



**PXE International**  
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**eNewsletter**  
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**Do you have a question about your life, symptoms, and PXE that you want answered?**

Well, we cannot answer it without gathering the experience of thousands of PXEers. Your participation in the PXE International patient registry is the most effective way to help yourself and others with PXE.

So far only 350 PXEers have shared their experiences. This means that only 8% of potential PXEers have registered. When asked a question today, we can only base the answer on a narrow sliver of people's experiences and data.

Click the button below to register.

Already registered? Click the button to answer new questionnaires and connect your electronic health records.

[Click here to share your experience](#)

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## **Meet a PXE Researcher**

PXE International works with many dedicated researchers at the forefront of ABCC6 research in hopes of learning more about PXE and finding possible treatments. This month, we're shining a spotlight on Koen van de Wetering, DVM PhD.



Dr. van de Wetering is a biochemist and an assistant professor at the Department of Dermatology & Cutaneous Biology at Thomas Jefferson University. He became a Doctor of Veterinary Medicine at Utrecht University in the Netherlands in 1999 and went on to complete his PhD in Biochemistry/Lung Surfactant in 2003. In 2011, he completed his postdoctoral work on ABC Transporters and Metabolomics at the Netherlands Cancer Institute in Amsterdam.

PXE International spoke with him about his passion for research. See the

interview below:

### **What drew you to research on ABC transporters?**

I initially started my work on ABC transporters because of their anticipated involvement in making cancer cells resistant against chemotherapeutics. Over time it became clear, however, that most ABC transporters actually don't play a crucial role in drug resistance of real human tumors. From that time on, the focus of my research moved towards understanding the function of these proteins. What do we have these pumps for?

To study the function of the ABC transporters, we set up screens to see what they pump out of cells in living organisms like humans and mice. The screens that we had initially set up for ABCC2, ABCC3 and ABCG2 worked remarkably well to decipher the function of these proteins. At that time, it was still a mystery what ABCC6, the ABC transporter involved with PXE, was doing and we thought our methods could be used to explain why not having this protein results in the clinical manifestations of people with PXE.

It turned out to be a little more difficult than I thought, but eventually, together with Robert Jansen and Piet Borst at the Netherland Cancer Institute, we found what ABCC6 was pumping out of cells: Adenosine triphosphate (ATP). Although we were initially surprised by this finding, this made a lot of sense: ATP is important for the production of the calcification inhibitor pyrophosphate (PPi). In people with PXE, liver cells do not release ATP into the blood, so there is not enough calcification inhibitor pyrophosphate made. This explains why people with PXE have calcification in skin, eyes, and blood vessels. I should also stress that my wife, Leonie Huitema, was working on mineralization at that time. Talking about how mineralization is restricted to certain areas in the body was important for making sense of our discoveries at the time. We discussed the results of our exciting experiments daily at dinner!

### **You studied veterinary medicine, how has this informed your research with ABCC6?**

My study of veterinary medicine brought me two important things. First, I always look at how different animals develop differently. To give an example, there are bony fish and cartilaginous fish. Bony fish have a mineralized skeleton ("calcified"), whereas cartilaginous fish have a skeleton formed of cartilage. Knowing which processes lead to these differences might also have therapeutic consequences as they point to important pro-calcification and anti-calcification factors. Second, one of my Veterinary Medicine teachers introduced me to mass spectrometry, the technique we used to understand the function of various ABC transporters, as a way to find out what these pumps release from cells.

### **When you aren't teaching or in a lab, what's your favorite hobby?**

There are several things I enjoy doing when not working. First of all, I like hiking. In the Alps I can make hikes of multiple days and spend the night in "huts", which are located in remote but wonderful places. The scenery on the surrounding mountains in these huts is breathtaking. I also enjoy running, although I don't do that as much as I should. I still have the goal to run a marathon. Though I have to admit that goal is still far away. Dining-out in special restaurants is another thing I enjoy doing. Tasting new things, eating food from different cultures, I find that

really enjoyable!

Want to know more about what Koen van de Wetering is working on? [Watch his webinar on targeting the pyrophosphate shortage in PXE here.](#)

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## PXE and Eyes Bulletin

We updated the Eye Bulletin, a resource with information about PXE and eyes. If you have any questions about anything related to eyes, this is the first place to look. In this bulletin, you can find information ranging from how PXE affects the eyes to how to protect your eyes and current treatment options. If you have any questions about PXE and eyes that is not answered by this bulletin, please send your question(s) to [Tamara Wurst](#).

You can find the Eye Bulletin and all other bulletins on the website by clicking the button below.

[PXE Bulletins](#)

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## Mindfulness Through Gestalt Awareness Practice

In response to the stress many PXEers reported feeling because of the pandemic, Sharon offered three online experiences of [Gestalt Awareness Practice](#), something she has studied for about five years. In these short practice experiences, Sharon helps open a space for you to feel support moment to moment. She offers tools to help ground you, even in a time of challenge. This practice can lead to self-support and openness to what is. Recordings of these guided practices for PXEers and others are [available here](#).

In addition, Sharon is offering a weekend retreat called Introduction to Gestalt Awareness Practice. She is offering a 10% discount to any PXEer or their supporting friends and family. Right now there are 2 weekend offerings in May. Click the dates below for more information:

[May 15 & 16 - 8:00 to 15:00 EDT/14:00 to 21:00 CEST](#)

[May 22 & 23 - 10:00 to 17:00 PDT/13:00 to 20:00 EDT](#)

[Listen to the Guided Meditations](#)