What are Myeloproliferative Neoplasms?

The Philadelphia Chromosome Negative Myeloproliferative Neoplasms (MPNs) are a closely related group of hematological malignancies in which the bone marrow cells that produce the body’s blood cells develop and function abnormally. The disorders are progressive and can strike anyone at any age, and for which there is no known cure.

The MPNs refer to three related neoplasms: Essential Thrombocythemia (ET), Polycythemia Vera (PV) and Chronic Idiopathic Myelofibrosis (MF). An MPN is featured by the proliferation of one, two or three bone marrow cell lineages.

The MPNs are considered rare diseases with a prevalence estimated of less than 200,000 in the United States.

Occasionally, the term MPN, for Myeloproliferative Neoplasms, is substituted for the term MPN.

Essential Thrombocythemia (ET) Polycythemia Vera (PV) Chronic Idiopathic Myelofibrosis (MF)

Chronic Idiopathic Myelofibrosis (MF)

Chronic Idiopathic Myelofibrosis (MF) is a malignant hematological neoplasm characterized by an enlarged spleen, varying degrees of anemia and low platelet counts, red cells in the peripheral blood that resemble tear drops, the appearance of small numbers of immature nucleated red cells and white cells in the blood, varying degrees of fibrosis of the marrow cavity [myelofibrosis] and the presence of marrow cells outside the marrow cavity [extramedullary hematopoiesis or myeloid metaplasia]. The syndrome can lead to marrow failure characterized by severe anemia and frequently low platelet counts. Non-specific symptoms include fatigue, weight loss and night sweats. Symptoms due to an enlarging spleen are also common as the disease progresses. Optimal care is at present supportive and palliative but new strategies including stem cell transplantation show promise.

Current treatment is directed at alleviation of constitutional symptoms, anemia and symptomatic splenomegaly. New drug therapies are continuously being tested. Various treatment options include hydroxyurea, interferon, thalidomide with prednisone, and oxymethalone. Depending on risk factors and disease progression, chemotherapy and allogeneic stem cell transplant may be an option.

Clinical trials are ongoing at this time with a new class of drugs which appear to be effective at reducing constitutional symptoms for MF patients. Whether one or more of these drugs can change the clinical course of the disease remains to be seen.

For more information on ongoing MPN clinical trials visit www.MPNResearchFoundation.org and click on Clinical Trials.

About the MPN Research Foundation

The MPN Research Foundation is a non-profit organization whose primary mission is to fund medical research for Myeloproliferative Neoplasms.

The MPN Research Foundation supports innovative efforts to advance the scientific understanding of the causes of Ph negative MPNs, and to support the developments of new treatments and ultimately a cure for the MPN’s.

The Foundation’s Scientific Advisory Board (SAB) utilizes a rigorous selection process to ensure donations are allocated to the most meaningful research projects. The SAB includes highly regarded scientists from top institutions. A list of the SAB members can be found on our website.

The MPN Research Foundation produces several newsletters a year informing patients on key advances in MPN research. To register for our free newsletter, visit our website at www.MPNResearchFoundation.org.

Where to get more information

The MPN Research Foundation website, www.MPNResearchFoundation.org contains more information about the diseases, treatments available, links to informative articles and websites, and research grant awards.

MPN Research Foundation
180 North Michigan Avenue
Suite 1870
Chicago, IL 60601
www.MPNResearchFoundation.org

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What causes MPNs?
In MPNs, the stem cell in the bone marrow that is capable of producing all blood cell lines is abnormal and overproduces certain types of blood cells.
In March 2005, researchers found that a single genetic mutation now called JAK2V617F appears in most PV patients, and about 50% of ET patients and MF patients. The mutation appears responsible for many of the features of these disorders. It also suggests that the diseases in most cases are acquired rather than inherited.
This finding has impacted the diagnosis and treatment of patients with these disorders and has spurred additional research into the origins of dysregulated cell growth and its function in the MPNs.

How are they diagnosed?
Diagnostic tests for these disorders include complete blood counts (CBC) blood analysis, red blood cell mass studies, bone marrow biopsies (BMB) cytogenetic studies, and ultrasound or CT scan to detect an enlarged spleen or liver. MF is characterized by bone marrow failure evidenced by falling blood counts and the amount of fibrosis (scarring) in the marrow. With the recent JAK2 discovery, the diagnostic process has been simplified. Your doctor will order the tests he/she feels are necessary to adequately diagnose a Ph negative myeloproliferative disorder.

Essential Thrombocythemia (ET)
Primary or Essential Thrombocythemia is a chronic progressive myeloproliferative neoplasm characterized by bone marrow failure evidenced by abnormal and overproduction of certain blood cell lines. This disorder is characterized by profound marrow megakaryocyte hyperplasia, splenomegaly and a clinical course punctuated by fatigue and risk of bleeding, fatigue, and excessive itching after exposure to water. Prolonged survival can be interrupted by the development of other syndromes including myelofibrosis and acute leukemia. The most common cause of morbidity and mortality is the predisposition of polycythemia vera patients to develop life threatening arterial thromboses (heart attacks, strokes, intestinal gangrene) and venous (thromboses of the portal and/or hepatic veins, pulmonary embolism) thromboses.

Treatment to return hematocrit to normal values is associated with a reduction of the number of thrombotic events. Treatment options include: phlebotomy with low dose aspirin or myelosuppressive therapy. Maintaining a hematocrit below .45 and .42 for men and women respectively, along with low dose aspirin, interferon or hydroxyurea. The choice of treatment is based on a variety of risk factors including age, history of thrombotic events and drug tolerance.

Polycythemia Vera (PV)
Polycythemia Vera is a chronic progressive myeloproliferative neoplasm characterized by an elevated hematocrit, an increase in the red cell mass, and usually by an elevated leukocyte count, an elevated platelet count and an enlarged spleen. Polycythemia Vera differs from many other hematological malignancies in that prolonged survival is enjoyed by most patients if excessive production of red blood cells and platelets can be controlled. The clinical course can be characterized by episodes of thromboses, bleeding, fatigue, and excessive itching after exposure to water. Prolonged survival can be interrupted by the development of other syndromes including myelofibrosis and acute leukemia. The most common cause of morbidity and mortality is the predisposition of polycythemia vera patients to develop life threatening arterial thromboses (heart attacks, strokes, intestinal gangrene) and venous (thromboses of the portal and/or hepatic veins, pulmonary embolism) thromboses.

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Thank you for supporting MPN research!