Wilson's Disease Support Group - UK

NEWSLETTER **APRIL 2013**

Welcome

The Wilson's Disease Support Group - UK (WDSG-UK) is an all volunteer organisation which strives to promote the wellbeing of patients with Wilson's disease.

It publishes an annual newsletter with informative articles written by medical professionals and also articles written by patients, their families and friends about their experiences of the disease.

It promotes networking of Wilson's disease patients and their families by helping and encouraging contact with one another.

And the Group strives to promote a wider awareness of Wilson's disease within the medical profession.

AFFILIATED TO :

Hello Everyone,

I hope by the time you read this that Spring has finally arrived! Early April and I'm gazing out of my window at the last small pile of snow in the corner of the garden and the battered crocuses. I don't think I ever remember such a cold spring, though my eldest brother tells me that when I was a small child he remembers our dad having to dig the house out of snow drifts in late March; we used to live in *the wilds* of Derbyshire.

As far as the Support Group goes I'm afraid I've been about as much use as a chocolate teapot over the last few months! I have been waiting around since last Autumn for the hospital to give me a date to go in for a knee replacement operation, although it wasn't until early February that I was finally called in. Since then you might have noticed my absence from *facebook*, but I'm back now and it's good to note that the site has doubled in popularity over the past twelve months. We now have some 115 members and if you haven't already joined, then why not join now?

As regards the newsletter, Valerie has had to crack on with it on her own this year. But there's plenty to enjoy, including three patients' accounts of their experiences of Wilson's disease, an article by me, a Chairman's report from Rupert, a resumé of the 2012 annual meeting in Cambridge, and news and stories of fundraising activities that have taken place during the past year. In addition there are two medical items covering *Oesophageal Varices* and the *Genetics of Wilson's disease* and an article from Dr Walshe reminding us of the importance of being aware of and avoiding foods that are high in copper. Valerie has put together a table showing the approximate copper content of various foods, using as her main source **McCance and Widdowson's** *The Composition of Foods* (*1978 ed*.) She is keen to point out that it is **not** an exact science, but it does at least give us a rough guide. And last, but not least, we have the result of the 2012 caption competition, and the exciting news of the recent marriage of one of our committee members. Can you guess who?

Finally, don't forget to make a note of the date of our next annual meeting and 3rd AGM, which will take place in Cambridge on Sunday, 21 July. Booking forms and the agenda are enclosed, together with membership renewals for 2013-14. Many thanks to everybody who fills them in and returns them. Hope to see you all in the Summer.

Linda

IN THIS ISSUE ...

Chairman's Report for 2012-2013

The last year has seen WDSG-UK make steady progress towards fulfilling the aims of our constitution, and in particular in raising awareness of Wilson's disease within the medical community. We made a generous donation towards the financial support for the **Wilson's Disease Centennial Symposium**, which was held on 5-6 October 2012, and we have participated in two UK government consultations on proposals for **NHS Specialised Services for Inherited Metabolic Disorders** and on the **UK Plan on Rare Diseases**, respectively. The Group has built upon its links with **Rare Disease UK (*RDUK*)**, **Genetic Alliance UK** and the **British Liver Trust (*BLT*)** as well as continuing to work with ***CLIMB*** (Children Living with Inherited Metabolic Diseases) and ***CLDF*** (Children's Liver Disease Foundation.)

Communication with members and others, whose lives are touched by Wilson's disease, is now achieved mainly by e-mail, either directly or *via* Facebook, and I owe a special thank you to **Linda** and to **Valerie** for all the advice, support, and hard work they continue to provide through these media. I should also like to thank the rest of the committee, **Jerry, Anne-Marie** and **Mary Fortune** (co-opted last summer), for their input and support over the past year.

