Polycythemia Vera Cancer Cluster Investigation in Northeast PA

**Determinant and accuracy of polycythemia vera diagnosis and use of the JAK2V617F test in the diagnostic scheme**

**Study Purpose**
The purpose of this study was to look at how patients with suspected polycythemia vera (PV) are diagnosed and if the cases are reported to state or local cancer registries. ATSDR also wanted to determine if treating physicians used the JAK2V617F mutation test, introduced in 2005, in the diagnosis process.

**What Was Studied**
PV is part of a group of diseases called myeloproliferative neoplasms (MPN). MPNs are a group of blood cancers where the bone marrow makes too many blood cells. The MPN designation also includes essential thrombocytosis (ET) and primary myelofibrosis (PMF). In 2005, several groups of researchers identified a gene mutation, JAK2V617F, that was present in more than 95% of PV patients. In 2008, JAK2V617F mutation testing was included in the 2008 World Health Organization (WHO) guidelines for diagnosing PV and other related MPNs.

In 2006, the Pennsylvania Department of Health asked ATSDR to help study PV patterns among residents in northeast Pennsylvania. ATSDR reviewed medical records, conducted genetic testing for the JAK2V617F mutation, and confirmed the presence of a PV cluster in three northeast Pennsylvania counties: Carbon, Luzerne, and Schuylkill. The study also found that greater than 50% of PV cases reported to the cancer registry did not have PV or that information was insufficient to confirm the diagnosis. Underreporting to the cancer registry occurred, with 15 of the 33 confirmed cases not being reported.

**Features of this Study**
At Geisinger Health Systems, investigators used electronic medical records for patients with a billing code of PV to determine if the PV diagnosis was correct and if the cases were reported to either the Pennsylvania Cancer Registry (PCR) or to the Geisinger Tumor Registry. Additionally, investigators checked to see if treating physicians implemented the JAK2V617F mutation test as part of the diagnostic scheme. For case identification, investigators used electronic health records for patients seen at a Geisinger Health Systems facility January 1, 2004 through December 31, 2009. To be included in the study, patients had to have at least one visit with a hematologist/medical oncologist and a billing code for PV.

**Gathering and Analyzing the Data**
Investigators obtained medical information specific for PV diagnoses for each patient and identified and included 277 patient health records in the study. Some information was missing from patient health records, resulting in smaller numbers for some parts of the study. A single study physician (the reviewing physician) reviewed each chart to identify the initial and final diagnosis as indicated by the treating physician. Reviewing physicians:
• Obtained sufficient information in 204 patient records to make a diagnosis.
• Used information within the health record and progress notes to confirm PV diagnosis.
• Further reviewed the health record for cases that did not meet the diagnostic guidelines for PV to
determine if the patient had secondary polycythemia, another MPN, or another diagnosis.
• Found sufficient information in 268 patient records to determine whether the case was reported to a
cancer registry.

Conclusion and Key Results

**JAK2V617F** testing has made a major impact in successfully identifying the correct type of
polycythemia (true polycythemia vera versus secondary polycythemia) in patients evaluated in a
large, community-based hematology/oncology practice in northeast PA.

- Of the 268 patients with enough information in their health record to evaluate cancer registry
  reporting, only 122 were reported to a cancer registry, and 34 of these had a final diagnosis of PV by
  the treating physician.
- Of the 204 PV patients with enough information in their health record to evaluate the accuracy of the
diagnosis and use of the **JAK2V617F** test, 121 had a final diagnosis of PV by the treating physician.
- Of these 121, the reviewing physician confirmed 90 true PV cases.
- When the reviewing physician used the strict 2008 WHO guidelines for diagnosing PV, 49 of the
  patients met the necessary criteria.

Physician use of the **JAK2V617F** mutation test increased from 2006–2009.

- In 2006, 40% of physician-diagnosed PV patients were not tested for the **JAK2V617F** mutation
  compared to 13% in 2009.
- Physician usage of other critical tests is inconsistent, possibly leading to errors in diagnosis.
- The study shows errors in PV diagnosis and underreporting of diagnosed cases to cancer registries
  in northeast PA.
- Physician education may be needed to improve PV diagnoses.

For More Information


Call ATSDR’s toll-free PV information line at 866-448-0242, or email Dr. Elizabeth Irvin-Barnwell, ATSDR
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