INTERNATIONAL ECTODERMAL DYSPLASIAS RESEARCH CONFERENCE

TRANSLATING DISCOVERY TO THERAPY

OCTOBER 21-24, 2021

nfed

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NATIONAL FOUNDATION FOR ECTODERMAL DYSPLASIAS

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WELCOME!

Dear Colleague,

It is our pleasure to welcome you to this unique International Research Conference, Transforming Discovery to Therapy sponsored by the National Foundation for Ectodermal Dysplasias (NFED). You represent a very prestigious group of scientists and researchers.

The purpose of this international conference is to provide a unique format to promote open discussion and exchange of ideas among an international group of scientists to help advance therapeutics for rare diseases that affect ectodermal tissues. The conference goal is to develop opportunities and overcome barriers to help advance early diagnostics, treatments and cures that address the significant morbidity in people with developmental ectodermal defects. To accomplish this goal, the conference will seek to facilitate collaborative research.

We are pleased that you are able to join us in this extraordinary opportunity. The conference participants represent the world leaders in cutting edge research in many areas. Our combined expertise and collaboration are vital to developing a five-year roadmap to expediting treatments and cures for not only ectodermal dysplasias but also other rare conditions.

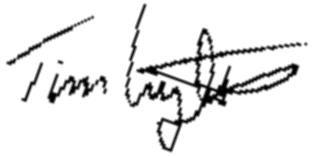
This conference will set our five-year agenda for the NFED Research Program and we plan to fund projects immediately following the conference.

Welcome and enjoy the conference!

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

Becky Abbott, MPH
Conference Organizer
Director, Research and Treat-
ment Advocacy
National Foundation for
Ectodermal Dysplasias

Maranke Koster, PhD
Conference Co-Chair
Professor, Department of
Anatomy and Cell Biology
Brody Schoo l of Medicine
East Carolina University



J. Timothy Wright, DDS, MS
Conference Co-Chair
Bawden Distinguished Professor
Pediatric and Public Health
Adams School of Dentistry
The University of North Carolina

HOUSE RULES

Below are some guidelines we ask all attendees to follow so that everyone is able to have a pleasant experience and absorb as much as they can from these amazing sessions.

1. You will be muted upon entry. Please remain muted until the end of the session when the moderator announces that we will open the session for questions and answers.

2. If a session is running late, please be patient. The moderator will end the session and start the next session as soon as possible.

3. Please be advised we will have student writers taking notes and pictures of the event.

4. Please give everyone an opportunity to participate and be mindful of time restraints.

5. To easily identify each attendee, we ask that you enter your name in Zoom as First Name, Last Name, Credentials and Affiliation/Institution.

6. We encourage you to enable your camera during sessions to enhance the conference experience and discussions. Please make sure your camera is stable and positioned properly.

7. If you have any technical issues or questions before or during the sessions, please reach out to kayla@nfed.org and she will respond promptly as allowed.

AGENDA

All times are listed in Eastern time.

All sessions on Thursday are in Woodlawn unless otherwise noted.

THURSDAY, OCTOBER 21

7 a.m. - 8 a.m.	Breakfast
8 - 8:15	Introduction *Mary Fete and Jacques Monnet*
8:15 - 9:05	Preclinical Studies on the Neurovascular Manifestations of Incontinentia Pigmenti *Markus Schwaniger, Josephine Lampe*
	Role of RIPK1 Kinase Activity in Inflammation and Cell Death *Matija Zelic and Dimitry Ofengeim*
9:05 - 9:45	The Human Genetic and Immunological Determinants of Life-Threatening COVID-19 *Jean-Laurent Casanova*
	Auto-Abs to Type I IFN and Viral Diseases in Women with IP *Jeremie Rosain,Paul Bastard*
9:45 - 9:55	Break
9:55 - 10:35	Other Clinical Manifestations of IP *Christine Bodemer*
10:35 - 11:15	From the IP Phenotypic Variability to the Role of NEMO in Cell Death *Matilde Valeria Ursini, Alessandra Pescatore, Francesca Fusco*
11:15 - 11:45	Incontinentia Pigmenti in Adults *Angela Scheuerle*
11:45 - 12	Break
12 - 1:30	Lunch with Discussion *Jacques Monnet: Identify Next Steps and Deliverables*
5:30 - 7	Welcome Reception - Salon A
7 - 9	Dinner - Salon A

AGENDA

FRIDAY, OCTOBER 22

All times are listed in Eastern time.

All sessions on Friday are in the Queen City Ballroom unless otherwise noted.

7 - 8	Breakfast
8 - 8:10	Welcome *Mary Fete*
8:10 - 8:35	Discovery and Trends in Science *Harold C. Slavkin*
8:35 - 9	Scope and Diversity of Patient Issues *J. Timothy Wright*
9 - 9:30	Setting Goals and Defining Deliverables *Philip Reilly*
9:30 - 10:10	Affected Individuals and Family Issues Session *Moderator: Karl Nelsen* *Families: Becky Abbott, Ruth Geismar and Jaques Monnet*
10:10 - 10:30	Break
10:30 - 11:05	Development of a Comprehensive Genetic Landscape for Ectodermal Dysplasia Genes *Mark Kiel*
11:05 - 11:35	Comparative RNA-Sequencing Analysis Enables Individualized Treatments for Pediatric Cancers *Gina D. Mawla*
11:35 - 12:05	Ushering in a Genetics-Informed Era in Medicine, Beginning With Newborn Screening *Maria Kousi*
12:05 - 1	Lunch

AGENDA

FRIDAY, OCTOBER 22 (CONT.)

All times are listed in Eastern time.

All sessions on Friday are in the Queen City Ballroom unless otherwise noted.

1 - 1:30	Experience and Expectation: Progress over Four Decades *Angus Clarke*
1:30 - 2:15	Wnt Signaling in Ectodermal Development, Regeneration and Disease *Sarah Millar*
2:15 - 2:45	From Genes to Organs: Genetic Pathways in Ectodermal Dysplasias *Maranke Koster*
2:45 - 3:15	Decipher p63-Controlled Gene Regulatory Network During Development and Disease *Rui Yi*
3:15 - 3:30	Break
3:30 - 4:30	Breakout Groups

Group 1 - Queen City Ballroom
Diagnostics and Phenotyping
Angus Clarke and Maria Kousi

Tim Wright, Tim Fete, Mary Fete, Kathy Grange, Elaine Siegfried, Sarah Millar, Smail Hadj-Rabia, Maria Morasso, Ophir Klein, Rob Coppes, George Cotsarelis, Encarna Guillen-Navarro, Vincent Falanga, Markus Schwaninger, Josephine Lampe, Alessandra Pescatore, Jeremie Rosain, Michele de Luca, Philip Reilly, Spiro Getsios, Matija Zelic, Kevin Byrd, Mary Kaye Richter, Shivani Awasthi, Anges Jaulent, Caroline Kant, Christine Bodemer, Stefano Sol, Bronwyn Dillon, Maria Carmen Martínez-Romero, Cendrine Grangeon

AGENDA

All times are listed in Eastern time.

All sessions on Friday are in the Queen City Ballroom unless otherwise noted.

FRIDAY, OCTOBER 22 (CONT.)

Group 2 - Woodlawn
Prevalence and Bioinformatics
Mark Kiel

Maranke Koster, Clayton Butcher, Becky Abbott, Karl Nelsen, Clark Stanford, Holm Schneider, Daniel Aberdam, Riu Yi, Yorick Post, Peter Marankovich, Brad Amendt, Matilde Valeria Ursini, Jean-Laurent Casanova, Francesa Fusco, Angela Scheuerle, Paul Bastard, Hal Slavkin, Jean DePascal, Pascal Schneider, Dimitry Ofengeim, Yang Chai, Fabio Mammano, Jaques Monnet, Marketa Saint Aroman, Alain Delarue, Marlene Guiraud, Valerie Ortis, Cecile Fournier, Ezia Spinosa, Cynthia DeKlotz, Christine Cazeau, Rena D'Souza, Caroline Miklaszewski, Marylaure Laborderie

4:30 - 5:15 Breakout Reports and Discussion
Ophir Klein and Tim Wright

5:15 - 7 Poster Session and Cocktails

Data Sharing and Global Collaboration Accelerate the Research in Rare Diseases:
The Incontinentia Pigmenti Genetic Biobank
Ezia Spinosa

Identification of Disaggregating Compounds in the Treatment of Skin Defects
in AEC Syndrome
Stefano Sol

Auto-antibodies to Type I Interferons and Viral Diseases in Women
With incontinentia pigmenti
Jeremie Rosain

7 - 9 Dinner and Continued Discussion

AGENDA

SATURDAY, OCTOBER 23

All times are listed in Eastern time.

