mpd-support

Sharing knowledge, supporting a cure

GUY'S & St THOMAS' CHARITY

Charity No. 251983

The newsletter for myeloproliferative patients October 2007

JAK2 spurs scientists to develop better treatments

Are we moving towards a cure? Dr Claire Harrison reports on the latest genetic research

The discovery of the JAK2 V617F mutation in 2005 has triggered an explosion in research into myeloproliferative disorders (MPD). There are hopes of new drugs, better treatment options and swifter, more certain diagnosis.

The acquired genetic mutation affects how quickly blood cells are made and perhaps how sticky they are. Almost all patients with polycythemia vera (PV) and about half those with essential thrombocythaemia (ET) have the mutation – they're JAK2 V617F-positive.

Scientists are also asking what causes MPD in JAK2 V617F-negative people and are scrutinising other parts of JAK2 as well as genes with related functions.

Last year, two new mutations – MPLW515L and K – were found in MPL, the gene for the thrombopoetin (platelet hormone) receptor. Rarer than JAK2 V617F, they have so far been found in 1-5 per cent of ET and myelofibrosis (MF) patients. Earlier this year, collaborative research by centres in Cambridge, Harvard, Belfast and London discovered four new mutations in yet another region of the JAK2 gene. They are also rare and seem to trigger a condition closely related to PV. More mutations may well be discovered.

Scientists are wondering how one mutation can trigger three different diseases – ET, PV and MF. It's now clear that this is partly due to the quantity of JAK2 V617F in blood cells.

For example, JAK2 V617F levels are much lower in the blood cells of positive ET patients than in positive PV and MF patients. Also, when ET patients transform to PV or MF, JAK2 V617F levels appear to increase.

Professor Vanucchi from Florence has shown that PV patients with high levels of JAK2 V617F might face higher clotting or thrombotic risks.

The great promise is of more effective treatment. Researchers have identified several potential new drugs. Some of these should enter trials in the US this year and in Europe next year.

And researchers are seeking better ways of using existing drugs as well. A trial starts in Europe next year comparing pegylated interferon with hydroxycarbamide. The trial will assess whether these treatments reduce JAK2 V617F levels, and if this improves a patient's condition.

We've taken exciting steps in the last two years and we think the best is yet to come. ■

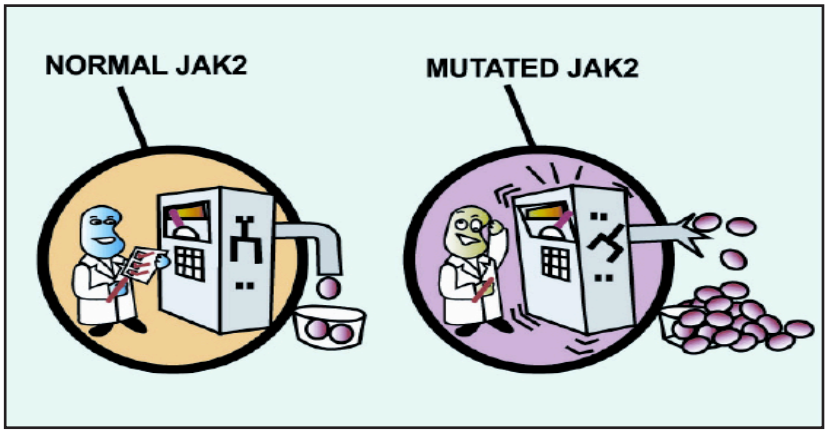

ILLUSTRATION BY LAWRENCE LAM

Bone marrow transplant cures patient's myelofibrosis

Stem cells weren't the only solution when Richard learned just after his honeymoon that he had myelofibrosis. Ann Marie Jahn explains how his persistence and management skills also helped the final result

Newly-married Richard and Susan had put off their honeymoon for several months after their wedding. They were looking forward to spending some time relaxing together in a breathtaking location – the Italian Dolomite Alps.

Richard, 53, had been feeling somewhat unwell before their trip. He had found himself out of breath when taking a long walk, but put it down to a tiring schedule. But when Richard arrived in the Alps, he began to feel fatigued and weak. He saw a local Italian doctor, who suspected bronchitis. Richard spent the rest of his holiday confined to bed.

When he returned home, he saw his GP, who suggested a blood test to look for what might be an underlying problem.

A few days later, Richard received an urgent phone call from his GP: "Mr Sanders, we have the results of your blood test, and you are extremely anaemic. We're going to need to run a test called a bone marrow biopsy. And I'm afraid you're going to need a blood transfusion."

