Studies Supported and Ensuing Publications - 5.31.12

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

Total Studies Supported 9.11

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
P1

4. “Quality of Life of Persons with Disorders of Keratinization”
Philip Fleckman, MD
Date approved: 10/21/96

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).

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


8. “Growth Failure in Infants and Children with Ichthyosis”
Mary L. Williams, MD and Melvin B. Heyman, MD
Date approved: 7/3/01


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

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
A3

P9

13. “Cross-Linked Protein Analysis: Application to the Ichthyoses”
Robert H. Rice, PhD
Submitted: 11/16/05
Date Approved: 11/30/05

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
PMID: 19683336

P10

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

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
A4

P11

A5

P12
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: _

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), epidermolytic 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

