



Summer 2010

Polycythemia Vera in Northeast Pennsylvania

Public Health Activities and the Role of Healthcare Providers Serving Carbon, Luzerne and Schuylkill Counties

About ATSDR

ATSDR serves the public by using the best science, taking responsive public health actions, and providing trusted health information to prevent harmful exposures and diseases related to toxic substances.

ATSDR is a sister agency to the Centers for Disease Control and Prevention (CDC) and has partnered with PADOH since 1989.

Summary

In 2004, the Pennsylvania Department of Health (PADOH) identified a cluster of polycythemia vera (PV) in northeast Pennsylvania. In 2006, the PADOH asked the Agency for Toxic Substances and Disease Registry (ATSDR) to help study patterns of PV in the area. The investigation aimed to find all residents of the three counties who were diagnosed with PV between 2001 and 2005, confirm the diagnosis of PV among these persons and collect information from people found to have PV.

From December 2006 to July 2007, ATSDR conducted a survey of possible PV cases to collect information on work and residence histories, health status and other factors that might be related to PV. After its initial investigation, ATSDR concluded:

- Thirty-three cases of PV were confirmed by testing for the janus kinase 2 (JAK2) mutation.
- There were potential environmental exposure sources common to some of the high-rate areas.
- More research is needed to understand the reasons for the high rate of PV in this area.

As a public service, in 2009 ATSDR conducted a public health screening for the JAK2 mutation in the tri-county area. The screening also allowed ATSDR to locate, confirm and characterize PV cases in the area.

As a follow-up to this investigation and screening, PADOH is tracking cases of PV in the tri-county area. ATSDR is working with research partners to look for trends and risk factors for PV. And the Centers for Disease Control and Prevention (CDC) are working to improve reporting systems for PV and other diseases. These groups are also providing information about PV to local healthcare providers and the public.

The purpose of this fact sheet is to provide information about PV-related activities conducted by PADOH and ATSDR in northeast Pennsylvania.

Past ATSDR and PADOH Activities

Initial ATSDR Investigation (2006–2008)

In 2004, four cases of PV were found in people who lived on one road in the Tamaqua area in northeast Pennsylvania. In a follow-up investigation, the PADOH found more PV cases than expected in parts of Carbon, Luzerne and Schuylkill Counties.

From December 2006 to July 2007, PADOH and ATSDR began a search for all residents in these counties who had been diagnosed with PV between 2001 and 2005. PADOH and ATSDR gathered and analyzed PV patient information, such as individual traits, ancestry and occupational experiences. ATSDR also reviewed environmental data from the Pennsylvania Department of Environmental Protection (PADEP) and the U.S. Environmental Protection Agency (EPA) to determine if any exposures were common to the high-rate areas. In 2008, ATSDR presented the final findings of this initial investigation:

1. Thirty-three cases of PV were confirmed by testing for the JAK2 mutation. The confirmed cases had no common occupations, ancestry, lifestyle choices or exposures. The Pennsylvania Cancer Registry (PCR) did not accurately reflect the true number of PV cases in the area. In some areas, PV rates were higher than those in the rest of the tri-county area; in only one of these areas was the difference statistically significant.
2. There were potential environmental exposure sources common to some of the high-rate areas. It is not known whether a relationship exists between any of these sources and the PV cases. This initial investigation was not designed to study such relationships. Further, the cause of PV is unknown. Therefore, it is difficult to link the illness to any environmental agent or any other factor.
3. More research is needed to understand the reasons for the high rate of PV in this area.

PV and the Environment

A few studies, published more than 25 years ago, reported that PV could possibly be caused by exposure to chemicals (benzene, embalming fluid, petroleum products) or radiation. These reports were limited by small patient numbers, and their findings have not been confirmed by subsequent studies.

Public Health Screening (2009)

In 2009, ATSDR partnered with the Mt. Sinai School of Medicine to offer two rounds of free JAK2 screening to all residents who had lived in Schuylkill, Carbon or Luzerne counties for at least 1 year. The primary goal of this screening was to provide a public service. In addition, the screening enabled ATSDR to locate, confirm and characterize PV cases in the area.

Of the 1,170 persons tested in the 2009 screening, 19 (1.6 percent) were found to have the JAK2 mutation; 5 persons had previously been diagnosed with PV or a similar blood disease. Because other large-scale screenings have not been conducted to date, the prevalence of the JAK2 mutation in the general population is unknown.