> 'I realised I needed to drive the process if I wanted to live'

Richard soon received the devastating news that he had myelofibrosis. Richard wondered what more he could do. The queue at the clinic showed that his regional hospital's staff was massively overworked. They also appeared unfamiliar with treating myelofibrosis in a middle-aged person.

Richard decided to consult a haematology professor in London who made the following suggestions: Richard should get back to work, lose weight and start thinking about a bone marrow transplant as the only way out.

Haematologists at his local hospital decided to include him in a Mayo Clinic trial, combining a course of thalidomide with occasional blood transfusions. This was fairly effective for a few months. Over time, the thalidomide worked less well, and he required more frequent transfusions. However, Richard was strong, had given up smoking, and was relatively young; he was thus a good candidate for a bone marrow transplant (BMT). He wanted to try for a cure.

During the bone marrow transplant, Richard's own diseased bone marrow would be replaced with stem cells from a healthy, matching individual. He could be cured, but the risks were high. Richard began a series of work-up tests to ensure he was strong

Richard's 'survival tips'

Be your own advocate Project-manage your own healthcare. Ask for what you need. "Don't just sit there like a lemon and wait for someone to help you!"

Enlist the experts If you use a regional hospital, don't be afraid to seek expert medical help elsewhere to get the best solutions for your condition.

Think positively Work at your care. Someone has to survive, so why not you?

Using your skills

Richard used management skills honed in the business world to get the best treatment. Patients with different life experiences can in fact use all sorts of skills to improve their care. Close family ties, good communication with the medical team, online research or just persistence can all be effective problem-solving skills.

enough for the chemotherapy phase ahead.

Today, standing with hands on his hips, Richard describes his ordeal: "When I started chemotherapy, I got myself out of bed every morning, dressed, and read the paper sitting up in a chair. The other patients lolled around in their beds until noon, but I knew I had to keep pushing myself," he explains.

After chemotherapy, Richard was transferred to the isolation ward of the bone marrow transplant floor. The transplant took place on his first night in isolation. It was almost anti-climatic after all the anticipation: a bag of stem cells arrived at his bedside, looking for all the world like strawberry jam.

Richard watched as the marrow transfused into his own bloodstream, and then waited with growing impatience to see the results. However, as the days passed, Richard's blood-cell counts remained at near-zero levels: a slight flicker, but no significant change.

This was a serious setback, but Richard and the transplant team lobbied for another chance, and the donor agreed. Richard then underwent a second transplant. Day 1 passed, and his blood counts were slightly up. Day 2, a bit higher. Day 3, much higher. The transplant had taken.

Richard was discharged after three months in hospital, and was almost reluctant to go home. He was shaky on his feet and very emotional as he said goodbye to the team that had kept him sane, and given him the gift of future life.

He found new and unexpected challenges after his discharge from hospital. Two months after his release, his blood counts began to fall. Richard had developed Evans Syndrome, a condition in which the body attacks its own platelets and red blood cells. He was treated with a massive dose of steroids, which induced type II diabetes.

In contrast with the round-the-clock attention Richard had received in the BMT unit, he worked though his diagnosis of Evans Syndrome as an outpatient.

Richard became weaker at home and was finally readmitted to the intensive care unit. He then began a slow road back to recovery.

Richard's experience after his BMT is not uncommon. As cutting-edge treatments allow more patients to survive cancer, survivors are often left to struggle through the aftermath on their own. Richard's experience is unfortunately a common one for cancer survivors.

Richard left work for 23 months during his illness. He is now 57 and his blood counts are within normal ranges. He is back at work, and is enjoying time with his family. "How am I doing? I am being *normal*," says Richard. ■

Our thanks to Richard for sharing his story with us. He continues to feel well.

Bone Marrow Transplant Option

- Bone marrow transplants can cure MF, but involve substantial risks
- The transplant works by replacing diseased stem cells with healthy cells from a matched or partially matched donor, often a sibling
- Careful follow-up remains essential in the days, weeks, months and years after a transplant, to manage host-vs-graft disease and other risks
- Rapid advances in the field of bone marrow transplant have led to fewer side-effects and better patient results. For instance, so-called "mini transplants" use less aggressive chemotherapy to reduce patient discomfort

Myelofibrosis

- MF is a rare disease, affecting two people in 1m
- MF can occur at any age, but primarily affects people aged 50 to 70
- Patients with MF have trouble producing enough healthy blood cells, suffer from anaemia and bleeding problems
- Prognosis varies, depending on the patient's age and general health
- Not all patients need a bone marrow transplant. Many treatment options are available. You can find more information at www.mpd-support.co.uk
- Trials in the US are assessing new drugs for myelofibrosis. We'll report on progress in forthcoming newsletters

Expert Briefing

Dr Claire Harrison answers questions about an enlarged spleen

Q Dear Dr Harrison, I have myelofibrosis, and my blood counts are low but stable. Recently my spleen has been getting bigger, so my haematologist has started me with hydroxycarbamide (HC). Since then my platelets have dropped from 100 to 80. What should I do?