All sessions on Saturday are in the Queen City Ballroom unless otherwise noted.

7 - 8	Breakfast
8 - 8:45	Opposite Sides of a Spectrum: Oral Wound Healing vs. Chronic Non-Healing Wounds *Maria Morasso*
8:45 - 9:30	P63-related Ectodermal Syndrome: From Stem Cell Modeling to Drug Repurposing *Daniel Aberdam and Smail Hadj-Rabia*
9:30 - 10:15	Plenary Speaker *George Cotsarelis*
10:15 - 10:30	Break
10:30 - 11:15	Combined Cell and Gene Therapy for Epidermolysis Bullosa *Michele De Luca*
11:15 - 11:45	Lessons from Impaired Healing *Vincent Falanga*
11:45 - 12:15	Bioprinting Human Tissue for Therapeutic Applications *Spiros Getsios*
12:15 - 1:15	Lunch
1:15 - 2	In Vivo Versus Ex Vivo Gene Therapy for Epidermolysis Bullosa *Peter Marinkovich*
2 - 2:30	The Human Genetic and Immunological Determinants of Life-Threatening COVID-19 *Paul Bastard*
2:30 - 3	Development, Renewal and Plasticity of Oral and Dental Tissues *Ophir Klein*

AGENDA

SATURDAY, OCTOBER 23 - CONT.

All times are listed in Eastern time.

All sessions on Saturday are in the Queen City Ballroom unless otherwise noted.

3 - 3:30 New MicroRNA Technology and Therapeutic Applications
Brad Amendt

3:30 - 3:45 Break

3:45 - 4:45 Breakout Groups

Group 1 - Queen City Ballroom
Would Healing and Tissue Repair
Maria Morasso and Peter Marinkovich

Tim Wright, Tim Fete, Mary Fete, Kathy Grange, Elaine Siegfried, Sarah Millar, Smail Hadj-Rabia, Maria Morrasso, Ophir Klein, Rob Coppes, George Costerelis, Encarna Guillien-Navarro, Vincent Falanga, Markus Schwanniger, Josephine Lampe, Alessandra Pescatore, Jeremie Rosain, Michele de Luca, Philip Reilley, Spiro Gestios, Matija Zelic, Kevin Byrd, Mary Kaye Richter, Shivani Awasthi, Anges Jaulent, Caroline Kant, Christine Bodemer, Stefano Sol, Bronwyn Dillon, Maria Carmen Martínez-Romero, Cendrine Grangeon

Group 2 - Woodlawn
Gene/Tissue Specific Approaches
Michele de Luca and Maranke Koster

Maranke Koster, Clayton Butcher, Becky Abbott, Karl Nelsen, Clarke Stanford, Holm Schneider, Daniel Aberdam, Riu Yi, Yorick Post, Peter Marankovich, Brad Ahmendt, Ursini Valeria, Jean-Laurent Casanova, Francesa Fusco, Angela Scheuerle, Paul Bastard, Hal Slavkin, Giovanni DiPasqualle, Pascal Schneider, Dimitry Ofengeim, Yang Chai, Fabio Mammano, Jacques Monnet, Marketa Saint Aroman, Alain Delarue, Marlene Guiraud, Valerie Ortis, Cecile Fournier, Ezia Spinosa, Cynthia DeKlotz, Christine Cazeau, Rena D'Souza, Caroline Miklaszewski, Marylaure Laborderie

AGENDA

SATURDAY, OCTOBER 23 - CONT.

All times are listed in Eastern time.

All sessions on Saturday are in the Queen City Ballroom unless otherwise noted.

4:45 - 5:30	Breakout Reports and Discussion *Brad Amendt*
5:30 - 6:30	Cocktail Reception - Salon A
6:30 - 8	Dinner with the NFED Board of Directors - Salon A
8 - 9:30	NFED Board of Directors Meeting - Woodlawn

AGENDA

SUNDAY, OCTOBER 24

All times are listed in Eastern time.

All sessions on Saturday are in the Queen City Ballroom.

7-8	Breakfast
8 - 8:30	Novel approaches in identifying disease pathways and implications for therapeutic design *Maranke Koster*
8:30 - 8:55	A Potent Antagonist Antibody Targeting Connexin Hemichannels Alleviates Clouston Syndrome Symptoms in Mutant Mice *Fabio Mammano*
8:55 - 9:20	WNT10A Role In Ectodermal Dysplasias And Screening Approaches For New Therapeutics *Encarna Guillen-Navarro and Maria Carmen Martínez-Romero*
9:20 - 9:45	Understanding Tear Secretion with Lacrimal Gland Organoids *Yorick Post*
9:45 - 10:05	NIH and NIDCR's Vision for Turning Discoveries into Health for All *Rena D'Souza*
10:10 - 10:30	Epigenetic Regulation Of Progenitor Cells During Tooth Root Development *Yang Chai*
10:30 - 10:45	Break
10:45 - 11:15	Development of a Stem Cell Therapy for Head and Neck Cancer Treatment Induced Xerostomia *Rob Coppes*
11:15 - 12	Molecular Pathway Directed Therapeutic Approaches *Pascal Schneider*

AGENDA

SUNDAY, OCTOBER 24 - CONT.

All times are listed in Eastern time.

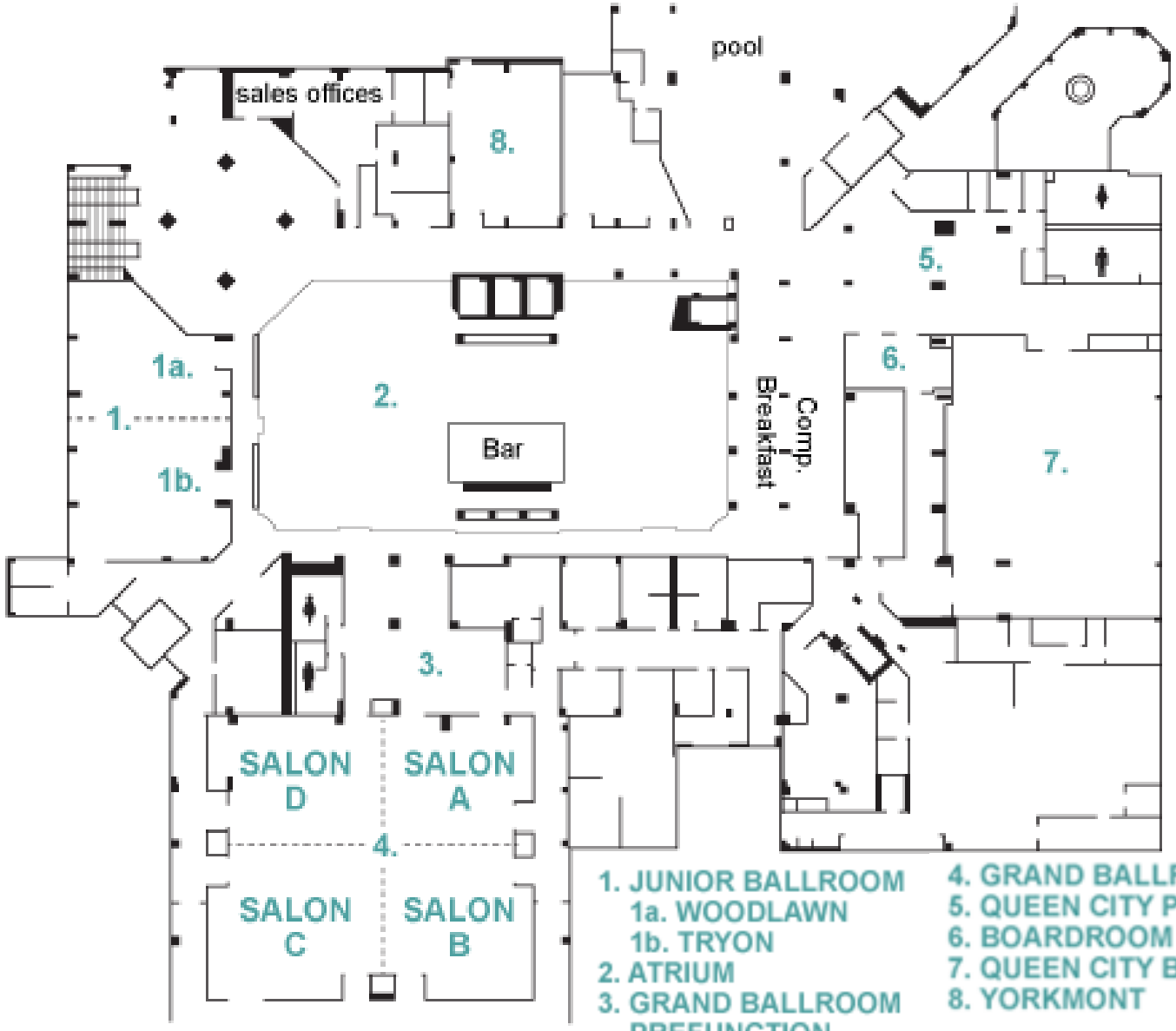
All sessions on Saturday are in the Queen City Ballroom.

