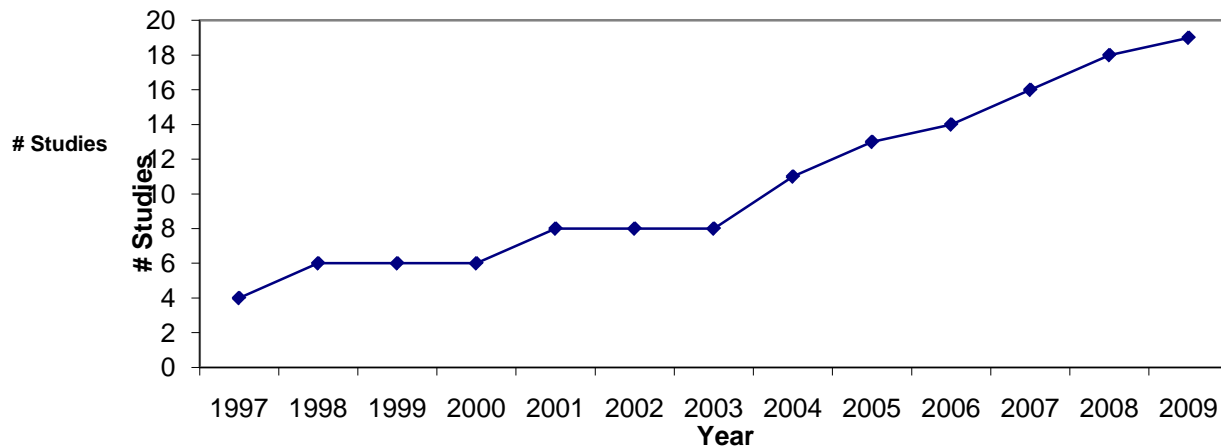


Studies Supported and Ensuing Publications - 9.1.09

Studies supported - 19

Total publications - 18

Total Studies Supported 9.09



Studies supported and ensuing publications

“Phase III Randomized, Double-Blind, Placebo-Controlled Study to Evaluate Safety and Efficacy of Glylorin™ in the Treatment of Congenital Primary Ichthyosiform Erythroderma, a Subtype of Congenital Primary Ichthyoses”

Matthew J. Stiller, MD

Date approved: 2/10/96

“Clinical and Genetic Studies of the Scaling Disorders and Other Selected Genodermatoses.” (EKV, PSEK, PPK, other erythrokeratodermias)

Sherri Bale, Ph.D.

Date approved: 2/22/96

Richard G, et. al. Mutations in the human connexin gene GJB3 cause erythrokeratoderma variabilis. Nature Genetics 20: 366-369, 1998 (with acknowledgement of Registry assistance).

“Evaluation of Barrier Function, Structure, and Lipid Content of Normal, Aged, and Ichthyotic Skin.”

Peter Elias, MD

Date approved: 10/21/96

Zettersten E, et al: Recessive x-linked ichthyosis: role of cholesterol-sulfate accumulation in the barrier abnormality. J.Invest Dermatol 111 (5):784-790, 1998 (with acknowledgement of Registry assistance).

“Quality of Life of Persons with Disorders of Keratinization”

Philip Fleckman, MD

Date approved: 10/21/96

Fleckman P, Hamill G, Weinstock MA: The National Registry for Ichthyosis and Related Disorders - Health Related Quality of Life and Patient Reported Outcomes. *J Invest Dermatol*, 121:A#420, 2003 (abstract).

“Clinical and Genetic Studies of the Scaling Disorders and Other Selected Genodermatoses” (Netherton Syndrome/Ichthyosis linearis circumflexa)

1998 -- Dr. Richard (NIH)

Continuation of 1996 Bale study, different disorder

“Predictors and Demographic Characteristics of People Using the Internet to Search for Information on Arthritis and Musculoskeletal and Skin Diseases”

Ann Taubenheim, MS, RN

Date approved: 4/3/98

Study completed (Doctoral Dissertation, University of Maryland, April 1999, Page 4 Conducting Survey Research On The Internet: Use of the Internet to Search for Information on Arthritis and Musculoskeletal and Skin Diseases (with acknowledgement of Registry assistance).

