The National Registry for Ichthyosis and Related Disorders was funded by NIAMS in 1994 to improve understanding of the pathophysiology, diagnosis, and treatment of the ichthyoses. Individuals with ichthyosis enroll in order to obtain and provide information about their disorder. A list of the disorders enrolled in the Registry can be found at www.skinregistry.com. Diagnosis is confirmed by strict clinical, histologic, and in the case of recessive X-linked ichthyosis, biochemical criteria. In appropriate cases, mutation detection is offered to individuals in whom known mutations have been shown to underlie their disorder. Extensive identifying, historical, clinical, and quality of life data are obtained during an interview by the Registry coordinator and are entered into a secure database.

Over 600 subjects have been enrolled. In most cases (>97%), enrollees ask to be informed of pertinent studies. Thus, a large group of affected individuals with well-characterized disorders interested in participating in studies of their disease is available for industrious investigators.

The Registry is available for “mining” at many levels. For example, a recent review of the quality-of-life data of enrollees was presented at the International Investigative Dermatology meeting and has been submitted for publication. Other investigators have used the Registry to identify and inform subjects about studies of specific clinical disorders such as erythrokeratodermia variabilis, X-linked recessive ichthyosis, CIE, and KID Syndrome (Taubenheim, 1990; Richardson et al., 1998; Zettersten et al., 1998; Fowler et al., 2004). In addition, a discussion group for investigators with clinical interests is maintained on the Registry web site. Investigators from within or outside of the US interested in pursuing this valuable resource are encouraged to contact the Registry for more complete information.

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References


Taubenheim A: Use of the Internet to search for information on arthritis and musculoskeletal and skin diseases. Doctoral Dissertation, University of Maryland, April 1999.