Short-Term Correction of Deficient Ectodysplasin A Signaling With a Replacement Protein: Long-Term Results in Nine Patients
Holm Schneider

12 - 1 Define Deliverables and Next Steps
Tim Wright

1 - 2 Working Lunch and Departure

HOTEL MAP


pool
sales offices
8.
5.
6.
1a.
1.
1b.
2.
Bar
Comp. Breakfast
7.
3.
SALON D
SALON A
4.
SALON C
SALON B
1. JUNIOR BALLROOM
1a. WOODLAWN
1b. TRYON
2. ATRIUM
3. GRAND BALLROOM PREFUNCTION
4. GRAND BALLROOM
5. QUEEN CITY PREFUNCTION
6. BOARDROOM
7. QUEEN CITY BALLROOM
8. YORKMONT

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

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.

NFED LEADERSHIP

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Vice President
Anil Vora

Treasurer
George Barbar

Secretary
Keith Throm

Krista C. Basile

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Roy Moffitt

Richard Stratz

Marianne Vermeer

J. Timothy Wright, D.D.S., M.S.

Honorary Member
Mary Kaye Richter

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University of Illinois – Chicago School of Dentistry

Vice-Chair
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Liaison from the Board of Directors
J. Timothy Wright, DDS, MS
University of North Carolina Chapel Hill

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Cynthia Marie Carver DeKlotz, MD

Dorothy Katherine Grange, MD
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Brandon Hopkins, MD
Cleveland Clinic Head and Neck Institute

Maranke I. Koster, PhD
East Carolina University

Richard A. Lewis, MD, MS
Baylor College of Medicine

Pilar L. Magoulas, MS, CGC
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Saint Louis University School of Medicine

Raj Sindwani, MD, FACS, FRCS
Cleveland Clinic, Head and Neck Institute

Ad Hoc Members
Karl Nelsen, PA-C, MS
NFED Family Member

Anil Vora
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.

We have made enormous strides in the understanding and treatment of the 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

- Identify and define a deliverable therapeutic to bring to trial in a 1-2 year time frame.
- Establish research priorities needed to achieve having deliverable therapeutic in the short term.
- Develop collaborations between groups to focus on deliverable therapeutic.
- Dissemination of conference proceedings through publication.

Long Term

- Support and sustain long-term collaborations directed at diagnostics and therapeutics.
- Development of effective therapeutics to ectodermal defects.

CONFERENCE CO-CHAIRS

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

Becky Abbott is the Manager of Research and Treatment 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 has practiced as a registered nurse for more than 30 years in a variety of specialties and practice areas. Mary also has mentored new nurses, developed education programs and curriculum for nurses, and provided education for professionals, patients, and families. She has held many nursing leadership roles. Mary manages NFED National Family Conferences, the Research Program, the Mary Kaye Richter Treatment Program, and coordinates scientific meetings, symposia, and grand rounds. She has several articles published on ectodermal dysplasias in peer-reviewed publications. Mary brings her experience in health care, nursing and insurance to the NFED. She joined the NFED team in 2002 and was promoted to Executive Director in January 2014.

CONFERENCE CO-CHAIRS

Maranke Koster, PhD

Dr. Maranke Koster received her MS degree from VU University in Amsterdam and her PhD degree from Baylor College of Medicine. She currently is a Professor in the Department of Anatomy and Cell Biology at Brody School of Medicine at East Carolina University. Research in the Koster laboratory is aimed at investigating the pathological mechanisms that underlie skin and cornea fragility in patients affected by the TP63-related ectodermal dysplasias, AEC and EEC. In collaboration with the Koch laboratory, her laboratory develops and analyzes in vitro and in vivo models to identify cell and molecular defects resulting from TP63 mutations. The ultimate goal of these studies is to contribute to the development of novel therapeutics aimed at treating skin and cornea lesions in AEC and EEC patients. Dr. Koster's research has been continuously funded by the NIH as well as private foundations. She has published over 50 manuscripts in peer-reviewed scientific journals and authored 7 book chapters. Dr. Koster also serves on the Scientific Advisory Council of the National Foundation for Ectodermal Dysplasias.

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 the Distinguished Bawden Professor in the Division of Pediatric and 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 the Immediate Past President of the AADR. He has Chaired the Counsel of Scientific Affairs for the American Dental Association and American Academy of Pediatric Dentistry. He has published over 200 peer reviewed scientific manuscripts, edited two texts and authored 24 text chapters. Dr. Wright is the Editor in Chief of the Journal of the American Dental Association. He serves on the Scientific Advisory Board and Board of Directors for the National Foundation for Ectodermal Dysplasias.

PARTICIPANTS

Daniel Aberdam, PhD

Daniel Aberdam received his PhD from the Weizmann Institute of Sciences (Rehovot, Israel) under the supervision of Prof. Leo Sachs. He was recruited at INSERM (France) as Director of Research in 1995 and served as head of INSERM unit in Nice and more recently in Paris. He became an expert in skin pathophysiology and more particularly on genodermatoses. He participated in the discovery of genes responsible for epidermolysis bullosa syndromes and used pluripotent stem cells to recapitulate skin and cornea embryonic development to identify both major molecular actors involved and potential therapeutic tools. It allowed more recently his team to demonstrate that a repurposed drug applied topically is able to treat severe skin erosions in ectodermal dysplasia patients. His team has furthermore modelized aniridia-associated keratopathy (AAK) in vitro by gene editing and identified small compounds able to rescue in vitro the pathological phenotype. In collaboration with partners of Aniridia COST and EU network, his team aims at understanding the mechanism of action of these drugs and validating in vivo these potential therapeutic compounds to treat AAK in patients.

Brad Amendt

Brad Amendt and his team have identified several new genetic modifiers of ectodermal organ development. Their laboratory has focused on the role of transcription factors in tooth development. The decreased or increased expression of these factors can affect ectodermal organ development. Several factors are closely associated with Wnt signaling and regulate the development and specification of stem cells during ectoderm development. These factors and others regulate the periderm, and a lack of periderm can affect epithelial invagination and ectodermal organ development. Recently, their research has identified microRNAs (miRs) as regulators of ectodermal organ development by targeting specific transcription factors. They developed a new system to inhibit miRs in mouse models and as a gene therapy reagent to treat diseases and regenerate tissues. miRs can specifically regulate the endogenous ectoderm stem cell niche to either inhibit stem cell differentiation or increase stem cell proliferation. Application of their new miR inhibitors to developmental and genetic anomalies has great potential for non-invasive treatments of ectodermal defects. New studies in their lab use these molecules for wound healing, to regenerate tissues, and dental supporting structures.

