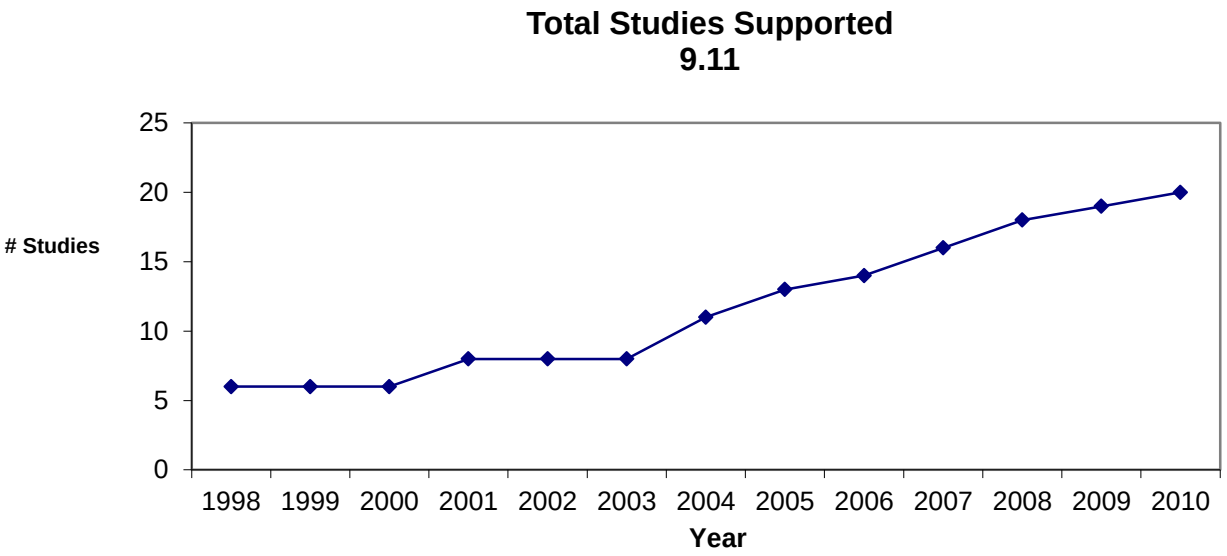
Studies Supported and Ensuing Publications - 5.31.12

Studies supported - 21
Studies supported since 9.04 - 11
Studies pending -
Total publications – 22



Letter 1,2,3,5,7,8,9,10,14,18,19,20
Data 4,11,12,16
DNA 15,16,18

Studies supported and ensuing publications

1. **"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

2. **"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 erythrokeratodermia variabilis. Nature Genetics 20: 366-369, 1998 (with acknowledgement of Registry assistance).
P1

3. **"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).
P2

4. **"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).
A1

5. **"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

6. **"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).
T1

7. **"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 erythrokeratodermai variabilis. J Invest Dermatol 120:601-609, 2003 (with acknowledgement of Registry assistance).
P3

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).

P4
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).
P5

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).
A2

8. **"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).
P6

Moskowitz et al: Pathophysioloical 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).
P7

9. **"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

10. **"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.

11. **"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).
P8

12. "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 Dermpath, 2005
A3

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
P9

13. "Cross-Linked Protein Analysis: Application to the Ichthyoses"

Robert H. Rice, PhD
Submitted: 11/16/05
Date Approved: 11/30/05

14. "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., 124:536-543, 2009.
PMID: 19683336
P10

15. **"Genetic and clinical studies of autosomal recessive congenital ichthyoses (ARCI) belonging to the 12(R)-lipoxygenase 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

16. **"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).
A4

Farasat S, Wei MH, Liewehr DJ, Steinberg SM, Bale SJ, Fleckman P, Toro JR: Novel transglutaminase-1 mutations and genotype-phenotype investigations of 104 patients with autosomal recessive congenital ichthyosis the USA. J Med Genet 46:103-111, 2008. PMID 18948357, PMC3044481.
P11

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).
A5

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.
P12

17. **"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:

18. **"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

19. **"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:

20. **In vitro activation of the sarco/endoplasmic reticulum pump in Darier Disease**

Ryan Hobbs, Kathleen Green, PhD, Amy Paller, MD
Sponsor: NIH
Type of Diagnoses to Enroll: subject with Darier Disease within driving distance to Chicago and subjects with Darier Disease with known *ATP2A2* mutations
Submitted: 10/6/10
Date approved: 11/10/10
Mailings: 3
Responses: _
Publications: _

21. **Clinical and Genetic Studies of Inherited Skin Disorders.**

Keith Choate, MD, PhD
Sponsor: NIH, Doris Duke

Type of Diagnoses to Enroll: subjects with ichthyosis where mutations have not been detected. Extension of #19 to include all Registry enrollees to use traditional and exome sequencing to identify mutations causing autosomal recessive congenital ichthyosis (ARCI), epidermolyic ichthyosis (EI), Erythrokeratodermia Variablis (EKV), Progressive Symmetric Erythrokeratosermia (PSEK), and disorders of keratinization which have not been possible to precisely classify those without a genetic diagnosis.
Submitted: 5/18/12
Date approved: 5/31/12
Mailings: __
Responses: __
Publications:

Other Publications

Mayes MD, Buyon JP, Pachman LM, Fleckman P, Giannini EH: Connective tissue disease registries. Arthritis Rheumatism 40:1556-1559, 1997. PMID 9324008
E1

Fleckman P, Hamill G: The National Registry for Ichthyosis and Related Disorders – the first 5 years. J Invest Dermatol, 114:869, 2000
A6

Fleckman P. Management of the Ichthyoses. Skin Therapy Lett 8: 3-7, 2003. PMID 14610614
P13

Fleckman P: The ichthyosis registry – a resource ready for use. J Invest Dermatol 123:x, 2004
E2