A Myelofibrosis affects your ability to produce all blood cells, including platelets. Some of your platelets are also pooled inside the spleen. Your platelet count has fallen from 100 to 80, but this variation is not too worrying and your haematologist will probably not be concerned.

As the medication you are taking starts to work, your spleen will shrink and release the platelets pooled inside. You may see an increase in your platelet count. If this treatment is not effective, your doctor can offer several other treatment options.

Changing what you eat and drink or how much you exercise will not change your platelet level. However, it's still essential to eat right and keep as fit as possible to help you avoid complications and improve your general well-being. We wish you good luck with your treatment. ■

You can find more information on treatments at www.mpd-support.co.uk

Please note that nothing contained in this newsletter is intended to constitute professional advice for medical diagnosis or treatment. You should always seek the advice of your physician or other qualified health provider prior to starting any new treatment or consult them on any questions you may have regarding a medical condition.

Peers offer friendship and skills

BY HELEN W WONG

Ever felt that you were the only person you've ever heard of with an MPD? Many people with myeloproliferative disorders feel isolated. It can be especially hard for those who can't travel and for younger patients who don't know anyone else coping with a serious diagnosis. Peer support can be a great way to share your experiences with others in similar circumstances.

How this works: peer support volunteers (who have an MPD) are paired with MPD patients seeking support. The volunteers offer emotional support, encouragement and friendship.

The programme is flexible and can work in a variety of ways. Patients can contact each other by telephone, email, or meet face-to-face. They can maintain a continuing relationship, or meet just once.

The buddy system works because peers have first-hand, experiential knowledge of MPDs. The volunteer's ability to listen and to share their own personal experience of living with an MPD brings a unique perspective to the support process.

> 'The volunteer's experience of living with an MPD brings a unique perspective to the support process'

Those seeking support benefit because they not only gain a friend who understands, but can learn new ways of coping with their MPD ■

For more information about volunteering or receiving support from the buddy programme, please contact us at www.mpd-support.co.uk

Keep your legs safe – look good too

BY MARJORIE LEONIDAS

Over 25,000 people in the UK die every year from complications of blood clots, but the Life Blood Thrombosis Charity is helping to prevent that.

Here are some of their recommendations: Don't cross your legs when seated or remain seated for long periods of time. Maintain a healthy weight, get regular exercise, don't smoke, and don't forget those compression socks!

A company called Sigvaris (www.sigvaris.com) offers a full range of compression socks and tights for both men and women in cotton, microfibre and microwool, in a selection of colours. Throw away those white tights! Instead, look great wearing comfy black cotton socks, while you prevent deep vein thrombosis. ■

Talking things over with your doctor

Your haematologist needs to address your concerns and you need to understand your treatment plan. Helen W Wong explains how good communications skills can make all the difference

Recent research shows that patients get well faster and feel more in control when they communicate well with their doctors. You can take steps to make the information flow more smoothly. Here's an approach to try:

Bring a list of questions So you can ask your doctor what concerns you most.

Discuss symptoms honestly Share your concerns openly with your doctor. Use descriptive terms to help the doctor understand your situation.

Know your goals Ask your doctor to define your healthcare goals – for example, your target blood pressure.

Double-check your understanding Repeat what you've understood back to your doctor using your own words. This helps you to remember and can clear up any misunderstandings.

Ask them to slow down Ask your doctor to speak slowly.

Ask for a drawing Ask your doctor to draw a picture or give you an illustration of the concept he or she is talking about.

Express your feelings Be honest about your feelings. Do you feel confused, overwhelmed, embarrassed, or frightened? Try using "I" messages to express your feelings: "I feel that... I am concerned that... I need more time to think."

Consider taking someone with you A family member or friend may help you understand information and remember instructions, especially if you are feeling unwell.

Ask for a summary Ask your doctor to repeat the main points and/or give you written instructions to take home.

