Dear Colleague:

Thank you for agreeing to enroll affected individuals into the National Registry for Ichthyosis and Related Disorders. The National Institute for Arthritis and Musculoskeletal and Skin Diseases of the National Institutes of Health has funded the Registry with the goal of supporting studies of a broad nature aimed at determining the cause of these disorders and how to treat them more effectively. The Registry is also supported by the American Academy of Dermatology, The Society for Investigative Dermatology, and the Foundation for Ichthyosis and Related Skin Types (F.I.R.S.T.). Our mission is to identify individuals and families in the United States with the ichthyoses, erythrokeratodermas, Darier disease, Hailey-Hailey disease, palmar-plantar keratodermas, pachyonychia congenita, extensive epidermal nevi, and related disorders, and to encourage them to participate in the Registry. Because of the numbers involved, individuals with ichthyosis vulgaris are excluded.

The Registry consists of a database listing voluntary participants who are affected with such disorders and will include extensive historical, social, clinical, biochemical, and morphologic criteria that are the basis of the diagnosis of specific diseases. Also included will be a large group of individuals with "undiagnosed inherited scaling skin disorders". The Registry offers a well-characterized group of individuals with specific diagnoses for study by skin biologists, pharmacologists, and others. Recent advances in the understanding of the molecular basis of inherited skin disorders suggest that this is an opportune time to study individuals and kindreds with such disorders of keratinization.

How Does The Registry Work?

There are three steps to enrolling in the Registry: obtaining consent, a dermatologist's evaluation, and a phone interview. If you call the Registry with the name and address of a patient who wants to enroll, we will send you the Physician form to complete for them and will follow up with the patient to complete their enrollment. We contact every individual we receive a completed Physician Form for and help them finish their enrollment. We are asking that you contact any patients you feel would be appropriate and interested about enrolling in the Registry, and ask them about it. You can tell them there are some forms to fill out, and a phone interview with the Registry Coordinator to complete their enrollment. The steps involved are as follows:

1. We ask you to assist your patient by having them sign the Consent/Assent Forms, and returning one set to the Registry (the Assent Form is for enrollees who are under the age of 18). The forms can also be completed and mailed from home. We will be happy to answer any questions they have before obtaining their consent.

2. We ask that you fill out a short Physician's Form describing their diagnosis and the clinical findings supporting it. When completed, you can mail or fax the form to the Registry.

3. When we receive a Consent/Assent Form and/or a Physician's Form, we will contact the person being enrolled and discuss the Phone Interview with them. We provide a Patient Enrollment Form for them and their family to review at home. When they have filled it out as completely as possible, they call the Registry and tell us when it would be convenient for them to be interviewed over the phone. As a subject, they may refuse to answer any questions they choose. All this information is entered into a secured database.

Biopsies, Serum Cholesterol Sulfate Testing, and Other Findings

In cases where a skin biopsy has been obtained, we ask that you mail a slide for review by the Registry dermatopathologist; the slide will be retained by the Registry. (Please use a plastic case to protect slide(s) when shipping them to the Registry). Provisions have been made for the determination of cholesterol sulfate in serum as a measure of steroid sulfatase activity from males with a clinical diagnosis of sulfatase-deficient X-Linked Recessive Ichthyosis; diagnoses that do not have pathognomonic histopathologic, clinical, or biochemical findings; and undiagnosed disorders.
(To prepare the cholesterol sulfate serum samples, draw 7ml of blood in a sterile red top tube; allow the sample to clot; spin the sample in a centrifuge; pipette off serum and place it into a freezer tube; store and ship the sample while frozen. Please contact the Registry prior to sending any frozen samples so we can be expecting them.)

These evaluations will be performed at no cost to you or the enrollee. You will be notified of the result. If you have any other clinical or historical information pertinent to their diagnosis, we would appreciate a copy of those findings for our files as well.