The 2009 public health screening in northeastern Pennsylvania was the first large-scale screening for the JAK2 genetic mutation in the United States.

Current and Future ATSDR and PADOH Activities

Current and future ATSDR and PADOH activities serve the primary goal of understanding the cause of the PV cluster in northeast Pennsylvania.

PADOH is tracking cases of PV in the tri-county area and examining the feasibility of medical records review and JAK2 testing for all newly diagnosed PV patients. To identify disease trends, PADOH is comparing PV incidence rates in the tri-county area with rates from previous years and the overall state rate.

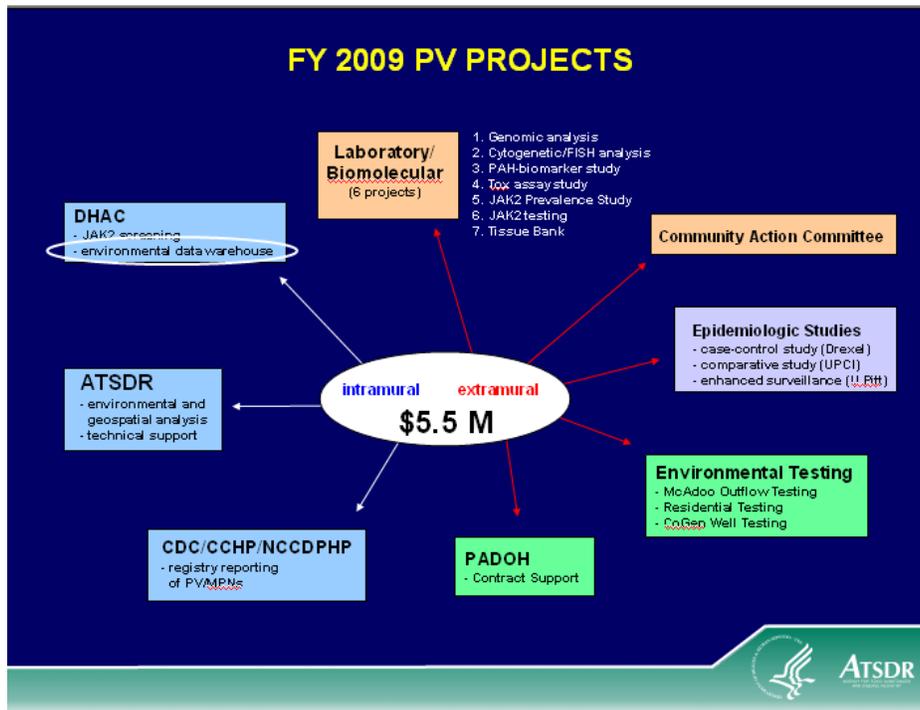
ATSDR is working with research partners to look for trends and risk factors for PV. Projects include:

- Identifying previous residence locations of PV patients in the cluster in order to determine potential environmental exposure sites and pathways.
- Identifying any differences between PV patients in the cluster and community members without the disease.
- Comparing the current cluster area with other locations having similar environmental conditions.

ATSDR will provide updates about the PV cluster in northeast Pennsylvania as more information becomes available.

The CDC is working to improve reporting systems for PV and other diseases. This will allow PADOH to track cases of PV in the tri-county area more accurately.

Finally, ATSDR/CDC and PADOH are providing information about PV to doctors, nurses, pharmacists and the public.



Role of Healthcare Providers

To assist ATSDR, PADOH and residents of the tri-county area, healthcare providers can:

- Provide medical care for patients with PV. (See the fact sheet “Polycythemia Vera,” provided in this information packet.)
- Respond to patients’ questions about the PV cluster and provide fact sheets as appropriate. (See patient information fact sheets provided in this information packet.)
- Report cases of PV to the PCR. (See the fact sheet “Reporting Polycythemia Vera,” provided in this information packet.)
- Encourage PV patients who reside in the cluster area to participate in the MPN tissue bank. For more information, contact:

Paul Roda, MD, FACP
Geisinger/Hazleton Cancer Center
Physician Liaison for Tissue Bank
570-459-2901

For More Information

For more information about PV, you can:

- 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