Paul Bastard

Paul Bastard is a MD- PhD student in pediatrics and immunology. He works at the Necker Hospital in Paris and the laboratory of human genetics of infectious diseases, headed by Jean-Laurent Casanova. His work focuses on the genetic and immunological predisposition to severe viral diseases, and study the autoantibodies to type I interferons in COVID-19 and other viral diseases.

PARTICIPANTS

Christine Bodemer

Professor Christine Bodemer is the Head of the Department of Dermatology at Necker Enfants-Malades Hospital in Paris, France since 2011. She is a Professor of Dermatology at the Paris Descartes-Sorbonne University since 2001. She obtained a doctorate in skin biology in 1997 and a qualification to lead a research team in 2000. She is more particularly specialized in pediatric dermatology. One of her main areas of interest is rare skin diseases in children. She coordinates a national expert center (MAGEC), the French national network (FIMARAD) and the European network (ERN-SKIN) on rare diseases of the skin. She is a member of many scientific societies, associate editor of various newspapers, such as the British Journal of Dermatology, and has been president of the French Society of Pediatric Dermatology and the secretary of the European Society of Pediatric Dermatology (ESPD). She is currently a member of the board of directors of the International Society in Pediatric Dermatology (ISPD). She has developed research and clinical networks in dermatology, and research programs supported by national and international grants, leading to translational innovative therapies and patents.

Jean-Laurent Casanova, MD, PhD

Jean-Laurent Casanova received his MD in 1987 and his Ph.D. in 1992, after training at the Pasteur Institute in Paris and the Ludwig Institute for Cancer Research in Lausanne. He was appointed professor at Necker in 1999 and with Laurent Abel, cofounded the Laboratory of Human Genetics of Infectious Diseases. He was appointed professor at Rockefeller University in 2008 and named HHMI investigator in 2014. He continues to partner with Dr. Abel, maintaining their lab in Paris and NY. Over the last 25 years, they discovered the first monogenic inborn errors of immunity, rare and common, which predispose otherwise healthy individuals to a single infectious disease, including a variety of viral, bacterial, and fungal infections. In response to the SARS-CoV2 Global Pandemic, Dr. Casanova co-founded the COVID Human Genetic Effort with Helen Su at the NIAID. He discovered monogenic inborn errors of type I interferon immunity underlying severe forms of COVID-19 in previously healthy individuals. This led to his discovery that pre-existing autoantibodies to type I interferons account for at least 10% of severe cases.

Yang Chai, DMD, DDS, PhD

Dr. Yang Chai is the University Professor and the George and MaryLou Boone Chair in Craniofacial Biology at the University of Southern California. He serves as the Director of the Center for Craniofacial Molecular Biology (CCMB) and Associate Dean of Research at the Herman Ostrow School of Dentistry of USC. Dr. Chai earned a DMD degree from Peking University School of Stomatology as well as DDS and PhD in Craniofacial Biology from the University of Southern California. He is most noted for his research on craniofacial development and birth defects, which has transformed the field and led to the successful rescue of cleft palate in animal models. He also studies stem cells and is currently using innovative 3D-printed scaffolds seeded with stem cells to regenerate tissue for patients who have lost bone due to trauma, congenital defects, or diseases. Dr. Chai has authored more than 160 scientific papers and numerous book chapters, and recently completed a book on craniofacial development. Dr. Chai has been continuously funded by the National Institute of Health for more than 25 years.

PARTICIPANTS

Angus Clarke

Dr. Angus Clarke is a Professor in Clinical Genetics at the School of Medicine, Cardiff University in Wales, UK. Coming from a background in pediatrics, Angus has worked in clinical genetics at Cardiff since 1989, with particular interests in ectodermal dysplasia and Rett syndrome, the genetic counselling process and the social and ethical issues around human genetics. He has authored or edited 10 books, often with colleagues, and numerous research papers and book chapters. He directed the Cardiff University MSc course in Genetic Counselling from when it was launched in 2000 until 2018. He contributes to policy discussions - he was a member of the Human Genetics Commission - and the drafting of policies and reports for the British and European Societies of Human Genetics. Angus has actively supported the Ectodermal Dysplasia Society UK for many years and is the Chair of their Medical Advisory Board. He was the principal investigator in the UK branch of the recent trial of a novel gene-based treatment for infant males affected by X-linked hypohidrotic ectodermal dysplasia. He hopes soon to be engaged in the trial of the same treatment molecule to be administered in utero to affected males still in the womb, in the second half of the pregnancy.

Rob Coppes, PhD

Rob Coppes did a PhD in molecular pharmacology and a post-doc in radiobiology. He became a full professor of radiotherapy with a focus on the radiation biology of normal tissues in 2012. He has a strong background in stem cell biology and radiation biology. His lab showed novel mechanisms and developed novel tools to investigate normal tissue responses using tissue and tumor derived organoids. Next to this, his lab developed a stem cell therapy to treat radiation induced xerostomia. He serves as editor or scientific board member of several international journals, society and institutes. In 2015, he received the Bacq-and-Alexander Award of the European Radiation Research Society which is given to an outstanding European researcher to recognize achievements in the field of radiation research.

George Cotsarelis, MD

Dr. Cotsarelis is the Milton B. Hartzell Professor and Chair of the Department of Dermatology at the University of Pennsylvania Perelman School of Medicine. His research focuses on hair follicle stem cells, alopecia and skin regeneration. He has published numerous high-impact research manuscripts, served on an array of grant review panels, and presented numerous invited lectures throughout the U.S. and abroad. Dr. Cotsarelis has held research support from the National Institutes of Health for over twenty-five years. Dr. Cotsarelis' patients with hair loss disease inspire his research, and Dr. Cotsarelis is a world renowned clinical and research expert in alopecia. He has received numerous awards, including the Society of Investigative Dermatology's Montagna lecturer, the American Academy of Dermatology's Marion Sulzberger Award and Kligman/Frost award, and the Mancunian Skin Club's Lifetime Achievement Award, and he has an honorary degree from China Medical University.

PARTICIPANTS

Rena N. D'Souza, DDS, MS, PhD

Dr. Rena D'Souza is the Director of the National Institute of Dental and Craniofacial Research, National Institutes of Health. She is deeply committed to the organization's mission — advance fundamental knowledge about dental, oral, and craniofacial health and disease and translate these findings into prevention, early detection, and treatment strategies that improve overall health for all individuals and communities across the lifespan. Dr. D'Souza oversees the institute's annual budget of approximately $485 million, supporting basic, translational, and clinical research. Prior to becoming NIDCR's director, Dr. D'Souza served in the position of Associate Vice Provost for Research for the University of Utah and as Assistant Vice President for Academic Affairs and Education for the Health Sciences. She held the Ole and Marty Jensen endowed chair in the School of Dentistry where she served as inaugural dean. As a clinician-scientist, D'Souza has been strongly committed to discovery and mentoring throughout her academic career. Dr. D'Souza is active on several trans-NIH committees and maintains an active research laboratory in the National Institute of Child Health and Human Development (NICHD), NIH.

Michele De Luca, MD

Michele De Luca is a Full Professor of Biochemistry, Director of the Centre for Regenerative Medicine "Stefano Ferrari" at the University of Modena and Reggio Emilia and Scientific Director and founder of the university spin-off Holostem, the first biotech entirely devoted to development of ATMPs for cell and gene therapy based on epithelial stem cells. He received his MD at the University of Catania and his specialization in endocrinology at the University of Rome. De Luca is author of over 130 peer reviewed international publications, has been invited speaker in over 350 international meetings and seminars and has received numerous international awards, including The New York Academy of Science & Takeda Pharmaceuticals Innovator in Science Award, Eurordis Black Pearl Award, ISSCR Innovation Award, ISSCR Public Service Award and Louis-Jeantet Prize. He is a leading scientist in the field of epithelial stem cell biology aimed at clinical application in regenerative medicine and played a pivotal role in epithelial stem cell-mediated cell and gene therapy.

