MPDlife

The newsletter for
people with MPDs
June 2014

New test for myeloproliferative neoplasms

Dr Jyoti Nangalia updates readers on an exciting MPN research development

A new blood test can now be used to diagnose the vast majority of patients with myeloproliferative neoplasms (MPNs). The research was first published in December 2013, in the *New England Journal of Medicine*.

Many readers will know that MPNs cause the over-production of blood cells such as red cells, known as polycythaemia vera (PV), or platelets, known as essential thrombocythemia (ET) and can also cause scarring of the bone marrow called myelofibrosis (MF).

In 2005 scientists identified abnormalities in the JAK2 gene in patients with MPNs. Since the discovery, blood testing for JAK2 has become a routine part of the diagnostic work of patients with suspected MPNs. However, whilst the vast majority of patients with PV are positive for the JAK2 test and can be readily diagnosed, only about half of patients with ET or MF have a positive test. Diagnosing these patients is currently time consuming and requires multiple, often invasive tests, such as a bone marrow biopsy.

Easier and quicker diagnosis of MPNs via a simple blood test.

In the study led by Professor Tony Green from the University of Cambridge, and Dr Peter Campbell from the Wellcome Trust Sanger Institute, researchers identified a new gene called CALR that was found to be abnormal in 40% of MPN patients. In particular, this was found in the majority of patients with ET or MF that were negative for the JAK2 test.

Professor Tony Green said: 'Diagnosing these chronic blood cancers is currently difficult and requires multiple tests. Now, most patients with a suspected MPN will be able to be given a diagnosis after a simple blood test.'

Professor Tony Green

Scientists sequenced the DNA from blood samples of consenting patients with MPNs from Addenbrooke's Hosptial and Guy's and St Thomas' Hospitals. By analysing the DNA sequence, they were able to identify mutations in the gene CALR in patients. Additionally, they found that patients with the CALR mutation, unlike those with the JAK2 mutation, had higher platelet counts and lower haemoglobin levels.

Dr Jyoti Nangalia, who found the mutations in CALR said: 'Not only will the identification of CALR lead to a new, less invasive test, we also hope that it can lead to new treatments, just as the discovery of JAK2 did. The CALR gene is involved in a cell function, aiding with the folding of proteins made by the cell. This has not been implicated in these disorders before, so our research raises as many questions as it answers.'

Dr Jyoti Nangalia

Based on these findings a blood test has been developed for use in hospital haematology laboratories and is already available in a number of hospitals across the UK to help diagnose MPNs.

Dr Peter Campbell said: 'There is now a sense of completeness with these disorders. The vast majority of our patients can now have a definitive genetic diagnosis made. In the next year or two, we will see these genetic technologies increasingly used in the diagnosis of all cancers, especially blood cancers.'

To find out more

Further details can be found at the *New England Journal of Medicine* in the paper entitled "Somatic CALR mutations in myeloproliferative neoplasms with nonmutated JAK2" by Nangalia J, Massie C, *et al.*

MPN Voice

Readers will notice that we are now referring to our charity as MPN Voice rather than MPD Voice. This change brings us in line with the WHO classification of MPDs as myeloproliferative neoplasms.

We are also in the process of finalising the changes needed to improve the current website which will introduce some new sections and update the current content, making it even more accessible and useful for new and returning visitors.

Please do send us feedback on areas that you would like to see in the future. Email **info@mpnvoice.org.uk**

MPD voice

GUY'S & ST THOMAS' CHARITY

Charity number
251983

You are what you eat

health update

Louise Broughton shares how radical eating changes have helped her to overcome debilitating side effects of medication and MPN

Louise – 60 years young

Diagnosed with PV in 2005, Louise suffered with aquagenic pruritis, (extreme itchy skin) and other well documented side effects of MPN including fatigue and headaches. Six months after starting Hydroxycarbamide treatment she was also diagnosed with hypothyroidism and treated with Levothyroxine. She took aspirin and had regular venesections.

In 2011 a severe urine infection was treated over several months with continuous courses of antibiotics. Louise describes months of severe side effects after being told by her GP of many years, that these were not a result of the antibiotics and to keep on with the medication. When the infection persisted, Louise sought the opinion of a private urologist who found and successfully treated a huge abscess at the neck of her bladder.

Although the infection was cured, the antibiotic had caused a rare but known side effect –"Nitrofurantoin induced peripheral neuropathy".

