

Frontiers in Ichthyosis Research Meeting Summary  
June 23-25 – Orlando, FL – Regal Sun Resort

Frontiers in Ichthyosis Research, an international meeting of investigators actively involved in research directly related to ichthyosis, was held June 23-25 in Orlando, FLA. The meeting immediately preceded the biannual family conference held by FIRST, the ichthyosis patient support organization. The meeting was designed to foster collaboration between investigators, and between patients and investigators. It was an opportunity for the most deeply engaged individuals to begin a dialogue about efficient and effective ways to utilize scarce resources to advance research in ichthyosis. Invited speakers were asked to present ongoing research and their perspective on broad and significant challenges and opportunities for the future.

Leonard Milstone (Yale University, New Haven, CT) introduced the meeting by noting that it was being held on the tenth anniversary of the announced completion of the human genome project. Identification of genes associated with human disease was an important spin-off from that world-wide effort and the ichthyoses were no exception. In the past twenty years we have come to recognize that the ichthyosis phenotype can be attributed to an unexpectedly large number of genes, whose coded proteins have a broad array of functions. This new perspective beautifully demonstrates that no aspect of epidermal biology can be taken for granted. It is no intellectual stretch to say that he (or she) who understands ichthyosis understands epidermal biology. This new appreciation opens the door for renewed collaboration between basic scientists interested in epidermal biology and keratinization and those interested in ichthyosis. Opportunities for epidermal science to impact the lives of others have never been so great.

Despite the rarity of ichthyosis, study of patients with ichthyosis has had a disproportionately broad impact on two areas: a) fundamental discoveries about critical skin functions; and b) development of treatments that benefit millions with more common skin diseases. It is not unreasonable to anticipate that stimulation of research in ichthyosis will continue to have significance to the entire skin disease community.

The meeting was organized into 5 sessions with a mixture of formal presentations and lively discussion: Frontiers in Genetic Diagnosis, Frontiers in Understanding Pathogenesis, Frontiers in Shared Reagents and Resources, Frontiers in Preparing for Clinical Trials, and Frontiers in physician/scientist/patient collaboration. In the final open discussion, there was enthusiasm for convening meetings of this kind in the future, possibly in different countries. Two working groups were established. One group will explore the feasibility and parameters of a collaborative international effort to establish a registry of patients with ichthyosis. The second will consider practical and efficient ways to provide genetic diagnoses to patients with ichthyosis. It was recognized that vigorous advocacy by patient support groups in many countries might be necessary to generate requisite governmental, insurance-industry, or private support to make genetic diagnosis widely available. There was broad agreement that standardized, clinical evaluation tools would be highly desirable for future investigations into the natural history or response to therapy for each type of ichthyosis. It was suggested that an area in which patient support groups could assist is in the development of visual analog, validated, widely available, genotype-specific clinical severity scales. Finally, it was generally recognized that in the face of scarce resources – limited numbers of patients, limited numbers of knowledgeable clinicians, limited numbers of preclinical scientists working on ichthyosis, and of course limited funding – progress in translating new approaches to therapy might likely require agreement to establish and coordinate centers of research/translation excellence.

Acknowledgements: The conference was sponsored by National Institute of Arthritis, Musculoskeletal and Skin Diseases, the NIH Office of Rare Diseases, and FIRST.