Vincent Falanga

Dr. Vincent Falanga is Emeritus Professor of Dermatology and Biochemistry at the Boston University School of Medicine (BUSM). Dr. Falanga's research career began in the fields of molecular and cell biology, evolving over the years to groundbreaking human-based translational research. He made seminal contributions to the expansion and growth of single cells in low oxygen tension and to the effects and transcriptional regulation of transforming growth factor-ß 1 (TGF-ß1). Dr. Falanga later focused his efforts on human recombinant growth factors and was the first to use a recombinant growth factor (EGF) in human non-healing wounds. He showed that systemic anabolic steroids can heal the painful skin ulcers due to cryofibrinogenemia and that doubling the dose of systemically administered pentoxifylline will heal venous ulcers. In 1998, he was the lead author in the use of living bioengineered skin in non-healing wounds, which led to the first ever FDA approval of bioengineered skin for accelerating wound closure. In 2007, he was the first to successfully use autologous cultured bone marrow-derived mesenchymal stem cells in human chronic wounds. He continues to be active in the field of normal and impaired wound healing.

PARTICIPANTS

Francesca Fusco

Dr. Francesca Fusco is a researcher in the Human Molecular Genetics Laboratory at the Institute of Genetics and Biophysics of the National Research Council in Naples, Italy. Most of her scientific activity has been dedicated to the study of genetic aspects of human x-linked disease (incontinentia pigmenti, IP) by focusing on genomic instability, on the role of modifier genes and on transcriptional regulation of gene diseases. She is involved in the project on the role of genomic background in the severe phenotypes of IP patients, to find genetic variants acting as modifier genes. The produced results could be useful to anticipate the outcome of the IP disease. Previously, she was focused on characterization of complex genomic recurrent and non-recurrent rearrangements of NEMO/IKBKG locus, causing IP disease. She studied how the DNA sequence structure of this locus makes it a hot spot for genome instability, triggering specific mechanisms causing the NEMO/IKBKG mutations, producing a patent on one-step kit for IP molecular diagnosis. She contributed to the establishment of the first and the only Incontinentia Pigmenti Genetic Biobank (IPGB) of the BBMRI of which she is currently the Biobank Director.

Spiro Getsios, PhD

Spiro is the Vice President of Tissue Engineering at Aspect and manages all partnered and internal tissue engineering programs. Spiro was trained as a reproductive and developmental biologist at the University of British Columbia prior to pursuing a postdoctoral fellowship in cell biology at Northwestern University, Feinberg School of Medicine in Chicago, Illinois. At Northwestern, Spiro rose to the rank of a tenured Associate Professor of Dermatology and Director of Skin Tissue Engineering while securing federal and industrial funding for his team's research on cell-cell adhesion and communication systems. In 2016, Spiro joined GlaxoSmithKline Pharmaceuticals as a Director of Biology with a focus on delivering preclinical models for drug discovery and development. He is an author on more than 60 research publications.

Encarna Guillen-Navarro

Professor Encarna Guillén-Navarro is pediatrician and clinical geneticist. She is Head of Medical Genetics Section, Pediatric Department at the Virgen de la Arrixaca University Hospital (Murcia, Spain), Professor of Medical Genetics and Pediatrics at the University of Murcia, Principal Investigator of the Research Group on Rare Diseases and Clinical Genetics of the Murcia Institute of Biosanitary Research (IMIB-Arrixaca) and in CIBERER-ISCIII. During the two years she was the Murcia Health Minister, she developed a comprehensive care and research program for rare diseases in Murcia, Spain. At the European level, she has defended the digital transformation of health and the personalized medicine strategies for better prevention and care of rare diseases. She is scientific advisor in national and international rare diseases federations (FEDER, ALIBER, EURORDIS) and patient advocacy groups, such as AADE (Spanish Association of Ectodermal Dysplasia). She organized the 7th International Conference on Ectodermal Dysplasias in 2017, and with consecutive competitive public grants and a multidisciplinary approach, including patients and families. She is leading the ectodermal dysplasias research in Spain.

PARTICIPANTS

Mark Kiel, MD, PhD

Dr. Mark Kiel is Co-Founder and Chief Science Officer at Genomenon, where he oversees the company's scientific direction and product development. After spending 15 years preparing for a life of academic research, Mark became convinced that revolutionary change in genomics was more likely to emerge out of industry. In 2014, he founded Genomenon - an AI-driven genomics company addressing the challenge of connecting pharma researchers with evidence in the literature to help diagnose and treat patients with rare genetic diseases and cancer.

Ophir Klein, MD, PhD

Ophir Klein is Professor of Orofacial Sciences and Pediatrics, the Larry L. Hillblom Distinguished Professor in Craniofacial Anomalies, and the Charles J. Epstein Professor of Human Genetics at the University of California, San Francisco (UCSF). He serves as Chief of the Division of Medical Genetics, Chair of the Division of Craniofacial Anomalies, Medical Director of the Craniofacial Center, Director of the Program in Craniofacial Biology, and interim Director of the Institute for Human Genetics. He attended Yale University School of Medicine, where he received a PhD in genetics and his MD. He then completed residencies at Yale-New Haven Hospital in pediatrics and at UCSF in clinical genetics. Dr. Klein's research focuses on understanding how organs form in the embryo and how they regenerate in the adult, with a particular emphasis on the processes underlying craniofacial and dental development and renewal as well as understanding how stem cells in the intestinal epithelium enable renewal and regeneration.

Smail Hadj-Rabia

Smail Hadj-Rabia is a doctor in the Department of Dermatology of the Hôspital Necker- Enfants Malades, APHP, Imagine Institute, in Paris, France. He is involved in care and research in rare skin diseases mostly with ectodermal dysplasias. He is a founding member of MAGEC, a French Center of Expertise for Rare Skin Diseases. He is an active partner of the Gendodermatoses Network, the European and Mediterranean Network on rare skin diseases and the ERN-skin Network. Together with Holm Schneider, he coordinates the subthematic group of ectodermal dysplasias in the ERN Network. More recently, he described a novel form of ectodermal dysplasia called HELIX and identified Claudin-10 as the disease-causing gene. In collaboration with D. Aberdam, he has tested a topical treatment for skin erosions in ankyloblepharon-ectodermal defects-cleft lip and/or palate (AEC) syndrome patients.

PARTICIPANTS

Maria Kousi, PhD

Maria started her genetics training at the Aristotle University of Thessaloniki, Greece and subsequently moved to the University of Helsinki, Finland where she obtained my MSc and PhD in human genetics. Her doctoral work focused on the genetic dissection of childhood-onset neurodegenerative diseases, leading to the molecular genetic diagnosis of approximately 100 patients and paving the way for prenatal screening in affected families. During her post-doctoral studies at Duke University, she focused on the dissection of pediatric-onset neurodegenerative and neurodevelopmental disorders, through the combinatorial implementation of genetic studies with in vivo and in vitro functional assays to dissect the biological effect of patient-specific variation. In parallel, she was a core member within the Task Force for Neonatal Genomics (TFNG), a project designed to provide genetic diagnoses through whole exome sequencing to fetuses and infants with clinically challenging to diagnose debilitating genetic conditions. Within TFNG, she led the efforts focusing on the diagnosis of neurological and neuromuscular disorders. In 2018, she became jointly appointed at Harvard University and MIT, expanding her work on functional genomics of neurodegenerative diseases using patient iPSC-derived cortical neuronal cultures. In 2020, she joined Third Rock Ventures where she is working towards creating new companies that use genetics to provide clinical diagnoses, shorten diagnostic odysseys and where possible intercept disease through data-driven individualized therapies.