1. WDSG-UK Meetings

Dr Richard Sandford at the July 2012 meeting

The WDSG-UK management committee met in Knowle, near Birmingham in November 2012, and we held the second WDSG-UK AGM as part of our annual meeting for members, family and friends in Cambridge on 15 July 2012. The genetics of Wilson's disease are a particular concern for our members, and so we were very pleased that **Dr Richard Sandford**, Honorary Consultant in Medical Genetics at Addenbrooke's Hospital, Cambridge was able to give a presentation on this subject as part of our July 2012 meeting. Dr Sandford has written an article based on his talk, which may be found on *pp16-17* of this issue.

2. Donations and Fundraising

I should like to thank members, their families, friends and sponsors for the magnificent additional income of over **£2,500** which they brought in through donations and fundraising in **2012-13.** In addition, I am grateful to Univar for continuing to sponsor us so generously. And to those of you who have completed Gift Aid forms allowing us to claim back an additional **25p** in the **£1.00** on your donated money, I am pleased to report that last year we received a refund from **HMRC** of over **£250.00** on gift aided money from the previous financial year (**2011-12**.)

3. UK Government Consultations

Changes are taking place in the National Health Service (NHS) at present, which are likely to affect the way rare diseases are treated in the future. April 1st 2013 marked the official start of the work of the NHS Commissioning Board. The Commissioning Board has initiated a consultation exercise on the provision of specialised services within the NHS, and has also issued a document (E6a) on 'Specialised Services for Inherited Metabolic Disorders [IMD] (Adults)', which sets out a plan for the establishment of Specialised IMD Centres for the treatment of these diseases. Prompted by a request from **Genetic Alliance UK**, we took part in the consultation. This input allowed us to make specific points related to the diagnosis and treatment of Wilson's disease. Our detailed reply was organised and written by **Jerry** (Tucker), Vice-Chair, WDSG-UK, and a copy of Jerry's report is available on request.

Genetic Alliance UK
Supporting. Campaigning. Uniting.

In last year's Newsletter (*pp 14-15*), I described the UK Rare Disease Plan and the consultation on this plan initiated by the Department of Health. **Rare Disease UK** organised a meeting in London on 3 May 2012 to help its members respond effectively in the consultation exercise, and **Valerie** and I attended this meeting. WDSG-UK sent its response to the consultation in May 2012, and again our report is available on request. Clearly, the recommendations in the UK Rare Disease Plan, if accepted by the government, will be subsumed into the organisation and operation of IMD Centres. At present, WDSG-UK is continuing a dialogue with clinicians and others about the services we would like to see offered to Wilson's disease patients and their families. The picture is therefore a little unclear at present, but we hope to report more precisely on the changes to the NHS and how these affect the treatment of Wilson's disease, in next year's newsletter.

RARE DISEASE | UK

4. NHS Blood and Transplant *(NHSBT)* Meeting and British Liver Trust *(BLT)* Meetings

On behalf of **WDSG-UK, Valerie** and **Mary Fortune** (a **WD** patient and liver transplant recipient) attended an **NHSBT** strategy meeting in Birmingham in July 2012. They, and representatives from other patient organisations with interests in the transplantation of liver, kidney, heart, lung, cornea and bowel, were asked how they thought **NHSBT** could best canvass their views on any future decision making policies relating to organ transplantation. The meeting was also attended by **Andrew Langford**, CEO of ***BLT*** and **Catherine Arkley**, CEO of ***CLDF***, who have between them organised a further two meetings for liver patients' groups in London this year, which **Valerie** and **Mary** have attended, and at which **NHSBT**'s proposed new liver transplant allocation system was discussed at length.

5. *BLT's* 4th Annual Patient Support Groups' Representatives' Conference

Valerie and **Linda** represented **WDSG-UK** at this two-day meeting in Daventry last September and used the opportunity to network with other patient representatives and publicise the Group. Of the talks that were given, the most enlightening was from **Andrew Langford** who reported that in the absence of a national liver strategy in the UK, ***BLT*** was running its own publicity campaign called ***Love Your Liver.*** In January 2013 it would be taking mobile fibroscan units to five cities in the UK and inviting the public to attend for free liver screening. He reported that of the adult patients who were screened in the ***Love Your Liver*** campaign of **2012**, **1** in **4** was found to have some form of liver abnormality. This method of screening could pick up undiagnosed liver disease such as cirrhosis due to Wilson's disease in the future.

