

# Powerful Patient 2008 week 36

Host: Joyce Graff, WebTalkRadio.net

## Genetic Alliance

Sharon Terry, President and CEO of the Genetic Alliance, shares the story of her own family's struggle with a rare genetic disorder, and the work that she and her organization are doing to help families understand and utilize genetic information to manage their health.



Sharon Fontaine Terry

### 1) About Our Guest

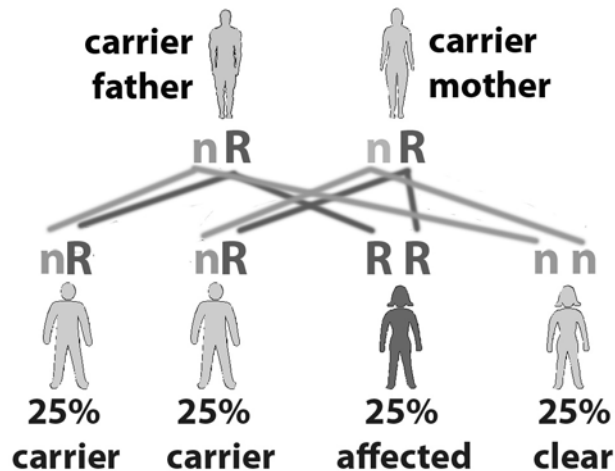
Sharon Terry, M.A., is a former geologist and college chaplain. In 1993 her two children were diagnosed with pseudoxanthoma elasticum (PXE), a disease which can lead to blindness about age 30-40. She and her husband decided they needed to take action to find a solution while there was still time to save their children's vision.

When two children both have a rare disease, and neither parent has that disease, then geneticist would say that this indicates that the disease has been passed as a recessive trait.

Everyone has two copies of each gene. In the case of a recessive trait like PXE or Cystic Fibrosis or Sickle Cell Anemia, each parent has only one altered copy of the gene in question, and has no sign of disease. When these two parents come together to make a baby, each child has a 25% chance of having two altered copies of the gene, which results in a child with the disease. (See figure below.)

#### Recessive pattern of inheritance.

Here each parent carries one altered copy of the gene R and one normal copy n. Each parent gives one copy of the gene to each child. Thus each child has a 25% chance of having two normal copies of the gene, a 50% chance of having one normal and one altered gene (and would thus be a carrier), and a 25% chance of having two altered copies of the gene, which would produce the disease. The sex of the child does not matter for this disease. Boys and girls have an equal chance of having the disease.



Armed with this knowledge from genetics, the Terrys knew they were looking for a recessive gene. They mapped out a plan, read and interviewed experts to learn whatever they could find, and set out to gather blood samples from every PXE family they could locate.

After many years of hard work they did in fact find the gene. The search continues for a realistic treatment. As with so many rare disease groups, the synergy of bringing together the families and the researchers is facilitating amazing progress.

Sharon became active in the Genetic Alliance, founded in 1981 by Joan Weiss, MSW. Sharon now heads the Genetic Alliance, as well as continuing to head the PXE Alliance, and chairs the Coalition for Genetic Fairness which was instrumental in the passage of the 2008 Genetic Information Nondiscrimination Act.

Sharon is at the forefront of consumer participation in genetics research, services, and policy. Learn what one mother has been able to accomplish by tackling head-on the issues that confront her children, and bringing them to the attention of the broader community. Her community has expanded to include others with PXE, then others with genetic illnesses, then others who can benefit from knowing their family medical histories, then others who have medical and genetic information to protect -- these issues in the end affect us all.

## **2) About PXE**

For additional information and resources for families with pseudoxanthoma elasticum (PXE), see <http://www.pxe.org>

## **3) About the Genetic Alliance**

Read more about the Genetic Alliance and its services at <http://www.geneticalliance.org>

## **4) Related U.S. Legislation**

For information about the Genetic Information Nondiscrimination Act of 2008 (GINA), what it covers and what it does not cover, see <http://www.genome.gov/10002328>

Sharon also mentioned the “Wired Act” which is under consideration in the Congress. See <http://www.ehealthinitiative.org/policy/congToHoldOrNotToHold.msp>

## **5) Family Health History**

For more information on compiling a family health history, see:

- Genetic Alliance, [http://geneticalliance.org/ws\\_display.asp?filter=fhh](http://geneticalliance.org/ws_display.asp?filter=fhh)
- U.S. Surgeon General’s Family Health History Initiative, <http://www.hhs.gov/familyhistory/>
- Family Health Portrait tool, <https://familyhistory.hhs.gov/>

- *Your Family Health Tree*, by Joyce Graff and Myra Vanderpool Gormley,  
<http://vhl.org/power/healthtree.pdf>

In conversation Joyce and Sharon mention MyFamilyHistory.com  
<http://www.myfamilyhistory.com>