Josephine Lampe, MS

Josephine Lampe earned her bachelor's and master's degree in human biology at the University of Greifswald (Germany) and is currently working as a PhD student in the Department of Pharmacology and Toxicology at the University of Lübeck. Her research focuses on the blood-brain barrier with a special interest in incontinentia pigmenti.

Fabio Mammano

Fabio Mammano graduated with a degree in physics at the University of Parma and specialized in biophysics at the International School of Advanced Studies (Sissa) in Trieste. He did his post-doc in Great Britain, in the Department of Physiology, University of Bristol, and then returned to Italy in 1995 as a university researcher at Sissa. From 2014 to 2018, he directed the Institute of Cell Biology and Neurobiology of the National Research Council (CNR). He is currently a full Professor of Applied Physics at the Department of Physics and Astronomy of the University of Padua and visiting professor at the Shanghai Institute for Advanced Immunochemical Studies of Shanghai Tech University. He also coordinates a research group at the CNR at the Institute of Biochemistry and Cell Biology and is the MUR / CNR National Delegate for the INFRAFRONTIER / IMPC distributed research infrastructure. For 30 years, Professor Mammano has been studying a class of proteins, the connexins, whose alterations cause more or less severe forms of deafness and skin diseases. His research is mainly aimed at finding innovative therapies for the treatment of these diseases.

PARTICIPANTS

M. Peter Marinkovich, MD

M. Peter Marinkovich, MD, is Associate Professor of Dermatology, and a founding member of the Stanford Program in Epithelial Biology. He is Director of the Stanford Bullous Disease Clinics. He has a longstanding interest and expertise in basement membrane biology, as well as the pathophysiology, clinical features, and therapeutic development of new molecular therapies for epidermolysis bullosa.

Mª Carmen Martínez-Romero, PhD

Mª Carmen Martínez-Romero, PhD is biologist and works as a molecular geneticist. Currently she is a laboratory consultant of Center for Biochemistry and Clinical Genetics at the Virgen de la Arrixaca University Hospital (Murcia, Spain. She teaches genetics and biotechnology at the Catholic University of Murcia UCAM and is a research collaborator of the Group on Rare Diseases (RD) and Clinical Genetics of the Murcia Institute of Biosanitary Research (IMIB-Arrixaca) and in CIBERER-ISCIII. She developed her doctoral research in the molecular characterization and phenotypic correlation of Spanish patients affected by ectodermal dysplasias. She actively collaborates in competitive public projects (funded by ISCIII, Spanish Ministry of Economy and Competitiveness, PI14/01259, PI17/00796, PI21/01082 and co-funded with FEDER). She has experience in the field of prenatal and postnatal genetic diagnosis, being a promoter of the implementation new genomic technologies at the lab and responsible for the quality management programs of the Center for Biochemistry and Clinical Genetics.

Gina Mawla, PhD

Dr. Gina Mawla is a postdoctoral scholar in the Department of Molecular, Cell and Developmental Biology in Dr. Olena Vaske's lab at the University of California, Santa Cruz (UCSC). She is a member of the Treehouse Childhood Cancer Initiative at the UCSC Genomics Institute, founded and led by Dr. Vaske. Dr. Mawla's research focuses on leveraging and applying information gained from genomic and transcriptomic sequencing techniques to inform the specific diagnoses and treatment plans of patients afflicted by rare pediatric diseases. She is currently developing novel experimental and high-throughput analysis approaches to discover and functionally characterize new druggable molecular targets of pediatric cancers. Dr. Mawla holds a PhD in biology from the Massachusetts Institute of Technology and a BS in molecular, cell and developmental biology from the University of California, Santa Cruz. She is a recipient of the Institutional Research and Academic Career Development Award (IRACDA), which recognizes her interest in and commitment to teaching and pedagogy in addition to research.

PARTICIPANTS

Sarah E. Millar, PhD

Sarah E. Millar, PhD is Director of the Black Family Stem Cell Institute, and Lillian and Henry M. Stratton Professorial Chair in the Departments of Cell, Developmental and Regenerative Biology and Dermatology at the Icahn School of Medicine at Mount Sinai. After graduating from Cambridge University, Dr. Millar received her PhD from the University of London and then trained as a post-doctoral fellow at the National Institutes of Health and at Stanford University. She is a graduate of the Executive Leadership in Academic Medicine (ELAM) program. Dr. Millar is internationally recognized as a leading researcher in epithelial biology. Her research group has made seminal discoveries on the roles of Wnt signaling and epigenetic regulatory mechanisms in development and regeneration of the skin and its appendages. She is an Editorial Board member for Developmental Cell and Experimental Dermatology, a Deputy Editor for the Journal of Investigative Dermatology, and has served on numerous national and international grant review committees.

Jacques Monnet, MBA

Jacques Monnet has been the president for 13 years of Incontinentia Pigmenti France, a non-profit organization dedicated to families affected by that condition. IP France has founded research projects in France, Italy and Germany. He has more than 40 years of experience in the pharmaceutical and vaccine industry, with international expertise in financial performance, project management and strategic studies. He earned his MBA at ESSEC Business School in Paris, France.

Maria Morasso, PhD

Dr. Maria Morasso received her PhD in biochemistry and molecular biology from the Instituto Venezolano de Investigaciones Cientificas (IVIC), Caracas Venezuela. There, she worked to study cell adhesion in melanoma metastasis. She did postdoctoral training at the National Institute of Child Health and Human Development (NICHD), National Institutes of Health, where she established her studies on epidermal specification during development. Dr. Morasso was recruited as a tenure-track investigator to the Laboratory of Skin of the National Institute of Arthritis and Musculoskeletal and Skin Diseases (NIAMS) to create the Developmental Skin Biology Unit. She is currently a senior investigator and laboratory chief of the Laboratory of Skin Biology (LSB) in NIAMS, NIH. Dr. Morasso is also an adjunct investigator at the Center for Cancer Research, National Cancer Institute (NCI). Research in the LSB has focused in understanding the roles of transcriptional regulators in skin differentiation, barrier formation, and wound healing. Another emphasis has been the study of the regulatory pathways controlling the development of ectodermal appendages (hair and tooth) and the molecular mechanisms underlying ectodermal dysplasias.

PARTICIPANTS

Dimitry Ofengeim, PhD

Dimitry Ofengeim, PhD is the head of Precision Neurology and Neuroinflammation in the Rare Neuroscience Research Therapeutic Area at Sanofi, based in Framingham, Massachusetts. He leads a team of scientists focused on understanding the role of neuroinflammation and the CNS innate immune system in multiple sclerosis and other neurodegenerative diseases. Dr. Ofengeim is an expert in mechanisms of acute and chronic neurodegeneration, and in particular has focused on understanding the interplay between cell death and inflammation in the CNS. His work was among the first to show that a novel form of cell death, called necroptosis, occurs in neurodegenerative diseases including MS, ALS, and Alzheimer's disease. Dr. Ofengeim's recent work has advanced understanding of disease-associated microglial states and functions. Prior to coming to Sanofi, he worked for two years at Biogen in the ALS and MS groups. He was awarded several fellowships from the National MS Society during his time at Harvard, including a postdoctoral fellowship and a career transition fellowship. He received his PhD at the Albert Einstein College of Medicine.

Alessandra Pescatore, PhD

Alessandra Pescaore completed her biology degree at the University of Naples "Federico II" in 2006 and her PhD in 2010, analyzing how newly found missense NEMO mutations cause incontinentia pigmenti (IP). In collaboration with the group of Dr. Gilles Courtois, INSERM (Paris), they provided insights into the function of NEMO in the NF-κB activation process (2006-2008). A fellowship from France Incontinentia Pigmenti Association allowed her to do a post-doc with Prof. Matilde Valeria Ursini, IGB-CNR Naples from 2011-2016, where she focused on cell death mechanisms and in particular, the role of NEMO in regulating cell death and IP. An EMBO-fellowship allowed her to establish a valuable collaboration with Pr. H. Walczak (2013, London UK), whose research was specifically focused on the biology of death receptor-ligand and the ubiquitin systems. After one year of teaching-experience at University of Baronissi, Salerno (2017-2018), she returned to the IGB-CNR (2019) as a permanent researcher. She focuses the cell death pathways and their intersection with IP and embryonic development.