Louise shares that, 'At this point I was feeling so ghastly. Malaise, headaches, nausea, searing stabbing pains in my legs, constant pins and needles in my feet and hands, tinnitus in my right ear. I felt like committing suicide!' Adding, 'I'm a hardy old boot and not the suicidal type. All this on top of that tedious PV symptom, fatigue.'

At such a low point, a friend suggested that Louise tried an elimination diet rather going back to the GP for yet more drugs.

Louise continues, 'For six weeks I only ate organic rice, fruit, vegetables and organic lamb, drinking water and naturally caffeine free tea; (the withdrawal symptoms over four days from caffeine were horrendous!).

Within two weeks I was feeling like a new woman. My headaches, nausea and malaise went, the stabbing pains much reduced, and oddly, the MPN fatigue reduced too. Over the following six months I began to introduce other organic foods into my diet, and found that I felt better than I had for years and all the symptoms disappeared, including fatigue.'

Current regime

Louise says, 'If I can, I avoid all non organic meat and fish or in the case of fish, I eat varieties line caught in open waters. I never ever eat chicken, unless organic, and even then I am careful. Marks and Spencer's is OK as is chicken from a local soil association accredited farm, others make me very unwell. I never touch dairy produce with the exception of the occasional small amount of organic butter.

I don't consume alcohol, processed or tinned foods, drinks or caffeine, including decaffeinated products which are chemically treated. I never consume non organic sugar whether white or brown, or rock or table salt , only pure sea salt.

Quite a lot of non organic vegetables and fruits are OK, but I now avoid non organic potatoes, onions, peppers, celery, mushrooms, butternut squash and beetroot, and non organic apples, pears, plums, grapes, strawberries, raspberries and blueberries. Thick skinned fruits are fine although I'm careful with mangoes and watermelons depending on the country of origin.

I was amazed to see a lot of the above are listed as to be avoided by the Penny Brohn Cancer Care website, almost two years after I'd come to this conclusion myself. I've never been a great devotee of organic food until now, regarding it as overpriced and unnecessary. How I've changed my tune!'

Maintaining the plan

Louise admits this is not an easy option and shares that, 'It's an absolute pain eating out, and going on holiday.' However she is committed to sticking with these changes, 'As the alternative makes me feel lousy and the pluses of feeling well outweigh the inconvenience.'

She does allow herself the occasional treat admitting, 'I had a glass of champagne when our daughter was married in December, and another glass on my 60th birthday and sometimes I'll nibble a bit of chocolate or a small piece of cake or scone if I'm out for tea.

I'm convinced that we are what we eat, and that the world's populous is very slowly being poisoned by the foodstuffs that they consume, mainly due to the added chemicals, preservatives, hormones and antibiotics that are pumped and sprayed, not only into and onto processed foods but also into animals, farmed fish, cereals, crops, fruit and vegetables, in the form of animal fodder, antibiotics, hormones, pesticides and fertilisers.'

Louise concludes; 'I believe that for those of us coping with diseases and cancers, taking chemicals into our bodies in the form of drugs it's doubly important to give ourselves an extra fighting chance by not stuffing them ever fuller with chemicals in the form of food and drink.'

The Bristol Approach to Healthy Eating

A guide on how to maintain a healthy balanced diet whilst living with cancer

Penny Brohn Cancer Care

Many of the dietary changes and principles Louise incorporated into her lifestyle eating are similar to the recommendations of The Penny Brohn Cancer care charity which has developed a healthy eating booklet and guidelines for patients and those wishing to reduce their risk of developing cancer. The guidelines are based on current evidence related to nutrition and cancer and over 30 years experience of staff at the charity.

At the heart of the approach is the belief that people are healthiest when they follow a diet composed of foods in their most natural state. The Bristol Approach encourages consumption of whole food which is fresh and unprocessed.

Visit the Penny Brohn Cancer Care website to download the booklet with a full list of dietary recommendations and further details about which foods should be increased and those which should be avoided or reduced.

www.pennybrohncancercare.org

Drugs trial success

patient story

Violet Slade started on a drugs trial in August 2013 and shares the transformation it has made in her life

Violet has led an amazing life, travelling internationally to the corners of the world. From an early age her love of animals has ruled her destiny resulting in a career in the world of dogs, training, teaching and judging.

In 1996 Violet visited Russia as a working dog specialist judge. The memories of the poverty and living conditions are still etched in Violet's memory and it led to her returning frequently to take medicine and vital supplies. She recalls, 'I started getting sick on my returns and In 2010 after a routine blood test at my surgery, I was sent to Addenbrooke's cancer unit. Further tests led to a diagnosis of PV and thrombocytosis as well as rheumatoid arthritis.'

