

# PXE International Accelerates Research: Advocacy Organizations are an Essential Component of the Research Enterprise

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## Challenges

Limited Research Funding  
Incomplete data

Extreme Phenotype  
Natural History  
Epidemiology

Difficult Sample Acquisition  
Patient Confidentiality  
Services and Funding

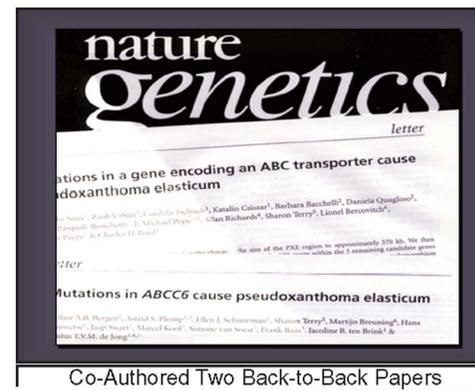
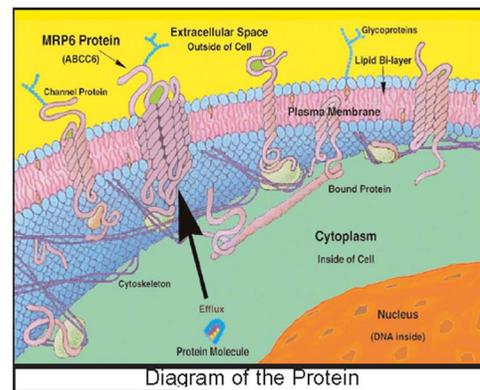
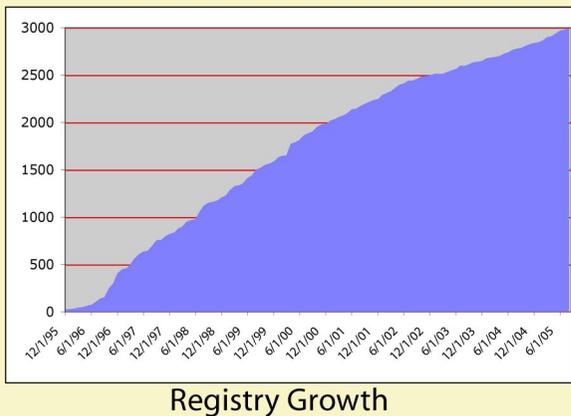
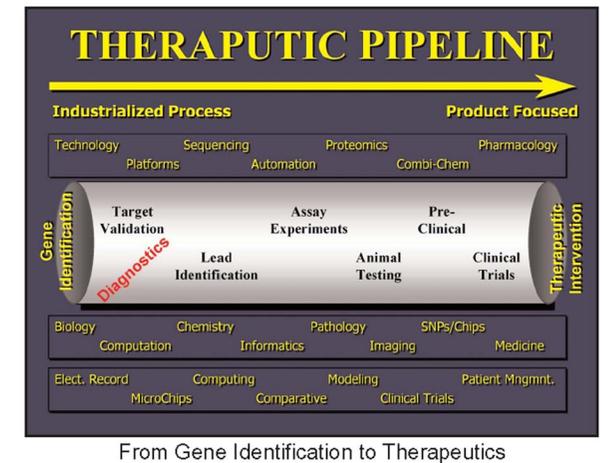
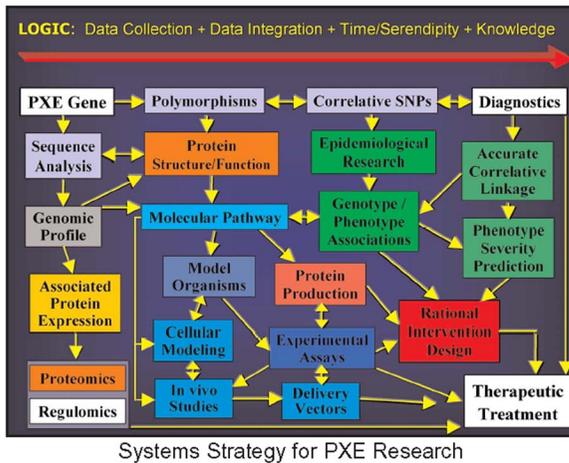
## Methods

Integrated samples and clinical data - BioBank  
Web portal for registry functions and education  
International fundraising and research grants  
Annotated bibliography of relevant research  
Laboratory consortium management  
International collaboration  
Global benefit sharing - IP  
MTAs and MOUs  
Respect for various cultures: research, clinical, patient and cultural

## PXE International Experience

Manage integrated database of:  
3000 affected individuals  
1000 pedigrees  
2000 DNA & tissue samples  
650 epidemiological study participants  
Initiate and/or fund research projects

Basic Science  
Animal model  
Protein and functional assays  
Genetic  
Mutation Analysis  
Comparative genomics  
Non-coding regions  
Molecular  
Cellular studies  
Clinical  
Natural history  
Phenotype/genotype  
Ethical participant management

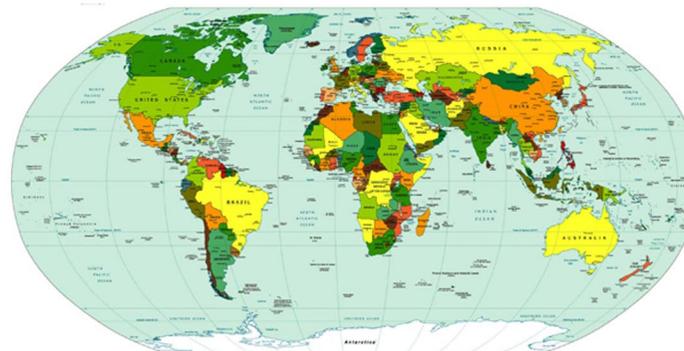


## RESULTS

Advocate viable research projects  
PXE Research Consortium  
Sponsored international meetings  
Multidisciplinary collaborations  
Focused agenda accelerates research  
New paradigm for partnerships  
Long term strategic planning

### Worldwide Scientific Collaboration

Australia  
Belgium  
Canada  
France  
Germany  
Hungary  
India  
Italy  
Japan  
The Netherlands  
South Africa  
United Kingdom  
United States



### International Patient Registry

Albania	Egypt	Malaysia	Singapore
Arab Emirates	Finland	Malta	South Africa
Argentina	France	Mauritius	South Korea
Aruba	Gabon	Mexico	Spain
Australia	Germany	Morocco	Sweden
Austria	Greece	New Zealand	Switzerland
Bahamas	Guam	Nigeria	Taiwan
Bangladesh	Hungary	North Korea	Tanzania
Basque Country	Iceland	Norway	Thailand
Belgium	India	Oman	The Netherlands
Brazil	Iran	Pakistan	Trinidad
Canada	Iraq	Panama	Turkey
Chile	Ireland	Papua New Guinea	Ukraine
China	Israel	Peru	United Arab Emirates
Colombia	Italy	Philippines	United Kingdom
Costa Rica	Jamaica	Poland	Uruguay
Cyprus	Jordan	Portugal	Venezuela
Czech Republic	Labanon	Romania	Viet Nam
Denmark		Russia	West Indios
Ecuador		Saudi Arabia	Yemen