These simple tools can facilitate communication between you and your doctor. ■

Helen W Wong is completing her MSc in Health Psychology

Fine-tune your skills

After you talk with your doctor:

- Think about how the conversation went
- Were you able to express what you were thinking and feeling?
- What might you do differently next time?

A physician offers his perspective on how doctors think

Dr Jerome Groopman's engrossing new book, *How Doctors Think*, explores how physicians make the decisions that affect their patients. He discusses how doctors use technology, observation and interaction with their patients to reach conclusions.

Technology seems to offer certainty but may be inconclusive or misleading. What's more, both patients and doctors can create barriers to communication.

Dr Groopman explains how medical professionals make the right decisions, and sometimes the wrong ones. The book covers ten case studies of patients working with their doctors. In the final chapter, Dr Groopman offers advice to patients on what to look for and which questions to ask.

"After writing this book," says Dr Groopman, "I realised I have a partner who may, with a few pertinent and focused questions, protect me from the cascade of cognitive pitfalls that cause misguided care. That partner is my patient or family member or friend." ■

Dr Groopman's book is published by Houghton Mifflin Company, and is available for £11.50.

Preserve health with eight crucial steps

BY ANN MARIE JAHN

As an MPD patient, it's important to focus on more than just your MPD, and here's why: MPDs make your blood "sticky," affecting blood flow. This can put you at a higher risk of thrombotic events such as deep vein thrombosis, heart attack and stroke.

It's imperative to do everything in your power to reduce your chances of developing preventable illnesses, for example diabetes or coronary artery disease. We didn't ask for or deserve an MPD, but it's within our power to fight back. Here's how:

1. Improve nutrition Make sure you get five or more portions a day of fruit and vegetables. Include green leafy vegetables every day and some yellow/orange vegetables every week. See the suggestions below about the best ingredients to add to your diet.

2. Lose weight if necessary to stay within normal levels. Weight loss information is available from www.cancer-research.org.uk and www.mayoclinic.com/healthyliving

3. Exercise regularly We all know that exercise lowers the risk of heart disease, strokes, deep vein thrombosis and diabetes in healthy patients. What's more, a study by Harvard University found that daily exercise can help patients with essential thrombocytosis reduce their feelings of fatigue.

4. Review your family history If some family members have a history of heart disease, diabetes, or breast or colon cancer, you may need more information on preventing these diseases. Additional illnesses such as diabetes can make complications more likely if you also have a myeloproliferative disorder. It's important to prevent additional diseases if possible and control them if they occur.

5. Get enough sleep Sleep can be difficult when we are anxious or worried, or busy for that matter, yet it's vital for health. Sleep allows our immune system to function correctly, and helps us cope with daily stresses.

6. Consider undergoing regular health-screening tests, from age 40 Basic tests include checking your weight, cholesterol and fasting blood sugar. Other essential tests are mammograms,

cervical cancer tests (Pap tests) and prostate cancer screening.

7. Control your stress level Identify what in your life may be putting you under excessive pressure. Make choices about your activities and workload based on your priorities. Try to focus on what you can control, for instance getting more information and help if you need it. Problem-solving techniques can also reduce stress.

8. Manage your own health Read your blood test results in detail. If you feel something is wrong, make sure your doctor understands your concern that something significant has changed.

The final word: You can't control it all, but do control what you can. ■

Nutritious Foods Shiri Morgan, Registered Dietitian at UCLA Medical Center, suggests that people include these very nutritious foods in their diets:

Avocados * Blueberries * Broccoli * Butternut Squash * Carrots * Soya Beans * Flax Seeds * Garlic * Kale and other dark green leafy vegetables * Lentils * Nuts and seeds of all kinds * Onions * Pomegranate * Quinoa * Salmon * Sea Vegetables (Dulse) * Tomatoes * Yogurt

Many of these foods are easy to find in supermarkets. Flax seeds, sea vegetables, dried soya beans and frozen soya beans (edamame) are available in health food stores, or online from www.goodnessdirect.co.uk

MPD Support now a charity

Dr Claire Harrison explains how we've grown and what's coming next

MPD Support is now three years old. Thinking back to those early meetings in 2004, we've achieved a great amount in this short time.

We launched MPD Support with a simple objective, to provide information and support to patients. Since 2004, we've held nine patients' forums, where patients meet each other and learn what's happening in the world of MPDs. We've developed a highly informative website for patients, and we've launched this newsletter.

