

PXE Awareness

*National Association for Pseudoxanthoma Elasticum
(NAPE, Inc.)*

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*Seasons
Greetings*



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President's Message

As 2009 winds down, we reflect on what has been a busy year. Clearly more who have PXE are being diagnosed, in the U.S. and around the globe. Seldom does a full week pass without contact from those newly diagnosed. Our website is a powerful tool for providing information in terms accessible by lay persons. We plan in 2010 to present an improved website with a special section for those newly diagnosed.



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In this issue we present our almost unbelievable problem - keeping up with PXE research literature. We should all be thrilled by such a problem. I am, and share some thoughts about it. We want to be sure everyone knows about new federal law protecting those with genetic disorders. Linda Austin prepared an article with a contact for the law itself.

The Postal Service has changed bulk mailing regulations. We now must discover on our own new rules prior to preparing a mailing. Changes in this issue conform to recent new regulations. And let me acknowledge help in the mail process by Bob Worrell, long time member who with his late wife, Barbara, participated regularly in our annual conferences. Bob and Barb helped us prior to her final illness. We miss her and are grateful to Bob for continuing to help NAPE as a way to honor her.

Best wishes for a wonderful holiday season.

Fran Benham



A Genetic Bill of Rights

by Linda Austin



Genetic testing is the testing of a small sample of blood, other body fluid or tissue to determine if a person carries particular gene or protein markers which would indicate the possibility of developing a certain disease or a disorder such as PXE. According to the U.S. National Library of Medicine and the National Institutes of Health, about 900 different genetic tests currently exist. While the availability and number of genetic tests increase and can be beneficial for health and life decisions, people remain concerned that positive disease indicators resulting from genetic testing may negatively influence future or current employment and cause health insurance companies to either refuse coverage or charge higher rates. With the signing by then-President Bush in May 2008 of the Genetic Information Nondiscrimination Act (H.R. 493), the American people can rest easier knowing that the results of such tests are to be protected against invasion of privacy by employers or insurance companies.

In the later 1990's, when testing for breast cancer genes became readily available, women were worried that their test results could be used against them. The completion in 2003 of the mapping of the human genome then pushed a genetic rights bill through the Senate, but it took until 2007 for the House to finally pass a modified bill. The Genetic Information Nondiscrimination Act (GINA) was passed by the Senate in 2008 and signed into law shortly after. Sections of the law relating to health insurers went into effect in May 2009, sections relating to employers took effect this November. No health insurer or employer can require genetic testing or use genetic testing results to make health generalizations or rate adjustments for any group of people. The law also allows people to participate in research studies without fear their genetic information could be used outside the studies. The law does not, however, encompass life insurance, disability insurance or long-term care insurance. The federal law is only a minimum standard for all states, but any state is free to enact stronger laws against genetic discrimination.

The bill and additional information is available online at <http://www.govtrack.us/congress/bill.xpd?bill=h110-493>

A live link is posted in this newsletter issue on the NAPE website.

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Thoughts About Following PXE Literature

by Fran Benham

Prior to the identification of ABCC6 as the PXE gene mutation, the medical literature on PXE was sparse, generally limited to descriptions of individual or a few cases. After the announcement in 2000 of the gene discovery, PXE study articles increased so that today one can tap the Internet for numerous studies ranging from descriptive cases noted in regions where diagnosis is rare to analysis of the biology and chemistry of PXE. Remarkably, the studies are conducted by a growing community of global scientists. A quick review features reports from across the U.S., England, Holland, France, Germany, Belgium, Italy, India, Japan and China, countries with more than one research center reporting. And there are single sources from other countries, such as South Africa and Poland.

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The popular media often is the initial source about research - especially if it indicates something dramatic, overcoming blindness, for example. Such reports too often are misleading, but they typically name the research center involved. These groups usually have a website with more realistic expectations for the research.

The "big" topics in PXE continue to be vision, cardiology, and more recently, PXE as a metabolic disorder. There are reports specific to PXE on cardiology and metabolic research, and to a lesser degree, on vision issues, but for most vision studies, one needs to broaden the Internet search to AMD or degenerative retinal disorders. The number of studies is overwhelming. The search can be refined by adding the terms VEGF (vascular endothelial growth factor) or stem cell therapy. Even here, the number and complexity of studies are daunting. And all this does not cover significant concerns, such as low vision services and technology and rehabilitation services, so important to our community. So how can lay persons, some without computer skills, hope to keep informed?

If possible, start with family and friends who have computer skills and are concerned about you. If that doesn't provide enough support, talk to colleagues in social groups or church. Contact nearby rehabilitation services as they may have or know about support groups. If there is a visiting nurse association available, talk to them. They may help start a





group with a nurse leader. The same may be true of low vision service centers. Start your own group with others who have disorders such as AMD, glaucoma, diabetic retinopathy or other not uncommon degenerative retinal disorders. Researchers are informed by studies in all retinal degenerative disorders, and we may learn coping strategies, about local physicians and medical services, training programs, technology providers, etc., from others who struggle with various retinal disorders. Susan Golasz, NAPE member, formed such a group, the VIPs (Vision Impaired Persons) with various disorders. They meet monthly to share information, read aloud materials ranging from the popular media to medical literature, share community resource information and encourage each other. Susan reports good friendships have developed among group members as they support each other.

There are numerous groups which support those with vision loss. Some are activist oriented, including the American Council for the Blind and the National Federation of the Blind. Some focus on research fund raising, for example, the Foundation Fighting Blindness which conducts chapter meetings which provide research information. Lion Clubs are committed to helping those with vision loss and may be a good place to meet those who can help with information gathering. Even if a group does not focus on information about research, one may meet others to form a subgroup for that purpose.

Of course, NAPE will continue to support, encourage research and inform members. We report specifically about PXE, and if it seems important, about related studies. We expect in 2010 to keep readers aware of ongoing studies of PXE as a metabolic disorder and on possible AMD stem cell therapy studies. Should other studies reveal significantly important discoveries, they will be brought to your attention. For those deeply involved in understanding the biology and chemistry of PXE, the Internet is a valuable source.

What a marvelous problem - one we did not expect . . . so many PXE studies that lay persons are hard-pressed to keep track of them. When finding our mutant gene was a dream, we believed we could work together to find treatments and eventually a cure. We continue on that path and many have joined us. We welcome them as we continue to believe. NAPE will monitor scientific literature sources for information you may find useful. We encourage you to do the same with family, friends and in support groups.



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New Address

Name: _____

Street: _____

City, State, Zip _____

Old Address

Name, if different: _____

Street: _____

City, State, Zip _____

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