“Clinical and Genetic Studies of Disorders of Cornification and Other Selected Genodermatoses” (Netherton / ILC / Peeling Skin Syndrome or related disorders; EKV, PSE and related disorders; PPK, KID, Bart-Pumphrey syndrome, other disorders associated with hearing impairment or deafness)

2001 -- Dr. Richard (TJU):

Continuation of NIH study, additional disorders

Richard et al. Genetic heterogeneity in erythrokeratoderma variabilis. *J Invest Dermatol* 120:601-609, 2003 (with acknowledgement of Registry assistance).

Richard et al. Missense mutations in GJB2 encoding connexin-26 cause the ectodermal dysplasia keratitis-ichthyosis-deafness syndrome. *Am J Hum Genet* 70:1341-1348, 2002 (with acknowledgement of Registry assistance).

Sprecher et al. The spectrum of pathogenic mutations in SPINK5 in 19 families with Netherton syndrome. *J Invest Dermatol* 117:179-187, 2001 (with acknowledgement of Registry assistance).

Netherton Syndrome: Novel and Recurrent Mutations in SPINK5 and implications for screening and diagnosis. Gabriele Richard, Paulina Ratajczak, Shivan Amin, Humza Ilyas, Aida Tesfaye Kedjela Elaine C. Siegfried and Jouni Uitto. *J Invest Dermatol* 122: 2004 (abstract) (with acknowledgement of Registry assistance).

“Growth Failure in Infants and Children with Ichthyosis”

Mary L. Williams, MD and Melvin B. Heyman, MD

Date approved: 7/3/01

Fowler AJ, et al: Nutritional status and gastrointestinal structure and function in children with ichthyosis and growth failure. J.Pediatr.Gastroenterol.Nutr. 38 (2):164-169, 2004 (acknowledgement to the Registry omitted. Will follow in J Ped article).

Moskowitz et al: Pathophysiological basis for growth failure in children with ichthyosis: An evaluation of cutaneous ultrastructure, epidermal permeability barrier function and energy expenditure. J Pediatrics 145:82-92, 2004 (with acknowledgement of Registry assistance).

“Clinical and Genetic Studies of Disorders of Cornification and Other Selected Genodermatoses” (Netherton syndrome, Darier disease, Hailey-Hailey, EKV)

Gabriela Richard, MD

Date approved: 2/7/04

“Utility of Ultrastructure in the Diagnosis of Ichthyosis”

Anna L. Bruckner, MD, Mary L. Williams, MD, Melvin B. Heyman, MD.

Sponsor: Not Listed

Type of Diagnoses to Enroll: Harlequin fetus and survivors, Lamellar Ichthyosis, Congenital Ichthyosiform Erythroderma (CIE), Netherton syndrome, Epidermolytic Hyperkeratosis (EHK), Ichthyosis "En Confetti's", Ichthyosis Bullosa of Siemens (IBS), and the Erythrokeratodermas.

Submitted: 5/4/04

Date approved: 6/23/04

Mailings: 1 mailing; 239 Registry contact letters were sent.

“Clinical and Genetic Studies of Pachyonychia Congenita”

Mary Schwartz

Sponsor: International Pachyonychia Congenita Registry (IPCRR)

Type of Diagnoses to Enroll: Pachyonychia Congenita

Submitted: 12/15/04

Date approved: 12/17/04

Mailings: N/A

Responses: N/A

Leachman SA, Kaspar RL, Fleckman P, Florell SR, Smith FJ, McLean WH, Lunny DP, Milstone LM, van Steensel MA, Munro CS, O'Toole EA, Celebi JT, Kansky A, and Lane EB. Clinical and pathological features of pachyonychia congenita. J Invest Dermatol Symp Proc 10:3-17, 2005 (with acknowledgement of Registry assistance).

“A Systematic Review of Histology in the Ichthyosis Registry”

Leslie Robinson-Bostom, MD, John DiGiovanna, MD, Philip Fleckman, MD

Sponsor: none

Type of Diagnoses to Enroll: all with histology

Submitted: 1/4/05

Date approved: 1/9/05

Mailings: N/A

Responses: N/A

Ross R, DiGiovanna JJ, Argenyi Z, Fleckman P, Robinson-Bostom L:
Histopathologic differentiation of mosaic epidermolytic hyperkeratosis(EHK) from
generalized EHK. Abstract, Am Soc Dermatol, 2005

