

PXE Awareness

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

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Lee Ducat – 30 Years and Going Strong

**Lee (right) with NAPE's Fran Benham
Celebrating Lee's 30 Years at NDRI's Helm**

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NAPE, a non-profit 501(c)(3) support group whose mission is to provide education and support for PXE-affected persons, publishes *PXE Awareness*. Articles in this newsletter are provided for information only and are not a substitute for professional medical advice. You should not use information in this newsletter to diagnose or treat medical or health conditions. Please consult your healthcare provider before beginning or changing any course of treatment.

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Contents

President’s Message	4
NDRI – 30 Years and Growing	5
Gene Patent Lawsuit – Update	7
FDA Approves New Wet AMD Treatment	7
Aching Legs	8



President's Message From Fran Benham

We continue to follow news and research as it relates to PXE. The gene patent case is still working its way through the courts. The U.S. Supreme Court ruled in another case against human gene patents. It sent our case back to the lower court for reconsideration in light of that ruling.

The FDA has given final approval to a new wet AMD treatment – Eyelea. To date PXE patients have been well served by the earlier approved medications, as well as Avastin which though not approved for eye treatment has proved highly successful for treatment of wet AMD and PXE.



The NAPE office is gradually contacting members, state-by-state to create with NDRI a list of PXE patients for possible research projects. NAPE has been a leader in such efforts, which started with the identification of our defective gene. That opened the door for much more research, which we expect to lead first to a treatment – and eventually to our cure.

We have made so much progress, but more remains and is crucial. Please respond positively when NAPE calls for your possible participation. We are not contacting you to ask for money – but for something that only a few can give. That is, to push research by participation in projects that we hope will lead to a cure. It will take months to work through our membership list. When we get to you, please say “Yes”. If you do, NAPE will give your contact information to NDRI, and they will send you a brief questionnaire. Your task will be to respond quickly to NDRI. They will enter you into their database and may contact you for a research project.

NDRI - 30 Years and Growing

NDRI (the National Disease Research Interchange) celebrated 30 years of leadership by its founder, Lee Ducat, who is stepping down from daily administration to focus on international opportunities. Lee was honored in a very special evening in Philadelphia in March. Fran Benham represented NAPE as our Board wanted to recognize the remarkable role of Lee and NDRI in connecting patient groups with specific researchers.

NDRI, with a competitive NIH grant, created its Rare Disorder Network to identify patients with rare disorders for the research community. In addition to working with NAPE, for example, to build a large potential patient group for research, NDRI has full-time staff who work with scientists to inform them about our disorder with areas of needed research. This is extremely important as it broadens the possibility of research needed to identify treatments and a cure.

NAPE is delighted that NDRI is going global. Their growth over the past 30 years has resulted directly from requests by the scientific community to NDRI to take on more and more disorders. Scientists developed great respect for and confidence in Lee and her staff to gather and provide accurate data about patients who are willing to consider participation in specific research projects. NDRI is equipped to support scientists in various ways according to need.

Much has been learned about PXE since Dr. Ken Neldner initiated the first lab at Harvard with the support of Drs. Klaus Lindpainter and Berthold Struk. There remain important questions yet to be answered. We are hopeful that follow-up research on Dr. Jouni Uitto's discovery of the role of magnesium in PXE mouse models may lead to a treatment for PXE. Such research requires human participation in carefully drawn projects. NAPE is cooperating with NDRI to develop as large a patient list as possible.

We are contacting our members individually state-by-state asking for each of us who has PXE to work directly with NDRI.

Each one of us, of course, will determine which, if any, projects we will engage in. Those who have been with NAPE even before it organized formally will recall that we provided Dr. Neldner with the blood samples that allowed Berthold Struk to begin the research that led to the identification of our defective gene. In the same spirit that led us to that immensely valuable discovery, we need to take the next step, perhaps a number of steps, to find the treatment to inhibit the development of PXE. It has been done in mice, thanks to Jouni Uitto and his team. Let's work with NDRI to help determine if this finding can be transferred to humans. When NAPE contacts you, please say "yes" and begin another exciting chapter of cooperation with NDRI, PXE patients and scientists. We can do this if we all work together: NAPE, NDRI and scientists together will make the crucial difference.

Finally, we know that PXE is found around the world in most, if not all, racial groups. NDRI's move to the international arena means we can identify many more potential research participants and researchers in diverse settings. Thanks to NDRI under Lee Ducat's vision, we continue to solve our own problem. Thanks in advance to all who will join with our Board to make it happen. And special thanks to Lee Ducat and NDRI for making it easier for our scientists to focus their energies on the research.

Gene Patent Lawsuit Update

In May 2009, the ACLU and the Public Patent Foundation filed a lawsuit on behalf of twenty plaintiffs including pathologists, geneticists, patients and health advocacy groups, including NAPE, against the U.S. Patent and Trademark Office, Myriad Genetics and the University of Utah Research Foundation. They argued that two genes patented by Myriad violate the First Amendment and U.S. patent law as genes are products of nature which cannot be patented. Plaintiffs argue that patents stifle scientific research and genetic testing. The case has gone through several rounds, has been appealed and has received several rulings. The most recent, a 2 to 1 decision, pleased neither side. The plaintiffs, led by ACLU lawyers, requested a Supreme Court review.

The U.S. Supreme Court set aside the most recent ruling and sent the case back for further review by the Appeals Court. In doing so, it pointed to the Supreme Court decision in another case which invalidated patents in genes since genes are not human inventions, but products of nature which cannot be patented. On June 15, both sides are to file positions in the case. Oral arguments will be presented before the Appeals Court on July 12th. The decision is expected by the Appeals Court this Fall. At that time it is possible that the losing side will again request a review by the U.S. Supreme Court.

FDA Approves New Wet AMD Treatment

The FDA has approved Eylea for the treatment of wet AMD. Eylea inhibits a factor that makes unwanted blood vessels grow in the retina. Eylea blocks this factor, called VEGF, as well as another similar factor. During its development, Eylea was called VEGF Trap-Eye. The generic name for Eylea is aflibercept.

Eylea is the third AMD drug to be approved by the FDA. Macugen inhibits a single form of VEGF, while Lucentis, like Eylea, blocks all known forms. All three are given by injection into the eye.

Lucentis is given monthly, although some patients may need it only once every three months. Macugen is given every six weeks. Eylea is given once every two months after three once-a-month injections. Lucentis costs about \$2,000 per shot. Lucentis is a low-dose form of the cancer drug Avastin and many doctors prefer Avastin at a cost of about \$50 per shot. It has FDA approval for cancer and thus can be used “off-label” for other purposes. Eylea will cost about \$1,850 per shot.

PXE patients have enjoyed good success with Avastin and Lucentis, and may want to discuss Eylea with their ophthalmologists.

ACHING LEGS

We have received many questions about aching legs. This is a complication of PXE which has been discussed in earlier issues. Most of us can deal with it through exercise. Walking daily is important. You may need to start slowly, but will need to build up until you finally eliminate the pain. Then walking must become a habit – or the pain is likely to return. Make your walks a pleasant part of the day – with a family member, friend, even a dog. You may prefer a standing bicycle. The other option, which we should avoid, if possible, is surgery. Talk with your doctor who should know about your PXE. Then get moving – it's all up to each of us. Those who have no leg pain should walk too, to make sure it doesn't start.



A Pleasant Walk With Friends