**Research Projects**

The existence and goals of the Registry are being advertised to skin scientists in the U.S. Investigators desiring access to the database will submit a brief proposal that will detail the types of patients in which they are interested. After approval, appropriate patients will be identified through the database. Where patient contact is desired, appropriate affected individuals are mailed information inviting them to contact the investigator to participate in the proposal. In this way, affected individuals will have the option of participating in each appropriate study while their privacy is protected.

**Registry Database**

Confidentiality of enrollees is assured. The database is secured. Enrollees are assigned a patient code, and data stored by code for confidentiality. Access to the database and modification of data is by password and will be limited to approved Registry personnel. Identifying information of enrollees is given to no one by the Registry. If desired, affected individuals may enroll in the Registry anonymously. This means that the epidemiologic information about their skin disorder will be entered into the Registry, but without name, address or other identifying data. Anonymous participation in the Registry will preclude the ability of affected individuals to participate in any of the research studies, but the information may be useful for epidemiologic studies. If an enrolled individual wishes to withdraw from the Registry at any time, all identifying data pertaining to that individual will be purged from the computer files.

**Referrals**

If you desire assistance in making the clinical diagnosis, we would be happy to discuss the patient with you. Where appropriate, we have arranged for the Regional Support Network Consultant Physicians of the Foundation for Ichthyosis and Related Skin Types (F.I.R.S.T. 1-800-545-3286) to see affected individuals. We can refer you or your patient to the nearest center. Funding for the Registry does not include funds for physician visits or travel expenses, but the regional consultant physicians have agreed to keep the costs at a minimum.

Thank you for your help. Your cooperation in filling out the Physician's Form is indicative of your willingness to supply the personal information requested. If you desire further information, please feel free to contact us.

Yours truly,

Philip Fleckman, MD  
Associate Professor of Medicine (Dermatology)  
Principle Investigator, National Registry for Ichthyosis and Related Disorders

Geoffrey Hamill, RN  
Research Nurse II  
Registry Coordinator, National Registry for Ichthyosis and Related Disorders
**PHYSICIAN'S FORM**

Date of Completion of this form: 

Person in your office to contact for more information: 

Your name as Referring Physician:  

Address:  

Phone number(s): (_____) (_____) State Zip Code  

Date you first saw the subject as a patient:  

Are you certified by the American Board of Dermatology?  

Date you first made the diagnosis:  

Referring Physician #2:  

Address:  

Phone number(s): (_____) (_____) State Zip Code  

Date you first saw the subject as a patient: 

PATIENT INFORMATION  

(Note: leave this part blank and continue if the patient wishes to participate anonymously or chooses not to participate)  

Informed Consent obtained?  

___ No ___ Yes  

Affected Individual's Name:  

Date of Birth:  

Social Security Number (optional):  

Date of Diagnosis:  

Address:  

Phone number(s): Home (_____) Work (_____) State Zip Code  

Person to contact if Affected Individual is a minor:  

Relationship to Affected Individual:  

Address:  

Phone number(s):  

DIAGNOSTIC CRITERIA  

What is the diagnosis? (Please check one)  

**More Common Ichthyoses**  

___ Bullous congenital ichthyosiform erythroderma (BCIE) (EHK)  

___ Non-bullous congenital ichthyosiform erythroderma (CIE) and Lamellar ichthyosis  

___ Recessive X-linked ichthyosis  

**Uncommon Ichthyoses**  

___ CHILD Syndrome  

___ Chondrodysplasia punctata syndromes  

___ Harlequin ichthyosis  

___ Erythrokeratodermas  

___ Erythrokeratodermas variabilis  

___ Progressive symmetrical erythrokeratoderma  

___ Disorders of Keratinization Lacking a Specific Diagnosis  

___ Other 

Comments:  

PLEASE COMPLETE BOTH SIDES OF FORM
CLINICAL FINDINGS

What definitive data do you have to support the diagnosis?

