development disease.

Dorothy K. Grange, MD

Dorothy K. Grange, MD is a Professor of Pediatrics in the Division of Genetics and Genomic Medicine in the Department of Pediatrics at Washington University School of Medicine. She is a clinical geneticist at St. Louis Children's Hospital. She received her undergraduate degree at Mount Holyoke College and her medical degree at the University of Florida College of Medicine. Dr. Grange completed a residency in pediatrics at the University of Wisconsin and a fellowship in medical genetics at the National Institutes of Health. She is boarded in Pediatrics, Clinical Genetics and Medical Biochemical Genetics. Dr. Grange is a member of the Missouri Genetics Advisory Committee and is currently the Chair of the Newborn Screening Committee. Dr. Grange has served on the Scientific Advisory Council for the National Foundation for Ectodermal Dysplasias since 2006. Clinical research interests include development of new therapies for genetic conditions. She is the principal investigator for the EDELIFE clinical trial for prenatal treatment of XLHED at the Washington University site.

PARTICIPANTS

Saravanan Thangarajan, MDS, MBA

Dr. Saravanan Thangarajan, a Harvard Medical School researcher and trailblazing global health leader, is redefining rare disease care and pioneering transformative healthcare innovations. His groundbreaking efforts have driven a 1.11% reduction in maternal and child mortality, improving the lives of over 7 million individuals in low- and middle-income nations. As an External Scientific Advisory Board Member for the Hypertrophic Olivary Degeneration Association, he spearheads strategic advancements in rare disease research. Renowned for his expertise in digital health and telemedicine, Dr. Thangarajan's leadership of the Tele-MANAS initiative achieved a 1.2% decline in suicide rates, underscoring his ability to convert innovation into lifesaving interventions. His mentorship at the Harvard Rare Disease Hackathon embodies his relentless drive to inspire breakthrough solutions in rare disease management. Through high-impact collaborations with WHO and UNEP, he amplifies the voices of patients and families, ensuring equity remains the cornerstone of global healthcare strategies for rare diseases. Dr. Thangarajan is a strategic leader shaping a healthier, more inclusive world.

Maddison Salois, BS

Maddison Salois received her bachelor of science in biology from Campbell University in North Carolina in 2020. She currently is a fifth year PhD student in the laboratory of Dr. Maranke Koster at The Brody School of Medicine of East Carolina University. Her research is focused on determining how mutant TP63 proteins disrupt pathways crucial for ectodermal development and maintenance in two ectodermal dysplasias: ankyloblepharon-ectodermal defects-cleft lip and/or palate (AEC) and ectrodactyly ectodermal dysplasia-clefting (EEC). Specifically, she is focused on mechanisms by which mutant TP63 proteins expressed in AEC and EEC patients cause functional defects in keratinocyte adhesion and migration. Their team's overall goal is to uncover the pathological mechanisms causing skin manifestations in AEC and EEC, which can contribute to the identification of novel therapeutic strategies to treat patients.

PARTICIPANTS

Markus Schwaniger, MD

Dr. Markus Schwaniger is the Chairman of the Institute of Experimental and Clinical Pharmacology and Toxicology and head of the Department of Pharmacology and Toxicology at the University of Lübeck in Germany. A board certified neurologist and pharmacologist, his research focuses on the blood-brain barrier with a special interest in incontinentia pigmenti. His current projects aim to establish innovative therapeutic approaches.

Shirley Parraga, BS

Shirley Parraga is a fourth-year medical student and research scholar at the Brody School of Medicine at East Carolina University working in the laboratory of Dr. Maranke Koster, PhD. Her research focuses include Ankyloblepharon-Ectodermal Defects-Cleft Lip/Palate Syndrome (AEC) and Ectrodactyly-Ectodermal Defects-Cleft Lip/Palate Syndrome (EEC). Additionally, she completed a research fellowship at the Center for Dermatology Research in the Department of Dermatology at the Wake Forest School of Medicine working under the leadership of Dr. Steven Feldman, MD, PhD. During her research year, she participated in various dermatologic studies with a focus on psoriasis, atopic dermatitis, pigmentary disorders, and hair loss. Her goal is to pursue a career as a pediatric dermatologist while gaining broad experience and serving a diverse patient population.

