



Pachyonychia Congenita Project

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We hope that making available the relevant information on Pachyonychia Congenita will be a means of furthering research to find effective therapies and a cure for PC.

Response to the Commentary “Significance of Patient Registries for Dermatological Disorder”

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TO THE EDITOR

We were pleased to see the commentary “Significance of Patient Registries for Dermatological Disorder” by Dr de Souza and Ms Miller (2012). Registries are vital for teasing out complexities of dermatologic disorders, especially rare dermatologic diseases. The authors highlighted the need for patient registries, as well as some of the challenges associated with them (e.g., inadequate standards, lack of adherence to standards, funding challenges, and so on). We appreciate the authors’ acknowledgment of advocacy-led registries. Advocacy organizations are playing an increased role in clinical research, including involvement in registries and biobanks (Terry *et al.*, 2007; Landy *et al.*, 2012). Whereas some foundation-led registries are designed for advocacy, educational, or fundraising purposes as noted by the authors, many foundations have developed registries to further the natural history and epidemiologic understanding of specific conditions.

The Pseudoxanthoma Elasticum International Registry (www.pxe.org) collects donor-reported epidemiological data and has a corresponding biobank of DNA and tissue samples. The National Psoriasis Foundation (www.psoriasis.org) also has a US-focused registry and biobank with corresponding DNA samples. (The psuedoxanthoma elasticum and psoriasis registries were not included in the selected patient registries in dermatologic disorders). These

organizations, together with the US Hereditary Angioedema Association (www.haea.org), are all members of Genetic Alliance Registry & BioBank, a cooperative model to share infrastructure and expertise across member organizations (Terry *et al.*, 2011). The three registries mentioned above, similar to many registries, are funded by the advocacy organizations that run them (the article erroneously lists industry funding for the HAEA scientific registry). In the commentary, the Cystic Fibrosis (CF) Registry is identified as a gold standard. It would be of benefit to the readership to identify areas in which each registry listed in the article matches or exceeds the CF registry. For example, the International Pachyonychia Congenita Registry offers free genetic testing to all participants, and serves patients in 50+ countries in seven languages (Irvine, 2012). Although this is just one example, there are likely numerous successes from the other registries listed and those not included.

The current system of academic research and rewards is fractured and unable to facilitate true collaboration and sharing. This is not sustainable, and as a community we have the opportunity and the obligation to improve the biomedical ecosystem. It is clear that advocacy organizations, individual advocates, and citizen scientists are essential partners for transforming the system. Advocacy organizations are vital partners for registries and biobanks, as they are poised to develop communities

of trust and leverage scarce resources. They understand the unmet research needs of the community, can facilitate collaboration between diverse stakeholders, and are appropriate and dedicated stewards of data and samples. We encourage providers, researchers, industry, and advocacy organizations to work together to make these resources as strong as they can be. We also thank the authors for reminding the scientific community how important patient registries are for dermatologic disorders.

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