Treatment started with blood thinning tablets, Hydroxycarbamide and intermittent venesection. Violet says 'I didn't understand all the instructions given to me. I came home convinced they had got it wrong. I was tired and sad, even depressed. I visited the unit every two weeks often giving blood, feeling weak and bloated with heartburn and indigestion. The fatigue was ruining my life. Travelling to dog shows was so difficult. Sleepless nights became the norm with disturbing dreams. My results at clinic did not improve, I flushed then shivered, my teeth suffered infections. I started to lose my hair and asked myself, "Is this my life from now on?"'

Life changed when, on a clinic visit, a doctor asked Violet to consider taking part in the MAJIC research trials. Violet read up on the latest information about the trials and after a little thought decided, 'What's to lose? I'm at an age to gamble a little. It might help others not just me.'

After assessment and pre trial tests, Violet started taking Ruxolitinib with blood thinning medication and reports, 'Almost at once my fatigue was gone and I gave up the drugs for the arthritis. Slowly my life came back. Now I sleep better and the flushes have gone. I eat only small meals, only fresh food, no alcohol or fizzy drinks, lots of juice and bottled water. I now rest for a while in the afternoon but I do have a busy life. For the past two years I have also followed the PH diet (green and healthy).'

Full of life and dedicated to animals

Violet now raises money for MPN Voice and is immensely grateful to the doctors and researchers who have supported her through her MPN journey. As well as all involved in MPN Voice, Violet also acknowledges the help given by her buddy who she says, 'Kept me sane when I was at my saddest times.'

The MAJIC trial is still open for MPN patients. Speak with your haematologist or visit **http://public.ukcrn.org.uk/search/StudyDetail.aspx?StudyID=11941** for further details.

Find out more about buddies

MPN Voice's Peer Support Programme offers individual support to all people with MPNs and to their partners and families. Many patients value the chance to speak with someone who has an MPN, a buddy who provides support via email or phone during what can be a difficult time. The goal of the programme is help patients cope with the common and emotional side-effects of diagnosis which may include feelings of fear, confusion and isolation. If you would like buddy support or feel that you can offer another MPN patient support email Maz Campbell-Drew **buddies@mpnvoice.org.uk**

Maz Campbell-Drew

Trials

Professor Claire Harrison comments on the latest MPN research and trials

Professor Claire Harrison

As we stand almost halfway through 2014 and on the brink of one of two major annual congresses what's new in MPN?

Undoubtedly what's made the biggest splash is the discovery of the calreticulin gene mutations. Following this we have seen data suggesting that having this mutation in ET or lacking any of the big three mutations in ET such as JAK, cMPL or CALR (as we call calreticulin) appears to be associated with a better prognosis and less risk of thrombosis. We also see that similar data is true for patients with MF. How can you get a CALR test? Most major hospitals now have access to this so please ask if you are interested.

Therapies and trials update

It's too early for CALR inhibitors but it certainly opens novel therapeutic options. On the trial front, after the withdrawal of fedratinib, SAR302503, comes an almost fully recruited trial with the drug pacritinib, the smoothened pathway inhibitors including drugs which target hedgehog including sonic hedgehog (what name will we think of next?!) and lots of combination studies. For PV and ET the MAJIC study is over 50% recruited and a major study in PV performed by Novartis and Incyte will announce details of the full results in Milan in mid June.

For the MPN Voice funded studies we are close to opening MEASURES, (a quality of life study) and are awaiting EU funding to top up our funding for the wider MOSAICC epidemiology trial (see page 4).

Personally I look forward to a hectic week in Milan which starts on the final day of my eldest son Henry's GCSEs. I also look forward to hearing about the many forums that will be held across the UK this summer and thank all the volunteers for supporting these and their teams.

Visit our website for a table of all trials and recruiting locations.

Patient advocacy

patient power

Jon Mathias, MPN Voice chair, explains what it is and why is it getting more important

Jon Mathias

Patient Advocacy has emerged over the past few years as an increasingly important part of the overall healthcare picture.

In the simplest terms, 'advocate' means someone who speaks on the behalf of another person or group of people. So Patient Advocates are people who represent patients in a variety of ways.

Sometimes there are situations where an individual patient may benefit from having someone speak on their behalf, rather like a barrister could argue a person's case in a court of law.

Another type of advocacy is where advocates act as a voice for a whole group of patients. In our case, people who have MPNs.

So it means speaking *for* patients, but who do advocates need to speak *to*?