Yorick Post, PhD

Dr. Yorick Post is a stem cell biologist with a PhD degree from the University of Utrecht in The Netherlands. Dr. Post conducted his graduate research in the laboratory of Hans Clevers at the Hubrecht Institute focusing on adult stem cell biology and organoid technology. In this role, he published research articles on heart regeneration and the establishment of organoid models. Organoids are self-organizing mini-organs that can be used to study physiological and pathological processes of a tissue of interest. Prior to this work, Dr. Post gained experience in molecular oncology, conducting research at the Netherlands Cancer Institute in Amsterdam and the Whitehead Institute part of MIT, earning a scholarship from the Dutch Cancer Society. Currently, he is a scientist at a biotechnology company, Surrozen, in South San Francisco, focusing on the Wnt pathway in injury and disease.

PARTICIPANTS

Philip Reilly, MD, JD

Phil Reilly focuses on creating and building companies that develop breakthrough therapies for orphan genetic diseases. He is a Venture Partner in Third Rock Ventures. He is passionate about working with patient groups, and helping to unite those suffering from rare diseases in order to foster a support network and potentially improve patient outcomes. Phil has authored or co-authored more than 100 articles in scholarly journals and has published seven books including most recently, Orphan: The Quest to Save Children with Rare Genetic Disorders. Phil currently serves on the Advisory Board to the Boston University School of Public Health, and has served as a trustee emeritus of Cornell University, and an Overseer of Weill Cornell Medical College.

Angela Scheuerle, MD

Dr. Scheuerle received her MD from The University of South Florida Morsani College of Medicine. She has a specific interest in craniofacial malformations and has practiced clinical genetics in Texas since 1995. From 2003 to 2014, she was in private practice at Tesserae Genetics at Medical City Dallas. She has been adjunct faculty at UT Southwestern in the Program of Ethics in Science and Medicine and the Eugene McDermott Center for Human Development since 2000. She joined the Pediatric Division of Genetics and Metabolism as full faculty in September 2014. She holds secondary appointments as Professor in the McDermott Center and in the Department of Pathology, where she is also co-director of the Fellowship in Laboratory Genetics and Genomics. Dr. Scheuerle has also worked with the Texas Department of State Health Services Birth Defects Epidemiology and Surveillance Branch since 1994. In 1997, Dr. Scheuerle joined the Antiretroviral Pregnancy Registry (APR), where she serves as teratologist and on the scientific advisory and steering committees.

Holm Schneider, MD

Dr. Holm Schneider obtained his MD from the University of Leipzig, Germany, and received German board certifications in general pediatrics, emergency medicine, and neonatology. He completed a long-term research fellowship at Imperial College London, U.K., established an independent research group at the University Hospital Erlangen, Germany, and was professor of experimental neonatology at Innsbruck Medical University, Austria, before returning to Erlangen in 2008. Since then, he has been working as a professor of pediatrics and consultant pediatrician at the University Hospital Erlangen, where he also heads the interdisciplinary Center for Ectodermal Dysplasias Erlangen (ZEDER), the German national reference center for ectodermal dysplasias and p63-associated disorders. Current research of his team is dedicated to specialized care and novel therapeutic approaches to severe genodermatoses. Dr. Schneider pioneered the prenatal drug therapy of genetic diseases. He conducted numerous preclinical and investigator-initiated clinical studies focusing on life-threatening congenital skin disorders. He has continued his pediatric practice while pursuing laboratory research and clinical trials and has been serving on medical advisory boards of various patient organizations for 15 years.

PARTICIPANTS

Pascal Schneider, PhD

Pascal Schneider is a senior lecturer and researcher in the Department of Biochemistry at University of Lausanne, Switzerland. He obtained a PhD there and performed post-docs at the University of Dundee, Scotland, and at the University of Lausanne. Ligands and receptors of the TNF family, in particular BAFF, APRIL and EDA, are his major research interests, with a focus on biochemical aspects of their functions.

Markus Schwaninger

Dr. Markus Schwaninger 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.

Harold C. Slavkin, DDS

Dr. Slavkin is Professor and Dean Emeritus at the Herman Ostrow School of Dentistry, University of Southern California. In June 2014, he retired after 46 years on the full-time academic faculty. He served as Dean of the dental school (2000-2008). From 1995-2000, he served as Director of the National Institute of Dental and Craniofacial Research (NIDCR) within the National Institutes of Health. One of the major accomplishments of his tenure at NIDCR was his leadership and the completion of the first ever Surgeon General's Report to focus exclusively on craniofacial, oral and dental health. His interests include "precision healthcare", health care policy (children and "mature senior adults"), craniofacial molecular biology, human genetics, developmental biology and aging, biomaterials, tissue engineering and regeneration, human and microbial genomics, immunity and inflammation, and precision dentistry and medicine as reflected within 520 peer-reviewed scientific publications and 91 contributed chapters to various books in biomedical research. He also authored Craniofacial Developmental Biology (1979), Birth of a Discipline: Craniofacial Biology (2012), and a novel "Atlanta" (2012).

PARTICIPANTS

Matilde Valeria Ursini

Matilde Valeria Ursini is a CNR Research Director and group leader of Human Molecular Genetics Laboratory at the Institute of Genetics and Biophysics (IGB) “A. Buzzati-Traverso” CNR in Naples, Italy and professor of human genetics. Prior, she worked in genetics for 30 years at the Lyon I University in France and at the Federico II University in Naples. Most of her scientific activity has been devoted to the discovery of the genetic basis of rare human diseases. Most recently, she studied the molecular basis of incontinentia pigmenti (IP), caused by a mutation in the NEMO/IKBKG gene. She studied IP with a classical genetic approach coupled with the study of the multiple roles of NEMO protein in the cell homeostasis. The use of cells expressing the IP associated NEMO mutant has revealed how alteration of cell death process may result in different cellular outcomes and pathological effects in IP patients. She has published 35 papers and patented a one-step kit for IP molecular diagnosis. She established the first word-wide Incontinentia Pigmenti Genetic Biobank and is the coordinator of the Biological Research Centre of the IGB belonging to the BBMRI-ERIC network.

Rui Yi, PhD

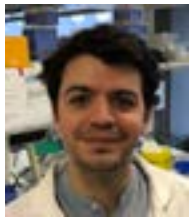
Rui Yi is the Paul E. Steiner Research Professor of Pathology at Northwestern University Feinberg School of Medicine. Dr. Yi studies mechanisms that govern cell fate specification, stem cell maintenance, aging and diseases by using genetically engineered mouse models, innovative single-cell genomic tools and intravital imaging. Currently, he is using precise genome editing to generate novel p63 mutations that cause human ectrodactyly-ectodermal dysplasia-cleft lip/palate (EEC) syndrome and applying single-cell genomic tools to examine gene regulatory networks affected by these p63 mutations during skin development and hair morphogenesis. He earned his PhD at Duke University.

Matija Zelic, PhD

Matija Zelic is a cellular and molecular biologist with expertise in innate immunity, neuroinflammation and cell death research. He received his PhD from the University of Massachusetts Medical School in Worcester, MA under the mentorship of Dr. Michelle Kelliher. During his doctoral studies, Matija focused on the role of RIPK1 and its kinase activity in inflammatory signaling and apoptotic and necroptotic cell death. He identified a role for RIPK1 kinase-mediated necroptotic death of liver endothelial cells contributing to vascular break-down and lethality in a mouse model of TNF-induced SIRS. In 2018, Matija joined the precision neurology and neuroinflammation group under Dr. Dimitry Ofengeim as a Postdoctoral Fellow at Sanofi. He has continued his work as a scientist in the Rare & Neurologic Disease Research Therapeutic Area, where he recently identified key roles of RIPK1 kinase activity in microglia and astrocytes in the context of multiple sclerosis (MS) disease progression. His research is focused on identifying and modulating cellular pathways leading to beneficial versus deleterious inflammatory signaling and death in glial cells in the context of neuroinflammation and neurodegenerative diseases (ALS, MS).