Ross R, DiGiovanna JJ, Capaldi L, Argenyi Z, Fleckman P, Robinson-Bostom L.
Histopathologic Characterization of Epidermolytic Hyperkeratosis: A Systematic
Review of Histology from the National Registry for Ichthyosis and Related Skin
Disorders. J Am Acad Dermatol 59:86-90, 2008

“Cross-Linked Protein Analysis: Application to the Ichthyoses”

Robert H. Rice, PhD

Submitted: 11/16/05

Date Approved: 11/30/05

“Netherton syndrome: Phenotype, Genotype, and Immunologic Defects”

Hans D. Ochs, MD, Ellen Renner, MD

Sponsor: NIH

Type of Diagnoses to Enroll: Netherton Syndrome

Submitted: 8/18/06, revision submitted 12/07

Date approved: 9/15/06

Mailings: 1 mailing, 14 letters sent

Responses: 6

Publications: Renner ED, et al: Comel-Netherton syndrome defined as primary
immunodeficiency. J.Allergy Clin.Immunol., E-pub ahead of print Aug 13, 2009.

**“Genetic and clinical studies of autosomal recessive congenital ichthyoses
(ARCI) belonging to the 12(R)-lipoygenase pathway”**

Judith Fischer, MD PhD, Philip Fleckman, MD, Leonard Milstone, MD

Sponsor: FIRST

Type of Diagnoses to Enroll: Lamellar/CIE with negative *TGM1* mutation testing

Submitted: 1/19/07

Date approved: 2/27/07

Mailings (consent): 38

Responses: 28

**“The spectrum of *TGM1* Mutations and Investigations of Genotype-
Phenotype in Congenital Recessive Ichthyosis”**

Jorge Toro, MD, PhD, Philip Fleckman, MD

Sponsor: NIH

Type of Diagnoses to Enroll: Lamellar/CIE with *TGM1* mutations

Submitted: 4/3/07

Date approved: 4/25/07

Publications: Farasat S, Wei MH, Liewehr DJ, Steinberg SM, Bale SJ, Ahvazi B,
Fleckman P, Toro JR: Transglutaminase-1 gene mutations in autosomal

recessive congenital ichthyosis: clinical and genetic investigations in a large cohort of 108 patients. *J Invest Dermatol*, 128 (S1):S111, 2008 (abstract).

Farasat S, Wei MH, Liewehr DJ, Steinberg SM, Bale SJ, Fleckman P, Toro JR: *Transglutaminase-1* and genotype-phenotype investigations of 104 patients with autosomal recessive congenital ichthyosis the United States. *J Med Genet* 46:103-111, 2009.

Herman ML, Farasat S, Steinbach PJ, Wei M, Toure O, Fleckman P, Blake PW, Bale SJ, Toro JR: *Transglutaminase-1* gene mutations in autosomal recessive congenital ichthyosis: a report of 23 novel mutations and modeling of TGase-1. *J Invest Dermatol* 129 (S1):S80, 2009 (abstract).

Herman ML, Farasat S, Steinbach PJ, Wei M-H, Toure O, Fleckman P, Blake P, Bale SJ, Toro JR. *Transglutaminase-1 (TGM1) Gene Mutations in Autosomal Recessive Congenital Ichthyosis: Summary of Mutations (Including 23 Novel) and Modeling of TGase-1*. *Hum Mutat* 30:537-547, 2009.

“The Economic Burden of Cutaneous Disease in Ichthyosis Patients and Families”

Suephy Chen, MD, Mary Spraker, MD

Sponsor: FIRST

Type of Diagnoses to Enroll: RXLI, EHK, LI/CIE, Netherton Syndrome

Submitted: 2/12/08

Date approved: 2/14/08

Mailings:

Responses:

Publications:

“Genes Modifying *TGM1*”

Jorge Toro, MD, PhD, Philip Fleckman, MD, Sherri Bale PhD

Sponsor: NIH

Type of Diagnoses to Enroll: Lamellar/CIE with *TGM1* mutations

Submitted: 4/23/08

Date approved: 4/30/08

Mailings (consent): 41

Responses: 29

“Clinical and Genetic Studies of Inherited Skin Disorders”

Keith Choate, MD, PhD, Catherine Yang

Sponsor: NIH

Type of Diagnoses to Enroll: subjects with ichthyosis where mutations have not been detected

Submitted: 7/8/09

Date approved: 7/15/09

Mailings: 24

Responses: 2
Publications: