

Wednesday, June 23

7:30 – 10 pm **Poster Session - Welcome Reception** (Horizons Ballroom 9 – 11)

Thursday, June 24

7:00 – 8:00 am **Breakfast Buffet** (Horizons Ballroom 9 – 11)

8:00 - 8:15 am **Welcome** (Horizons Ballroom 2 – 4)
David Scholl, PhD (President of FIRST & Diagnostic Hybrids) &
Leonard Milstone, MD (Yale University)

8:15 - 10:15 am **Frontiers in Genetic Diagnosis** (Horizons Ballroom 2 – 4)
Judith Fischer, PhD (Centre National de Genotypag) - Next generation sequencing and gene
identification in ichthyosis

Eli Sprecher, MD, PhD (Tel Aviv Sourasky Medical Center) - New Genodermatoses in the 21st
century: challenges and promises

Keith Choate, MD, PhD (Yale University) - Somatic genetic events facilitate identification of
disease-causing genes

Amy Paller, MD (Northwestern University) - Should every individual with ichthyosis be given a
genetic diagnosis?

10:15 - 10:30 am **Break** (Horizons Salon 1)

10:30 - 12:00 pm **Frontiers in Understanding Pathogenesis I** (Horizons Ballroom 2 – 4)
William Rizzo, MD (University of Nebraska) - Lipid pathways in the pathogenesis of ichthyosis

Michael Fitzgerald, PhD (Harvard Medical School) - Lipid transport in ichthyoses: Analysis of
ABCA12 molecular function

Peter Elias, MD (University of California San Francisco) - Treatment of the Ichthyoses Based Upon
Pathogenesis of the Cutaneous Phenotype

12:00 - 1:00 pm **Lunch** (Lakeview Restaurant East, Mezzanine Level)

FRONTIERS IN ICHTHYOSIS RESEARCH PROGRAM

Regal Sun Resort
1850 Hotel Plaza Boulevard
Lake Buena Vista, FL 32830



Thursday, June 24 (cont'd)

- 1:00 - 2:30 pm **Frontiers in Understanding Pathogenesis II** (Horizons Salon 2 – 4)
Alain Hovnanian, MD, PhD (University Paris) - From gene to function: Netherton syndrome as a disease model to understand the role of protease regulation in skin allergy

Pierre Coulombe, PhD (Johns Hopkins University) - Non-canonical functions of keratins

Hiroshi Shimizu, MD, PhD (Hokkaido University) - Skin cancer in ichthyosis
- 2:30 - 2:45 pm **Break** (Horizons Salon 1)
- 2:45 - 5:15 pm **Frontiers in Shared Reagents/Resources** (Horizons Salon 2 – 4)
Mary Williams, MD (University of San Francisco) - Biomarkers for clinical trials

Suephy Chen, MD (Emory University) - Burden of Ichthyosis

Robert Rice, PhD (UC Davis) - Proteomic analysis of epidermal scale

Dennis Roop, PhD (University of Colorado) - Stem cell therapy: a shared or personal resource?

Roger Kaspar, PhD (Transderm, Inc.) - Improving models for pre-clinical testing
- 6:30 pm **Meet in Lobby for Dinner** (Napa Room in Contemporary Resort; transportation provided)
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Friday, June 25 Patient-Investigator Colloquium

- 7:00 – 8:00 am **Continental Breakfast** (Horizons Salon 1)
- 8:00 - 9:30 am **Frontiers in Preparing for Clinical Trials** (Horizons Salon 2 – 4)
Heiko Traupe, MD (University Hospital, Muenster) - The long way from molecular insights to therapeutic innovation

Sancy Leachman, MD, PhD (University of Utah) - Preparation for clinical trials with siRNAs

Philip Fleckman, MD (University of Washington) - Making Registries More Useful – the US Ichthyosis Registry
- 9:30 - 9:45 am **Break** (Horizons Salon 1)



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Friday, June 24 (cont'd)

9:45 - 11:00 am **Frontiers in physician/scientist/patient collaboration** (breakout groups)

CIE – ARCI (Autosomal Recessive Congenital Ichthyosis) (Bay Lake Room)

Moderator – William Rizzo, MD

FIRST Ambassadors – Holly Friddle, Janet & Maggie McCoy

Epidermolytic Hyperkeratosis and other Keratinopathies (Spring Lake Room)

Moderator - Sancy Leachman, MD, PhD

FIRST Ambassadors – Mark, Kelly & Adam Klafter, Ryan Licursi, John Schoendorf,

Harlequin Ichthyosis (Highland Room)

Moderator: Philip Fleckman, MD

FIRST Ambassadors – Mark, Shannon & Lauren Hamill, Hunter Steinitz

Lamellar/Transglutaminase 1 deficiency (Park Lake Room)

Moderator – Amy Paller, MD

FIRST Ambassadors – Joe Andrews, Mark & Cora Dunkin, Laura Phillips

Netherton Syndrome (Rock Lake Room)

Moderator – Mary L. Williams, MD

FIRST Ambassadors – Mark, Wendy & Tyler Breen, Diana Gilbert, Randy LaBarbera

Undiagnosed (Sandy Lake Room)

Moderator – Keith Choate, MD, PhD

FIRST Ambassadors – Angela Godby, Brian See

11:00 - 12:00 am **Group Reports and Goals for the Future** (Horizons Salon 2 – 4)