PARTICIPANTS

Jacques Monnet

I am Jacques Monnet, Chairman of the French IP patients' organization for 17 years. I am the father of 3, of whom Laura, 24, who carries IP. Laura suffers from ocular and CNS disorders: epilepsia, learning difficulties, hemiparesis and failure of the left-and side optic nerve.I am neither a scientist, nor a physician. My expertise is in Finance, Project Management, Performance and strategy analysis. I have worked for 40 years in the chemical , Pharma Industry and the last 20 for the vaccine affiliate of Sanofi. Laura was diagnosed in the early weeks after birth. Laura suffers from brain and ocular disorders epilepsia, over by now, left hand side hemiparisis and failure of left-and side optic nerve, learning disabilities, and cognitive development deficiencies. But Laura is very courageous and brave. With educational support, she has been able to follow standard education till grade 9. Next, she succeeded to pass a degree in family and children care. She works now in a preliminary school. What has been key for her development is the support that she enjoyed all along her childhood and teenage years. (psychomotor therapy, speech therapy, occupational therapy, physiotherapy, orthoptist, psychological support, educational support) In France, the patient organization succeeded to have the 'IP standard carepath' document issued by the Ministry of Health. As an example, neuro and opthalmo treatments to be provided in the early days after birth are reported. Over the past 15 years, as a patients organization, we are proud to have supported financially the works carried by Markus Schwaninger, Jérémie Rosain, Christine Bodemer, Valeria Ursini, Francesca Fusco, and teams. We are grateful to them for their commitment, and breakthroughs to improve the knowledge on IP. Their works have driven to mitigate the most severe disorders carried by IP, and have opened new fields of investigation.

Emily Cole, MD, MPH

Emily Cole is an Assistant Professor at the Sue Anschutz-Rodgers Eye Center at the University of Colorado. She is an adult and pediatric vitreoretinal specialist with a clinical and research focus in retinopathy of prematurity and implementation of artificial intelligence into systems of care. She completed her retina fellowship training at the Kellogg Eye Center at the University of Michigan and ophthalmology residency at the Illinois Eye and Ear Infirmary at the University of Illinois Chicago.

PARTICIPANTS

Susanne Emmerich

In October 1994 a baby girl was born into my family. She had blisters on her arms and legs and was diagnosed immediately with Herpes. This did not seem correct and she was taken to various dermatologists, one of whom was Dr. Seth Orlow. He was familiar with IP, and suspected via anecdotal evidence, this was the cause of the symptoms. Unfortunately, no definite method could determine if this was correct and there was no

By February of 1995, I was determined to identify the gene so an accurate diagnosis could be made for those like my great niece who would be able eventually to benefit from IVF. I had received a science degree from college in 1958 but I had absolutely no experience with a project like this. I had worked in the fashion field, been married and widowed and remarried. I attended an ASHG meeting in Minnesota, posted an invitation to a coffee hour in a room I rented and invited anyone to attend that had an interest in IP. Approximately 20 people showed up and I was able to convince 3 to join me in my project. It subsequently became 5, each from a different country, France, US, England, Germany, Italy. We held 3 - 4 meetings a year, but the consortium members were in touch with each other frequently. Success finally came in 2000 and an article announcing the success appeared in the journal Nature.

The child in my family subsequently graduated from college, summa cum lauda, received a Phd in Psychology and is very happily married.

Teresa Faupel

Teresa Faupel is a researcher and PhD student at the Institute for Experimental and Clinical Pharmacology and Toxicology at the University of Lübeck, Germany. She focuses on brain endothelial cell death, typically found in people with Incontinentia Pigmenti (IP). Here, she aims to elucidate the different molecular mechanisms behind IP. Therefore, she uses a mouse model mimicking neurological symptoms of IP (mice with a knockout of NEMO specifically in brain endothelial cells). She characterizes the time course of changes such as the formation of string vessels found in the brains of these mice and uses genetic models to inhibit molecules involved in the cell death pathway with the ultimate goal to identify suitable targets to ameliorate endothelial cell death in IP.

PARTICIPANTS

J. Timothy Wright, DDS, MS

Tim Wright received his DDS degree from West Virginia University and completed his Pediatric Dentistry training and Master of Science Degree at the University of Alabama at Birmingham. He currently is a Professor in the Department of Pediatric Dentistry and Dental Public Health at The University of North Carolina, Chapel Hill and has served as Pediatric Dentistry Department Chair and Department of Endodontics Department Interim Chair. He is a Diplomat of the American Board of Pediatric Dentistry and a Fellow of the American Academy for the Advancement of Science and is a Past President of the AADR. He has published over 200 peer reviewed scientific manuscripts, edited text three texts, authored 30 text chapters and had NIH funding for over 25 years. He has chaired the Counsel on Scientific Affairs for the American Dental Association and American Academy of Pediatric Dentistry and is the Current AAPD Chair for the Evidence Based Dentistry Committee. Dr. Wright serves as the Editor in Chief of the Journal of the American Dental Association.

Jeremie Rosain, PhD

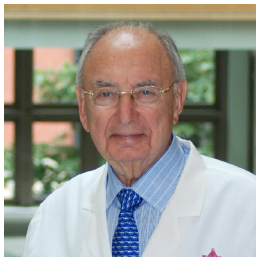
I'm a immunologist and geneticist with a primarily interest for rare disease affecting the immune system. I'm working both in a clinical laboratory and in a research laboratory

PARTICIPANTS

Audina Berrocal, MD

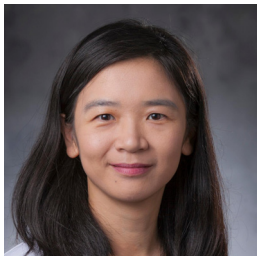
Audina “Nina” Berrocal, MD received her undergraduate education at Princeton University followed by medical school at Tufts University School of Medicine. She stayed at Tufts/New England Eye Center, where she completed her ophthalmology residency. Dr. Berrocal subsequently received vitreoretinal surgery and uveitis training at the Bascom Palmer Eye Institute, University of Miami Miller School of Medicine. She has since remained on the faculty at Bascom Palmer, where she holds currently the rank of Professor of Clinical Ophthalmology. She also serves as Director of the Pediatric Retina service at Bascom Palmer Eye Institute and Jackson Memorial Hospital and Vitreoretinal fellowship director. Dr. Berrocal is internationally recognized for her approach to pediatric retinal diseases. As a pediatric retina specialist and Director of Pediatric Retina and Retinopathy of Prematurity Service, she has extensive clinical, surgical and research experience in the diagnosis and treatment of ROP in this population. She has performed extensive basic and clinical research studies in ROP, and have co-authored over 70 peer-reviewed papers in these areas. She received the Bernadotte Foundation Award for her work in ROP. Additionally, she maintains a robust adult vitreoretinal surgical practice. Dr.Berrocal is an active member of The Retina Society, The Macula Society, Club Jules Gonin and holds leadership positions in many of these groups including the American Academy of Ophthalmology and the American Society of Retina Specialists and is one of the founding members of the Vit Buckle Society. She is the recipient of the ASRS Crystal Apple Award for her dedication to surgical teaching and of the Bernice Z. Brown Lecture Award for her dedication to the advancement of women.

PARTICIPANTS



Morton F. Goldberg, MD

Dr. Goldberg became Professor and Head of the Department of Ophthalmology at the University of Illinois at age 32, working there for 19 years. Following that, he became the William Holland Wilmer Professor of Ophthalmology and Director of the Wilmer Eye Institute at Johns Hopkins, serving for 14 years. Following this, he has remained on that fulltime academic faculty, to the present, at Johns Hopkins as the Joseph Green Professor of Ophthalmology and Director Emeritus of the Wilmer Eye Institute. His major fields of interest include vascular retinal diseases, such as sickle cell anemia; persistent fetal vasculature; laser photocoagulation; incontinentia pigmenti; and others. Honors have included the Howe Medal of the American Ophthalmological Society, The Laureate Award of the American Academy of Ophthalmology, the Life Achievement Honor Award of the American Academy of Ophthalmology, membership in the National Academy of Medicine, Chief Editor (for 10 years) of Archives of Ophthalmology; and presidencies of the Macula Society, the Association for Research in Vision and Ophthalmology; the Chicago Ophthalmological Society, and the Association of University Professors in Ophthalmology. Other honors include the Inaugural Cunha-Vaz Lectureship in Portugal, the Inaugural Ida Mann Lectureship in Oxford, and the Inaugural Venkataswamy Oration Award in Madurai, India. Dr. Goldberg has co-authored and edited about 600 original articles and chapters, and has written or edited 10 books on ophthalmic subjects. He was active in participating in the creation of the following two ophthalmology buildings: the Lions of Illinois Eye Research Institute in Chicago (for clinical care and basic research) and the Clarice and Robert H. Smith Building at Johns Hopkins (for eye surgery and basic research). Endowed professorships in his name have been established at the University of Illinois and at Johns Hopkins. In 1999, Ophthalmology Times selected him as one of the “ten greatest living ophthalmologists.” Currently, Dr. Goldberg is Co-Founder of the RYR-1 Foundation (www.ryr1.org), for which he serves as Co-Chairman of its Research Committee. The RYR-1 Foundation provides funding internationally, for numerous research projects related to inherited, congenital muscle diseases with RYR-1 mutations.