The world of healthcare is complicated and getting more so. Patients, especially those like us with rare disorders, rely on a chain of organisations to deliver the care and treatments we need. There are doctors, hospitals, researchers, drug manufacturers and a variety of government agencies. They are all involved to some extent in our care but sometimes the priorities of one group or another conflict. In the middle of all this, Patient Advocates can help ensure that the patients' needs are properly understood.

Doctors usually shield us from this complexity of modern healthcare and in most cases patients don't need to know what goes on behind the scenes. However, when things are more difficult, it becomes important to understand who makes the decisions, how they are made, and where the money to pay for it comes from. The risk is that, in these processes, the perspective of the patient can be overlooked.

Advocacy for patients tries to ensure that our point of view is expressed clearly and consistently. We have seen in recent months how important this can be, in the ongoing discussion about whether the new drug Ruxolitinib should be made available for MF patients in the UK.

This debate could be seen as the thin end of the wedge. As effective therapies emerge from years of painstaking and incredibly expensive research, can we afford to pay for them? Or, more accurately, will governments and/or health insurers view them as value for money? Are the prices that drug companies charge for their new products exorbitant or a reasonable return on the massive investment they have made in their development and testing?

Patients and consultants working together. Some members of The MPN Voice steering committee.

These are very difficult questions and nobody has a simple answer to any of them. However we, as patients, deserve our seat at the table in the debates and one of the roles of Patient Advocacy is to be our collective voice, to ensure that our views are understood.

Readers will be interested to hear what MPN Voice is doing on a practical level. One of the challenges with MPNs is their rarity, so the more patients we can represent, the louder our voice will be. Over the past few months, we have teamed up with patient groups in other European countries to create an organisation that represents a larger community. At the same time, we are building a network of contacts in organisations like NICE, (the primary decision-making body in the UK) and the most important companies in the pharmaceutical industry, in order that MPN patients' needs and priorities are properly heard.

It's interesting and important work, and we need help. If you're interested in Patient Advocacy, please get do get in touch **info@mpnvoice.org.uk**

MOSAICC Epidemiology study hits recruitment target

Glen Titmarsh, PHD student updates that 216 participants have now been recruited far surpassing the 100 MPN cases initially required with 104 currently participating. He says, 'We really appreciate the commitment everyone has made taking part in this study. If you are contacted by the MOSAICC study team we would really value your participation.'

If you have any questions please contact the team through the website's Contact Page (http://mosaicc.qub.ac.uk). Study recruitment will cease at the end of May but any further participants will be accepted while statistical analysis is being undertaken over the next few months.

Glen also published a second paper titled 'How common are the myeloproliferative neoplasms: a systematic review and meta-analysis' in the American Journal of Hematology. This paper gives an estimate of MPNs in the general population. The MOSAICC study team will be meeting later this year to discuss the findings of the study as it comes to a close. Furthermore, Glen hopes to present his work at international conferences in Portugal and San Francisco later this year.

FUNDRAISING update

Since joining the MPN Voice steering committee last year, Marilyn Webster our new fundraising co-ordinator has been busy supporting many individual fundraisers and developing new exciting fundraising opportunities to raise vital funds which underpin our many areas of patient initiatives.

Fundraising heroes

Hairaising tactics

Mosaicc PhD student **Glen Titmarsh** (pictured right) raised £400 in total from his Movember effort.


BEFORE


AFTER

Family affair

Thanks to the **Austin family** who to date have collectively raised £2690, having participated in numerous fundraising activities, including a skydive by Lynn and her husband, a half marathon by son in law Danny and a dress down day organized by Lynn's friend which raised £145.


Never too young to fundraise.

Fundraising fun

Andrea Headech has been busy fundraising for MPN Voice having organized two fun events, a spa night and a 60s themed disco. Together the events raised almost £3,000 and Andrea is participating in the June abseil too!

Thanks to all our featured fundraisers and those who send financial support on a regular basis. We really appreciate all contributions whatever the amount. If you are planning an event, please let us know so that we can include your achievements in future issues.

