



Summer 2010

Polycythemia Vera and Myeloproliferative Neoplasms

Schuylkill, Carbon and Luzerne Counties

About ATSDR

ATSDR is part of the U.S. Department of Health and Human Services. It is a sister agency to the Centers for Disease Control and Prevention (CDC).

ATSDR gives information to the public to help you avoid contact with harmful materials. We use the best science we can to help you protect your health.

ATSDR has partnered with PADOH since 1989.

Summary

Polycythemia vera (PV) is a rare blood disease with no known cause. If you have PV, your bone marrow makes too many red blood cells, thickening your blood. Thick blood can lead to weakness, sweating, itching, headaches, tiredness, shortness of breath, blood clots and heart problems.

Myeloproliferative neoplasms (MPNs) are diseases where the bone marrow makes too many blood cells—red blood cells, white blood cells and/or platelets (clotting cells in blood). PV is one of the MPNs.

To diagnose PV and other MPNs, healthcare providers consider your symptoms, review your health history, do a physical exam, order blood tests and sometimes take a bone marrow sample. Right now, PV and other MPNs have no cure. But treatment can avoid or delay health problems.

In the United States, about one in 100,000 people are diagnosed with PV each year. In parts of Carbon, Luzerne and Schuylkill counties, more people than expected have PV compared with other locations. The Pennsylvania Department of Health (PADOH) and the Agency for Toxic Substances and Disease Registry (ATSDR) are working to find out why.

This fact sheet gives information about PV and other MPNs.

What Is Polycythemia Vera (PV)?

Bone Marrow

Bone marrow is the soft, spongy tissue inside bones. It makes blood cells.

Polycythemia literally means “many cells in the blood”:

Poly = many
Cyt = cells
Hemia = blood

PV is a rare blood disease that develops very slowly. If you have PV, your bone marrow keeps making blood cells—red blood cells, white blood cells and platelets—even when your body does not need that many cells. Over time, having too many red blood cells thickens the blood. This is why some people call PV “thick blood.”

Thick blood moves more slowly through your blood vessels, so parts of your body get less blood—and less oxygen from the blood. At first, you have no symptoms. Over time, thick blood can lead to weakness, sweating, itching, headaches, tiredness, shortness of breath and blood clots.

Blood clots are dangerous when they block the flow of blood in a blood vessel. Blocked blood flow to the heart can cause a heart attack. Blocked blood flow to the brain can cause a stroke.

Signs and Symptoms of PV (after having PV for years)	
More Common	Less Common
<ul style="list-style-type: none"> • Abnormal blood cell counts • Itching, especially after bathing • Enlarged spleen • Weight loss • Weakness • Sweating 	<ul style="list-style-type: none"> • Bruising • Nosebleeds • Budd-Chiari syndrome (when blood vessels to the liver become blocked) • Mitchell’s disease (when blood vessels in the feet or hands become blocked, causing pain and redness) • Gout (a painful joint condition) • Bleeding • Enlarged liver • Decreased blood flow to fingers and toes • Blood clots • Headache • Ringing, buzzing, or other noises in the ear • Dizziness • Blurred vision • Tingling sensations • Chest pain

Risk Factors

We do not know why people get PV. We do know that PV takes years to develop. The average age at diagnosis is about 60 years old. PV is more common in men than women. You are somewhat more likely to develop PV if you have a history of blood clots, other heart problems or a high platelet count.

Rarely, PV runs in families. We do not know of any specific environmental risk factors for the disease. Researchers are working to understand the reason for the PV cluster in northeast Pennsylvania. They are also working to see if there are any differences between people with PV and people without PV in the cluster area.

What Are MPNs?

MPNs are a group of diseases in which the blood-making cells in the bone marrow do not act normally. Often, the bone marrow makes too many red blood cells, white blood cells or platelets.

The three main MPNs are:

- Polycythemia vera (PV). See description of PV on previous page.
- Essential thrombocythemia (ET). In ET, the bone marrow makes too many platelets. This can cause blood clots. It can also cause serious bleeding.
- Primary myelofibrosis. In this disease, scar tissue replaces bone marrow. As bone marrow disappears, red blood cell and platelet levels drop. The liver and spleen begin to produce blood cells. This causes these organs to become larger than normal.

The three types of blood cells are:

- Red blood cells—carry oxygen to all parts of the body
- White blood cells—help fight infection
- Platelets—help blood to clot (to stop bleeding from a cut)

Myeloproliferative neoplasm literally means “(abnormal) new growths from fast multiplying bone marrow”:

**Myelo = bone marrow
Proliferative = fast multiplying
Neo = new
Plasm = growth**

Most Common Signs and Symptoms of Three Main MPNs(after having the disease for years)

Polycythemia Vera	Essential Thrombocythemia	Primary Myelofibrosis*
<ul style="list-style-type: none"> • Abnormal blood cell counts • Itching, especially after bathing • Enlarged spleen • Weight loss • Weakness • Sweating 	<ul style="list-style-type: none"> • Weakness • Bleeding • Gout (a painful joint condition) • Eye migraines • Tingling sensations 	<ul style="list-style-type: none"> • Anemia • General discomfort • Weight loss • Enlarged spleen • Loss of oxygen to the spleen • Enlarged liver • Swollen lymph nodes

* Many people with primary myelofibrosis do not have symptoms.

