Plain Language Summary

How PXE International Helped Speed Up Research for Pseudoxanthoma Elasticum

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Study at a Glance

Title:
Advocacy groups as research organizations: the PXE International example

Lead Institution:
PXE International

Collaborating Institutions:
Genetic Alliance, University of California San Francisco, Jefferson Medical College, Brown Medical School

Study Type:
Perspective / Review Article

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Terry SF, Terry PF, Rauen KA, Uitto J, Bercovitch LG. Advocacy groups as research organizations: the PXE International example. Nat Rev Genet. 2007 Feb;8(2):157-64. doi: 10.1038/nrg1991. PMID: 17230202.

Summary

This article describes how **PXE International** went beyond being a support organization into an active partner in scientific research. Rather than simply raising awareness and funding research, the organization helps lead research efforts by building the first lay-led registries, collecting blood and tissue samples, creating a research consortium, and working directly with scientists.

One of PXE International's greatest achievements was helping identify the ***ABCC6* gene**, pathogenic variants in which cause pseudoxanthoma elasticum (PXE). The organization also developed the first genetic test for PXE, supported the creation of animal models, coordinated clinical studies, and planned one of the first clinical trials for vision loss associated with PXE.

The authors explain that rare diseases often face challenges because there are relatively few people affected by the condition, limited research funding, and little commercial interest. Advocacy organizations can help overcome these barriers by connecting researchers with patients, organizing biological samples and clinical information, and encouraging collaboration instead of competition.
The article also describes how PXE International shared its methods with many other rare disease organizations. This work led to the creation of the **Genetic Alliance BioBank**, which provides registries, biological sample storage, research agreements, and other resources that help accelerate research for many rare diseases.

Why this Matters for PXEers

This article highlights how individuals and families can play a direct role in advancing medical research. PXE International showed that an advocacy organization can do much more than provide support—it can become a driving force behind scientific discovery.

The collaborative model developed by PXE International helped identify the gene responsible for PXE, improve genetic testing, establish research tools that scientists still use today, and lay the groundwork for future clinical trials. The organization's approach has since been adopted by many other rare disease organizations, demonstrating that patient-led partnerships can significantly accelerate research and bring promising treatments closer to reality.

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