UPCOMING EVENTS

- **28 & 29 June 2014**
  Sponsored Abseil challenge
  Abseil down King's College Hospital. Minimum target £100 per person. Registration fee £25 per person (£10 if registering as a team of 3+)
- **July and October 2014**
  The Moonriders Cycle Series 2014
  Exciting, 50 mile themed overnight cycle ride around the capital. Minimum target £200 plus registration fee of £39
- **31 October 2014**
  Halloween cycle ride
  Join hundreds of other spooked up cyclists in London's biggest Halloween cycle ride. Minimum target £200 plus registration fee of £39
- **13 September 2014**
  The Thames Path Challenge
  A 25 km walk or run from Runnymede to Cookham. Minimum target £145 plus registration fee of £25
- **14 September 2014**
  The Deep RiverRock Belfast City Half Marathon
  £18 entry fee. Participants need to finish in 3 hours or less

2015

MPN Voice is delighted to announce that places have been secured for three iconic fundraising events

- **March 2015**
  The Adidas Silverstone Half Marathon
- **April 2015**
  The Virgin Money London Marathon
- **May 2015**
  The Bupa London 10,000

For details on all our events email **marilyn@mpnvoice.org.uk** or visit the fundraising section of our website.

Visit our **News and Events blog**:
www.mpdvoice.org.uk/news-events
or visit our Facebook and Twitter pages

Living with MPNs day

The November 2013 Living with MPNs day was fully subscribed with over 200 delegates from around the UK and Europe attending the School of Oriental and African Studies, London

A packed agenda from world leading MPN experts included talks on new drugs for MPNs, an overview on MPNs, Allogenic Stem Cell Transplantation, a guide to anti-coagulant treatments, a panel discussion from patients and their families and break out discussion groups for ET, PV, MF patients and families. MPN Voice would like to thank Professors Tony Green, Mary Frances McMullin, Nicolaus Kroger and Claire Harrison and Dr Gary Benson for presenting to the patients and their families and a special thank you to Professor Ruben Mesa who sent a well-received video presentation as he was unable to personally attend.

Feedback from the delegates indicated that the day had been well received. One delegate wrote, 'A really good day thank you. Very grateful for the opportunity to attend a source of much valuable information.' Another commented, 'Please do it again. Excellent, informative and enjoyable.'

All the doctors' presentations from the day are available to view as a video on the MPN Voice website (**www.mpnvoice.org.uk**).

We plan to run a similar event in 2015 and will update you in forthcoming newsletters.


Photo: Gordon O'Neill, IotaMEDIA

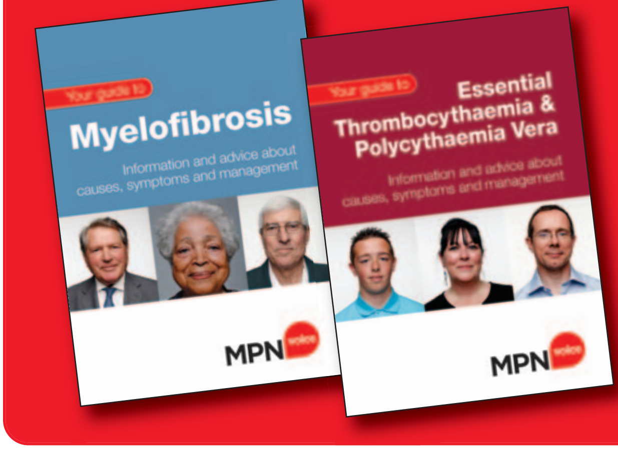
Prof Nikolaus Kroger (left) and Prof Claire Harrison (right) pictured at the 'LIving with MPNs day' in November

More help needed

As we increase our support for MPN patients the need for more volunteers grows in many areas. If you have time to spare and skills to share please contact Marilyn Campbell-Drew on **maz@mpdvoice.org.uk**

New leaflets available

We have been developing and updating our range of literature and have 2 new disease leaflets for Myelofibrosis and PV and ET available in A5 format. We have also extended the range of drugs leaflets and added leaflets for Danazol and Ruxolitonib. These can all be downloaded from the website (http://www.mpdvoice.org.uk/treatments/treatment-leaflets) or if you want to order bulk quantities for clinics or patient groups please email Maz on **info@mpnvoice.co.uk**



Forums

New regional locations for patient forums in 2014…

A great way to keep up to date with the latest on MPNs and meet other patients and families living with MPNS. Informal, relaxed and for you, please do come along to one of our patient forums this year.

- **6 June** Southampton
- **28 June** Manchester
- **10 October** Newcastle & Edinburgh

Dates for Cambridge and London will be confirmed soon and will be promoted via email and on the MPN Voice website.

Our continued thanks to Samuel Sebba Charitable Trust whose generous funding has enabled the expansion of MPN Voice regional forums.

MPDlife

- Want to be featured in our patient story?
- Do you have tips to share with readers on managing MPNs?

If so, please email the editor at the address below.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