NEW CAREER INVESTIGATORS

The NFED is proud to have provided grants to the following researchers to attend the International Research Conference.



Jeremie Rosain, PhD

Jérémie Rosain, an immunologist from clinical laboratories who trained at Necker Hospital for Sick Children in Paris, France. His hospital training focused on the immunological and genetic investigations of patients with primary immunodeficiency, including patients with ectodermal dysplasia with immunodeficiency. In 2018, he joined the laboratory of Jean-Laurent Casanova for his PhD. With his colleague, Paul Bastard, he has been investigating autoantibodies to type I interferons in patients with life-threatening COVID19. He is now more focused on studying and deciphering the pathophysiology of high prevalence of these autoantibodies in individuals living with incontinentia pigmenti.

Stefano Sol, PhD

Stefano Sol earned his PhD in biology at the University of Naples Federico II, under the supervision of Prof. Caterina Missero. During his PhD, he focused on the transcriptional mechanisms and genetic pathways required for normal skin development and function that are altered in inherited and acquired skin diseases. He studied the role of transcription factor p63, a master regulator of stratified epithelia, in ankyloblepharon-ectodermal defects-cleft lip and/or palate (AEC) syndrome and developed novel therapeutic strategies to rescue epidermal defects. Thanks to an “IBSA Foundation Fellowships Call 2018”, financed by IBSA Foundation, in 2018, he collaborated with Dr. Anna Mandinova at Massachusetts General Hospital on the identification of promising chemical compounds able to inhibit mutant p63 aggregation that the lab has recently identified. Currently, his work is focusing on assessing the efficacy and long-term safety of a drug-therapy for AEC syndrome.

Ezia Spinosa

Ezia Spinosa earned her master’s degree in biotechnology for medical, pharmaceutical and veterinary diagnostics with 110 / 110L at the University of Basilicata. She worked for her master’s thesis at the Institute of Genetics and Biophysics “A. Buzzati-Traverso” CNR of Naples in the group of MV Ursini and F Fusco. Her thesis was titled “Genotype-Phenotype Correlation in the Genetics Rare Disease Incontinentia Pigmenti (IP)”. In February 2020, she was awarded a CNR fellowship is currently a fellow at IGB in the same group. She is currently participating in the biobank project. Specifically, she takes care of accompanying the sample from arrival at the biobank to molecular diagnosis and storage. Her research is aimed at defining the genotype of IP patients. She sequences both the individual exons of the NEMO/IKBKG gene and does the sequencing and analysis of the entire exome and transcriptome of patients as part of a larger project focused on the defining the genotype -phenotype correlation in IP.

STUDENT WRITERS

Jessica Gugger

Jessica Gugger is a graduate student in the biomedical sciences program at The Brody School of Medicine at East Carolina University and is completing my master's thesis research in the laboratories of Drs. Maranke Koster and Peter Koch. She graduated from East Carolina University in 2020 with a bachelor's degree in biology. Her research is focused on investigating the molecular mechanisms responsible for causing the skin fragility experienced by patients affected by ankyloblepharon-ectodermal defects-cleft lip and/or palate (AEC) syndrome. She primarily focuses on analyzing the expression of genes involved in cell-cell and cell-ECM [extracellular matrix] adhesion in patient-derived keratinocytes. Their team hypothesizes that the abnormal expression of these genes is responsible for the skin erosions and hope that identifying the underlying mechanisms responsible will lead to a therapy for these individuals.

Shirley Parraga

Shirley Parraga graduated from the University of North Carolina at Charlotte in 2018 with a double major in biology and psychology. She is a medical student and research scholar at the East Carolina University Brody School of Medicine. She works alongside Dr. Maranke Koster and Dr. Peter Koch in the Department of Anatomy and Cell Biology. Her research involves analyzing the effects of ankyloblepharon-ectodermal defects-cleft lip and/or palage (AEC) and ectrodactyly-ectodermal dysplasia-clefting (EEC) TP63 Mutations on keratinocyte biology. Their hypothesis is that skin fragility in AEC and EEC patients is caused by cell adhesion defects. They are conducting experiments to compare expression and localization of cell adhesion and cytoskeletal proteins in keratinocytes that express TP63 mutations linked to AEC and EEC syndromes.

Maddison Salois

Maddison Salois is second-year PhD student at East Carolina University in the laboratories of Dr. Maranke Koster and Dr. Peter Koch. She is a trained embryologist and cell culturist. She is enthused to attend this conference!

Amanda Swanson

Amanda K. Swanson is a third year DDS student at the University of North Carolina Adams School of Dentistry. She received her undergraduate degree in biology and chemistry from Ohio Northern University. Amanda is currently studying under the mentorship of Dr. Tim Wright as a 2021-2022 fellow of the American Dental Education Association (ADEA) Academic Dental Careers Fellowship Program. Through this involvement, she is eager to continue learning about hereditary developmental dental defects and other syndromic conditions, as well as how research across disciplines can be utilized to advance patient care. She is currently working on a project in collaboration with endocrinologist Dr. Janet Rubin about the clinical presentation of tricho-dento-osseous syndrome and the subsequent importance of obtaining a genetic diagnosis.

FAMILY PANEL

BECKY ABBOTT
x-linked hypohidrotic ectodermal dysplasia (XLHED)

JACQUES MONNET
incontinentia pigmenti (IP)

RUTH GEISMAR
ankyloblepharon-ectodermal defects-cleft lip and/or palate (AEC) syndrome

KARL NELSEN
unknown subtype of ectodermal dysplasia

CONFERENCE ORGANIZING COMMITTEE

Becky Abbott

Angus Clarke

Mary Fete

Mark Kiel

Maranke Koster

Karl Nelsen

Holm Schneider

Tim Wright

Thank You…

…Tim Wright and Maranke Koster for your leadership and passion to make a difference in the lives of families affected by ectodermal dysplasias.

…NFED staff, especially Becky Abbott and Kayla Hollenkamp, for your countless hours of work to make this conference a landmark event.

…generous sponsors for underwriting the conference and investing in a brighter tomorrow.

…participants for sharing your time and expertise to help us chart a course for ectodermal dysplasias research.

Our shared commitment to inspiring investigators to study these rare conditions, advancing research and developing new and improved treatments gives our NFED family incredible hope.

Together, we will accomplish extraordinary things!

– Mary Fete

CHAMPION SPONSOR

THE LOUIS & GLADYCE FOSTER FAMILY FOUNDATION

The Louis & Gladyce Foster Family Foundation was established in 2011 to help support charitable causes important to Louis and Gladyce Foster and to support missions especially important to each new generation of the Foster and Witt families.

When Craig & Lisa Witt discovered that their son, Payton, had ectodermal dysplasia, they had never heard of it and didn't know where to turn. Fortunately, they found the NFED which provided answers and compassion during a very difficult time. They immediately knew that they wanted to support the NFED to help others & make a difference.

"My goal as a father is to be as supportive to my kids as I can. As a foundation trustee, my job is to provide opportunities for others and their families to feel supported as well. The goal is to make a real difference in the lives of those with ectodermal dysplasia, now and into their future!"

- Craig Witt, father and foundation trustee

CHAMPION SPONSOR

The Hubbard Broadcasting Foundation
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Pierre Fabre Group is a French company with 60 years of pharmaceutical expertise, inventor of dermo-cosmetics. Highly involved in oncology and dermatology, Pierre Fabre is also committed in XLHED according to its values and patient-centered commitments, summarized in its corporate purpose:

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EspeRare

As a not-for-profit drug developer committed to finding solutions for pediatric rare diseases, EspeRare fully understands and relies on the central role that Patients Organisations play in advancing tomorrows' treatments.

EspeRare is grateful to count the NFED as a key partner in their XLHED programme, and is delighted to be able support the NFED's international conference.

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