Scale
___ Absent ___ Present (If present, to what degree?) ___ mild ___ mild/moderate ___ moderate ___ moderate/severe ___ severe

Nature of Scale (Check all that apply)
___ Flat
___ Accuminate
___ Follicular
___ Fine (smaller than half an inch in diameter)
___ Plate-like (larger than half an inch in diameter)
___ Serpiginous
___ Double-edged
___ Other (Describe)

Distribution of Scale
___ Localized ___ Generalized

Localized areas involved (Check all that apply)
___ Scalp ___ Groin ___ Arms
___ Ears ___ Chest ___ Scapula ___ Legs
___ Eyelids ___ Under Breasts ___ Back ___ Palms
___ Face ___ Stomach ___ Buttocks ___ Soles
___ Other areas

Erythema
___ Absent ___ Present (If present, to what degree?) ___ mild ___ mild/moderate ___ moderate ___ moderate/severe ___ severe

Comments:

Blisters
___ Absent ___ Present (If present, to what degree?) ___ mild ___ mild/moderate ___ moderate ___ moderate/severe ___ severe

Comments:

Erosions
___ Absent ___ Present (If present, to what degree?) ___ mild ___ mild/moderate ___ moderate ___ moderate/severe ___ severe

Comments:

Other abnormalities of the skin, hair, or nails

Skin
___ Absent ___ Present (If present, to what degree?) ___ mild ___ mild/moderate ___ moderate ___ moderate/severe ___ severe

Describe:

Hair
___ Absent ___ Present (If present, to what degree?) ___ mild ___ mild/moderate ___ moderate ___ moderate/severe ___ severe

Describe:

Nails
___ Absent ___ Present (If present, to what degree?) ___ mild ___ mild/moderate ___ moderate ___ moderate/severe ___ severe

Describe:

Any other systemic abnormalities ___ None apparent

Cardiovascular ___ Musculoskeletal ___ Respiratory ___ Gastrointestinal ___ Neurologic ___ Metabolic ___ Circulatory ___ Reproductive

Describe:

(To what degree?) ___ mild ___ mild/moderate ___ moderate ___ moderate/severe ___ severe

Describe:

(To what degree?) ___ mild ___ mild/moderate ___ moderate ___ moderate/severe ___ severe

INHERITANCE

Are other family members affected? What is the apparent inheritance of the disorder in this family?

___ No affected family members ___ Grandparent(s) ___ Parent(s) ___ Sibling(s) ___ Children ___ Aunt(s) ___ Uncle(s) ___ Cousin(s) ___ Niece(s) ___ Nephew(s) (Maternal / Paternal)

If other family members are affected, list their relationship to the individual whom you are enrolling.

What is the apparent inheritance of the disorder in this family? ___ Autosomal Dominant ___ Autosomal Recessive ___ Recessive X-linked ___ Mutation ___ Unknown ___ Other

Describe:

DIAGNOSTIC TESTING

Skin biopsy #1
___ Not obtained ___ Obtained Date obtained: ______/_____/______ Date slide sent to Registry ______/_____/______

Skin biopsy #2
___ Not obtained ___ Obtained Date obtained: ______/_____/______ Date slide sent to Registry ______/_____/______

Steroid Sulfatase
___ Not obtained ___ Obtained Date obtained: ______/_____/______ Date serum sent to Registry ______/_____/______

Results:

Other testing
___ Not obtained ___ Obtained Date obtained: ______/_____/______ Type of test: ___________________

Results:

Other testing
___ Not obtained ___ Obtained Date obtained: ______/_____/______ Type of test: ___________________

Results:

If the diagnosis is not clear, are you willing to: (Check all that apply) ___ Obtain additional information requested by the registry?
___ Refer the patient to a regional expert in disorders of keratinization?

Physician Signature ___________________________ Date ______/_____/______

Please send this completed form to the Registry using the return envelope or by fax.
Please remind the enrollee to call the Registry to obtain an enrollment form and to be interviewed.