ATSDR
AGENCY FOR TOXIC SUBSTANCES AND DISEASE REGISTRY

pennsylvania
DEPARTMENT OF HEALTH

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JAK2 and Polycythemia Vera

Schuylkill, Carbon and Luzerne Counties

About ATSDR

The Agency for Toxic Substances and Disease Registry (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 the Pennsylvania Department of Health (PADOH) since 1989.

Other Mutations

The JAK2 mutation is the mutation most often tested for when a healthcare provider thinks a person might have PV. Other genetic mutations can also cause PV. Testing for these mutations is more difficult and uncommon. But your healthcare provider might order these tests if he or she strongly suspects PV even if a JAK2 test is negative.

Summary

The JAK2 mutation is a genetic change found in about 9 out of 10 people with polycythemia vera (PV). PV is a rare blood condition. In some parts of Schuylkill, Carbon and Luzerne Counties, more people than expected have PV compared with other locations.

A JAK2 blood test can help you find out if you have PV or if you *might* develop PV. A positive test does not mean you are sick or will get sick.

This fact sheet gives information about the JAK2 mutation and JAK2 blood testing.

What Is the JAK2 Mutation?

JAK2 is part of a signaling system (like a thermostat) that helps tell the bone marrow when to start and stop making blood cells. Most people with PV have acquired a mutation (change) in their JAK2. Because of this change, the bone marrow makes too many blood cells.

People with the JAK2 mutation might have or develop PV or one of the other myeloproliferative neoplasms (MPNs). On the other hand, they might never get one of these diseases.

PV and other MPNs take years to develop. You can be perfectly healthy and still have the JAK2 mutation. If you have the JAK2 mutation, you should visit your doctor regularly, even if you feel healthy. Medical care might help you avoid or delay health problems from PV or the other MPNs.

People without the JAK2 mutation likely will not develop PV—unless the mutation appears at a later time.

What Is the JAK2 Blood Test?

A JAK2 blood test can help you find out if you have PV or if you *might* develop PV. A positive test does not mean you are sick or will get sick.

To get a JAK2 blood test, visit your healthcare provider. They will draw a little blood and contact you with the results.

Should I Get the JAK2 Blood Test?

A JAK2 test is expensive (usually more than $1,000). Your healthcare provider will probably order the test only if he or she has a reason to think you might have a blood disease—for example, if you have an unusual blood count or an enlarged spleen.

What Happens if the Test Is Positive?

Having a positive JAK2 blood test does not mean that you have or will develop PV. However, you *might* develop PV or other MPNs later in life.

If your test is positive for JAK2, talk to your healthcare provider about the test, your health and next steps. Your healthcare provider can suggest ways to:

- Check to see if you develop PV.
- Avoid or delay health problems if PV occurs.

What are MPNs and PV?

MPNs are blood disorders in which the bone marrow makes too many blood cells. PV is an MPN in which too many red blood cells cause health problems. PV and other MPNs take years to develop. People with PV might experience headaches, tiredness, shortness of breath, blood clots and heart problems.

What Does a Positive Test Mean if I'm Healthy?

Having a positive JAK2 blood test does not mean that you are sick or will get sick. However, you *might* develop PV or other MPNs later in life. Your healthcare provider might want to check your blood counts and monitor your health carefully.

How Can I Learn More?

To learn more about JAK2, PV and other MPNs, you can:

- Ask your healthcare provider.
- 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

Is the JAK2 Mutation Found With Other Diseases?

The JAK2 mutation is also found with two other MPNs:

- *Essential thrombocythemia (ET)*
- *Primary myelofibrosis (PMF)*

About half of people with ET or PMF have the JAK2 mutation.