

Powerful Patient #4

Host: Joyce Graff, WebTalkRadio.net

Diagnosis: Understanding the Challenge

Example: Pheochromocytoma

Diagnosis is a challenge. You and your doctors will need to work together to get to the right diagnosis.

Listen and learn. Don't come to the doctor with your mind made up – you don't have the breadth of experience your doctor has. Come instead with **questions** and **symptoms**.

- What does it feel like?
- When do the symptoms occur?
- Is there a relationship to times of the day?
Times of the month?
- Is it related to anything you eat? Is it related to mealtimes?
- Leave out “because...” – your hypotheses may distract

Likewise, your doctor needs to listen to your story. Make sure you tell it clearly and succinctly.

- Prepare your story in advance – no more than 3 minutes. If you go on longer, the doctor will run out of time.
- Make a list
- Perhaps keep a log to show the relationships you are sensing

The doctor will have other questions for you that will help validate or exclude various possibilities. Answer as accurately as you can. Don't make up answers, don't say what you think he or she wants to hear – tell the **simple true story**.

Diagnosis is difficult. Some doctors are better at it than others. And even the best diagnostician may propose a diagnosis, do some tests, then realize the first diagnosis is not correct, and move to another one. This is not a fault, it is quite normal. It is good for you that the doctor is working on the problem – don't criticize if the first diagnosis is wrong, **work with your doctor** to move to a better answer.



Clenton Winford with nurse Angela Kokonis in the halls at the Clinical Center, U.S. National Institutes of Health, Bethesda, Maryland

1) **Our Guests**

Clenton Winford II from Texas is the artist whose music is used as our theme music for the show. His CD is available for purchase through this website and at <http://www.clentonwinford.com>

Clenton has a diagnosis of von Hippel-Lindau (VHL). The pheo he discusses with us in only one of the many challenges he has dealt with as a result of VHL.

Carol (not her real name) from California graciously shares with us the story of her son's pheo. There is no family history of pheos in their family. His pheo metastasized, and most of the difficulties of his treatment stem from the metastasis. When pheos are removed at earlier stages there are usually fewer long-term repercussions. As with most conditions, the key is finding them early and getting appropriate treatment. That's why diagnosis is so very important!

2) **About Pheochromocytoma and Paraganglioma**

Pheochromocytoma is a tumor of the sympathetic nervous system. 80% of pheos are found in or near the adrenal glands. 20% of them may appear outside the adrenals, anywhere along the sympathetic nervous system, which runs from your earlobe to your groin, on both sides of the body. When they are outside the adrenals, and especially when they are higher in the body, they are sometimes called **paragangliomas**. In the neck they are sometimes referred to as **carotid body tumors**. For the sake of simplicity, I will call them all pheochromocytomas in this discussion, or pheos for short.

Most pheos are not malignant. Some are. All pheos should be carefully checked for malignancy.

Pheos occur at random in the general population. About 30% of them are caused by one of five different genetic alterations which can be inherited.

- Von Hippel-Lindau <http://www.vhl.org>
- Neurofibromatosis (von Recklinghausen's disease) <http://www.ninds.nih.gov/disorders/neurofibromatosis/neurofibromatosis.htm>
- Multiple Endocrine Neoplasia <http://ghr.nlm.nih.gov/condition=multipleendocrineneoplasia>
- SDHB and SDHD, two variants of [succinate dehydrogenase](#) complex:
 - <http://en.wikipedia.org/wiki/SDHB>
 - <http://atlasgeneticsoncology.org/Genes/SDHDID390.html>

If someone in your family has had a pheo -- and especially if more than one person in your family has had a pheo -- you yourself may be at increased risk for a pheo. You may wish to discuss this with a genetic counselor, who can help you draw your Family Health Tree and determine whether you might have some increased risk. You may, or you may not, depending whether you inherited whatever genetic alteration may be involved.

See our publication **Your Family Health Tree** for guidance in preparing for a visit with a genetic counselor.

Pheos have traditionally been considered very rare, and most doctors were taught in medical school that they were so rare they would probably never see one in their practice. For that reason, they tend not to be top-of-mind when making a diagnosis. Since 1996, however, research is showing that pheos are not as rare as previously thought. But diagnosis of pheos remains poor, and according to some estimates as many as half the pheos there are are not recognized until after the patient has died. In fact, the autopsy of President Eisenhower showed that he had a pheo which was likely the cause or certainly a contributing factor in the heart issues he experienced.

Basically, pheos emit hormones that cause your cardio-vascular system to go into overdrive – symptoms may include:

- High blood pressure (sustained or spiking)
- Headaches
- Palpitations
- Visual disturbances
- Constipation
- Anxiety, panic attacks, mood swings...
- Night sweats or heavy sweating
- Facial flushing
- Nausea, vomiting

Each of these is not an uncommon symptom of many conditions. When three or more of them occur together, however, they are worth checking out for a pheo. There are simple inexpensive tests to look for the chemical products of a pheo in the blood and urine

- 24-hour urine collection
- Plasma free metanephrines test

The interpretation of these tests, however, is not as simple. An endocrinologist or nephrologist familiar with pheos is best prepared to assist you. The right person is usually found at a large teaching hospital. In the case of inherited tendencies, depending which genetic alteration may be at work, there can be very different profiles of the levels of these chemicals. [Pacak et al., *J. Int. Med.*, 257: 60, 2005.]

If you and your doctor suspect a pheo, please check out the information at <http://www.vhl.org/pheo>