Our reach is growing, as our website attracts hits from all over the globe. We recently received an email from a patient in Canada saying, "I've logged on here for more information because my doctor told me this was a great resource on the web!"

Many of you responded to our survey earlier this year asking how MPD Support can provide better service for you. You reported that you'd like frequent updates on research and drug trials, as well as additional patients' forums across the UK.

We've decided to take MPD Support a step further with these ideas in mind. We've launched a peer support programme, we've expanded this newsletter, and we're offering more meetings plus involvement in drug trials and research.

In addition, we've now opened an MPD Support fund under the auspices of Guy's and St Thomas' Charity. This charity is independent of the hospital trust and holds many special purpose funds for particular causes.

Our new status will give us many benefits: the charity will run our financial administration and ensure we can gain tax relief on donations. You can learn more about the GSTT Charity by visiting their website at www.gsttcharity.org.uk where you will find news about the charity's history, major projects and grants.

We're at an exciting crossroads in myeloproliferative disease. Cutting-edge research looks poised to deliver new treatments and understanding of these mysterious illnesses.

We look forward to meeting your needs in this changing environment with the best information and support we can provide. Thanks for your support! ■

Find advice for MPDs close to home

Many people with myeloproliferative disorders live outside London and want access to the best advice available.

Luckily there is no need to travel to London to find advice on MPDs.

You will find a list of haematologists with special interests in these conditions on our website. Just click on the link "healthcare professionals" to see a list.

Most patients with myeloproliferative disease are very well managed by their own local haematologists and don't need to see an MPD expert. But if you would like to do so, it's best to be honest with your haematologist and tell them why you would like to see an expert; this will help them to decide who the best person might be. ■

Runners get fit and raise funds

Dr Deepti Radia and Nurse Specialist Yvonne Francis ran for MPD Support and raised just under £1000. Yvonne Francis (pictured below, centre) com-

pleted her 5km run in 31:30.

Dr Radia (below, last on left in front row) discovered a great way to raise funds and promote fitness. Personal trainers Dan and Martin of Pro-Performance organised a 5km fun-run and offered to dedicate the event to MPD Support if Dr Radia

ran it too. "I don't do running," says Dr Radia, "but I decided to rise to the challenge. The morning was drizzly, but 11 of us in MPD Support T-shirts ran along the North Downs in Surrey, and we all finished." The runners raised £600 for MPD Support. "I still have to be convinced about running being fun," says Dr Radia. ■

A meaningful life with an MPD

Marjorie Leonidas explains the benefits of getting involved

When I was first diagnosed with polycythaemia vera I asked myself, "Why me?" I was active in my career, happily married and ready to start a family. Why was my life threatened, when all my peers were perfectly healthy?

When we learn we have this illness, we often feel angry and we look at the disease as the enemy. It takes a bit of mental gymnastics to turn our thinking around. We then realise with some shock that the disease might bring us a gift.

When I was diagnosed, I was frustrated at the limited help available to people with myeloproliferative disease. I decided to get involved and develop resources for other people in my situation. I worked with Dr Harrison and a group of dedicated patients to build our website and launch the first patients' forums.

We've made an impact. Patients and families across the UK are learning about MPDs from our website. The patients' forums are a welcoming place, where people with MPDs can make friends who truly understand their experience.

What's more, I discovered that my contribution could make a difference. My life is richer because of the friends I've made at MPD Support. In some sense I've found that having an MPD has added a positive dimension to my life. I invite all of you to get involved and join with us to help. ■

Learn more about getting involved with MPD Support at www.mpd-support.co.uk

To our readers

We've recently expanded MPD Support News to cover a wide range of topics. We hope you'll find this coverage informative and useful. Our thanks to Richard for sharing his moving story of what it's like to undergo a bone marrow transplant. We also appreciate the work of our contributors Marjorie Leonidas, Helen W Wong and Lawrence Lam. Please write to us with your suggestions for upcoming newsletters – we'd love to hear from you.

Sincerely, Claire Harrison and Ann Marie Jahn

"Which do you want first, the good news that sounds better than it is or the bad news that seems worse than you expected?"

Our next issue

- The latest news on the JAK2 inhibitor drug trials in the US and Europe: Roy Farndon reports
- How-to guide: Reading your blood test results
- MPDs affect your family too. Helen W Wong gives tips for communication

MPD Support News

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You can also contact the Guy's and St Thomas' Charity at info@gsttcharity.org.uk or visit their website for more information: www.gsttcharity.org.uk

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