Xi Chen, MD, PhD

Dr. Xi Chen is an adult and pediatric retina specialist and a clinician-scientist in translational research at Duke University with a special focus on imaging retinal vascular development and pathology. She completed ophthalmology residency training at Massachusetts Eye and Ear Infirmary and vitreoretinal fellowship at Duke Eye Center. She joined the faculty at Duke Eye Center in 2017. She works with her ophthalmology and biomedical engineering colleagues in the DARSI laboratory to study pediatric retinal development and diseases.

PARTICIPANTS

Philip Ferrone, MD

Dr. Ferrone earned his Bachelors of Science from Union College magna cum laude and received his M.D. from Harvard Medical School cum laude with special honors.

He completed an internship at Newton-Wellesley Hospital in Massachusetts and his residency at Duke University Medical Center. He subsequently completed a fellowship in vitreoretinal diseases and surgery at Associated Retinal Consultants in Royal Oak, MI. Dr Ferrone is board certified by the American Board of Ophthalmology. Dr Ferrone currently practices with Vitreoretinal Consultants of New York.

Dr. Ferrone is a member of the American Academy of Ophthalmology, NYS Ophthalmological Society, Retina Society, Macula Society, Club Jules Gonin, American Society of Retinal Specialists. Dr. Ferrone is Past- President of the American Society of Retinal Specialists. Dr. Ferrone is a Professor of Ophthalmology at Donald and Barbara Zucker School of Medicine at Hofstra/Northwell.

He has expertise in adult, as well as pediatric retinal disease, and he has authored many scientific articles as well as textbook chapters on retinal disease. Dr. Ferrone received a Senior Honor Award for his contributions to the American Society of Retinal Specialists and a Senior Achievement Award from the American Academy of Ophthalmology.

Eric Nudleman, MD, PhD

Eric Nudleman, MD, PhD is an Associate Professor of Ophthalmology at the Shiley Eye Institute, UC San Diego, where he serves as the Co-Director of the Retina Division and the Director of Pediatric Retina at Rady Children's Hospital. He holds the prestigious Viterbi Family Chair for Retinal Vascular Disease. He joined UC San Diego (UCSD) after completing his fellowship in vitreoretinal surgery at the renowned William Beaumont Hospital. Prior to his fellowship, Dr. Nudleman graduated from Stanford University with bachelors and doctoral degrees. He earned his medical degree at Albert Einstein College of Medicine of Yeshiva University in New York then went on to Washington University School of Medicine in St. Louis, Missouri for his residency. Dr. Nudleman is the recipient of many prestigious honors such as the Ronald G. Michels Fellowship, Heed Fellowship, the Doris P. and Harry I. Wexler Prize, Rosenbaum Research Award, and the Association of University Professors of Ophthalmology / Research to Prevent Blindness Resident and Fellow Research Forum Award. He has participated in multiple National Eye Institute and industry-sponsored clinical trials. At UCSD, Dr. Nudleman's clinical focus is on vitreoretinal diseases and surgery, with a special interest in pediatric vitreoretinopathies. His NIH funded laboratory focuses on developmental angiogenesis with a particular interest in identifying novel targets to treat retinal vascular diseases, including vascular leakage, neovascularization, and fibrosis.