Risk Factors

We do not know why people get MPNs. We do know that MPNs take a long time to develop, so they are more common in middle-age and older adults. ET and primary myelofibrosis are more common in women than in men.

How are PV and Other MPNs Diagnosed?

Diagnosing PV

To find out if you have PV, your healthcare provider will:

- Consider your symptoms—to see if they match those of PV.
- Review your medical history—to see if there is a history of blood clots or heart problems.
- Do a physical exam—to see if your spleen is too large, areas of skin are reddish or purplish or if the gums are bleeding.
- Order blood tests—to check for signs of PV:
 - JAK2 mutation
 - High level of red blood cells
 - High white blood cell count
 - High platelet count
 - Abnormal levels of B12, uric acid, oxygen and erythropoietin (a hormone that helps trigger production of red blood cells)

If your healthcare provider suspects PV, he or she might take a bone marrow sample—use a needle to remove a little bone marrow from the hipbone or breastbone—to look for abnormal cells.

Diagnosing Other MPNs

To diagnose MPNs, healthcare providers once again consider symptoms, review your medical history, do a physical exam, order blood tests and sometimes take a bone marrow sample.

JAK2 Mutation

JAK2 is part of a signaling system that tells the bone marrow when to start and stop making blood cells. A mutation in JAK2 can cause the bone marrow to make too many blood cells. About 9 out of 10 people with PV have this mutation. About half of people with ET or primary myelofibrosis have the mutation.

If your healthcare provider thinks you are at risk of developing PV or another MPN, he or she might recommend a JAK2 blood test. A positive test does not mean you definitely have or will develop PV or another MPN.

With ET, healthcare providers look for high levels of platelets, large platelets, clumps of platelets and megakaryocytes (cells that make platelets).

With primary myelofibrosis, healthcare providers look for a low red blood cell count (anemia) and young or oddly shaped red blood cells. A bone marrow sample is needed to diagnose primary myelofibrosis.

How Are PV and Other MPNs Treated?

At this time, there is no cure for PV or other MPNs. But treatment can help avoid or delay health problems from these diseases. You might not need treatment right away, or at all. Your healthcare provider will monitor your health carefully to decide if and when you need treatment.

Treatment of PV

With PV, most health problems are caused by too many red blood cells in the body—or too many platelets. If you need treatment, your healthcare provider will likely use a combination of treatments to lower your blood cell counts and control your symptoms. He or she will choose among these treatments based on your age, gender, health, nutritional status, symptoms, and blood test results. Possible treatments include:

- **Phlebotomy (blood draw).** Phlebotomy simply means drawing blood. Instead of drawing blood for tests or blood donation, you will have blood drawn to lower the number of red blood cells in your body. Sometimes this is enough. If not, you might also need to take medicine.
- **Medicine to slow production of blood cells.** The most commonly used is hydroxyurea. Other medicines include anagrelide, interferon-alpha, busulfan and chlorambucil; these drugs can increase the risk of leukemia (a type of cancer). But the need to treat PV sometimes outweighs this risk.
- **Medicine to control symptoms or bad side effects.** Antihistamines can help ease itching caused by PV. Aspirin can relieve pain and help lower the risk of blood clots.

Treatment of Other MPNs

Like PV, other MPNs require a combination of treatments. These include phlebotomy, removing platelets from the blood and blood transfusions to replace abnormal blood cells with new blood cells.

What About the Future?

Although PV has no cure, good medical care can help you avoid health problems. Many people with PV who get treatment lead normal lives.

People with PV symptoms who get appropriate treatment most often live for more than 20 years after diagnosis. People with PV symptoms who do not get appropriate treatment often die within 5 years of diagnosis.

Nutrition plays a role in health. You might want to talk about your nutritional status with your healthcare provider.

Now that we know about the JAK2 mutation, researchers are testing new medicines to treat PV and other MPNs. For example, medicines that block JAK2 are being tested in people with PV. You can learn more by talking to your healthcare provider or by visiting this website:

www.mpdfoundation.org

How Can I Learn More?

- Ask your healthcare provider about JAK2, PV and other MPNs.
- Visit ATSDR's Web page on PV:
http://www.atsdr.cdc.gov/sites/polycythemia_vera/index.html
- Call ATSDR's toll-free PV information line:
866-448-0242