POSTER PRESENTERS

Aliyah King, BA
Nancy Arias, MD
Nicole Somani, MD, MPH
Saravanan Thangarajan, MDS, MBA
Maddison Salois, BS
Shirley Parraga, BA
Emily Cole, MD, MPH

SPONSOR

PATIENT PERSPECTIVE

Susanne Emmerich and Jacques Monnet

CONFERENCE WRITERS

Reid Wilson, MD, Phd
Maddison N. Salois, BS
Shirley Parraga, BS
Nicole Somani, MD, MPH

AWARD DEDICATIONS

A TRIBUTE TO SUSANNE EMMERICH: A CHAMPION FOR FAMILIES AND RESEARCH IN INCONTINENTIA PIGMENTI

The National Foundation for Ectodermal Dysplasias proudly salutes **Susanne Emmerich** for her extraordinary contributions to the understanding and advancement of **incontinentia pigmenti (IP).**

For 25 years, as the **founder and director** of the Incontinentia Pigmenti International Foundation (IPIF), Susanne dedicated herself to connecting the brightest minds in medicine and research. Under her leadership, experts from around the world gathered annually to share knowledge, develop treatment protocols, and forge new paths in the study of this rare genetic condition. Thanks to their collaborative efforts, critical guidelines were established to address the significant **eye issues** associated with IP, improving care for those affected.

Susanne was also a driving force in **building a comprehensive medical library** on IP, ensuring that healthcare professionals had access to vital research and resources. Her unwavering commitment to education and awareness helped push the field forward, and her advocacy played a pivotal role in identifying the **gene responsible for IP**. This breakthrough made genetic testing possible, offering clarity and answers to families navigating an uncertain diagnosis.

Yet, beyond the scientific advancements, Susanne's greatest legacy lies in her **personal impact** on families affected by IP. She was a tireless advocate, always available to offer guidance, comfort, and reassurance. No matter where she was, her laptop was never far—ensuring she never missed an email from a family in need.

Her compassion, vision, and dedication have left an indelible mark on the IP community. We honor her **unwavering commitment, pioneering spirit, and profound kindness.** Thank you, Susanne, for being a beacon of hope to so many.

AWARD DEDICATIONS

A TRIBUTE TO MORTON GOLDBERG

It's our distinct honor to have **Morton F. Goldberg, M.D.** participating in this Incontinentia Pigmenti Conference.

On behalf of the families we serve who are affected by **incontinentia pigmenti (IP)**, we extend our deepest gratitude to Dr. Goldberg for his invaluable contributions to the study of IP and its **ocular manifestations**. His pioneering research has greatly advanced our understanding of the retinal and vascular complications associated with this condition, leading to **improved recognition and management for affected individuals.**

Dr. Goldberg's dedication to ophthalmology and his commitment to scientific discovery have left a **lasting impact** on the field, providing hope and guidance for both patients and clinicians. His work continues to shape our knowledge and inspire future research, **ensuring better outcomes** for those living with IP.

Thank you, Dr. Goldberg, for your **remarkable efforts in expanding our understanding** of this rare disorder. Your legacy in IP research will be felt for generations to come.

WITH DEEPEST GRATITUDE

For more than 20 years, the National Foundation for Ectodermal Dysplasias has been committed to finding solutions to improve the quality of life for individuals affected by ectodermal dysplasias like incontinentia pigmenti (IP).

We sincerely thank Oregon Health and Science University (OSU) for their generous sponsorship. Your support made this conference a reality. Special thanks also go to our dedicated co-chairs—Becky Abbott, Peter Campbell, Connie Chen, and Ian Han—whose countless hours of hard work and teamwork were instrumental in making this event a success. Your efforts are deeply appreciated!

We are also grateful to Susanne Emmerich for her extraordinary contributions to advancing our understanding of IP. Her unwavering commitment to education and awareness has brought much-needed clarity and answers to families facing this uncertain diagnosis. We also express profound gratitude to Dr. Morton Goldberg for his contributions and pioneering research. His work has advanced our understanding of IP and its complications, improving recognition and management for those affected.

Finally, thank you to all participants for attending this conference and sharing in our vision to alleviate the challenges that ectodermal dysplasias present to those affected. Your expertise and dedication are vital to creating a path toward better outcomes and a brighter future for individuals living with IP. Together, we are making meaningful progress.

- Mary Fete

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