6. Wilson's Disease: A Centennial Symposium: 5–6 October 2012

An opportunity for the UK medical community to commemorate the centenary of the first publication on Wilson's disease was taken last October in the elegant surroundings of the Royal College of Physicians, London. About 150 delegates representing clinicians, scientists and patients' groups from the UK, Europe, USA, Canada, and India attended this two-day meeting, at which eminent speakers covered the history of Wilson's disease, its various clinical presentations, diagnostic testing, treatments, molecular biology and genetics. An extended summary of the proceedings of this event will shortly be added to the list of **WDSG-UK** articles on our website – please visit http://www.wilsonsdisease.org.uk/WDSG-P2e.asp

The organisation of an international meeting is not an easy task. Much of the work was undertaken by **Dr James Dooley** over a twelve month period with the assistance of **Geoffrey Bowden** and **Simon Williams** from the Secretariat of the British Association for the Study of the Liver (**BASL**).

7. EuroWilson

As part of the Centennial Symposium, *EuroWilson* held a short meeting for patients' representatives. The meeting was hosted by Dr Jean-Marc Trocello and Emeline Ruano who presented the results of the second *EuroWilson Patient Questionnaire*. The design of this questionnaire had been discussed at a meeting for patients' representatives held last year in Munich (see *pp 8-10* of the WDSG-UK Newsletter, April 2012 for details). The results of the questionnaire are now available for download on the *EuroWilson* website at http://www.eurowilson.org/en/home/index.phtml

8. WDSG-UK Annual Meeting and 3rd AGM

The 2013 Support Group Meeting has been arranged for **Sunday, 21 July 2013** at the clubhouse of the city of Cambridge's Rugby Union Football Club. During the course of this meeting the **3rd WDSG-UK AGM** will be convened. An agenda for the AGM is also included. As part of the AGM, the election of officers and members of the **WDSG-UK** Management Committee for the year 2013-14 will take place. All members of the current committee have submitted their names for re-election for this period.

This report has reviewed the activities of WDSG-UK during 2012 and the first part of 2013. Through the combined efforts of your committee, WDSG-UK members, family and friends, we have tried to meet the objectives of the WDSG-UK constitution, at least in part, and we will continue to do so in the future.

Rupert Purchase,
March 2013.

Wilson's Disease Support Group Meeting & 2nd AGM

Cambridge Rugby Union Football Club, 15 July 2012

For once the summer rain had relented and Sunday, 15 July turned out to be a relatively dry day for the WDSG-UK Annual Meeting. Our venue for the fifth consecutive year was the Cambridge Rugby Union Football Club where **Linda** and **Valerie** welcomed the many Group members, old and new, including for the second year **Allie Johnston** and her mother, **Rita**, from Edinburgh. Also, we were pleased to be joined by **Malin Lundberg** and her friend **Camilla** who had travelled all the way from Stockholm to be with us and to welcome **Dr Walshe's** daughter, **Susan**, to our meeting for the first time.

Linda Welcoming Everybody

The Group's links with Addenbrooke's Hospital, Cambridge, remain strong. Current staff members **Dr Bill Griffiths**, Consultant Hepatologist, **Dr Richard Sandford**, Honorary Consultant in Medical Genetics and **Dr George Mells**, Clinical Reader in Hepatology, attended together with **Dr John Walshe** and **Kay Gibbs**, who were previously attached to the Department of Medicine at Addenbrooke's. Our medical advisers were augmented by the presence of **Dr James Dooley**, Consultant Hepatologist at the Royal Free Hospital, London.

Emma Colcott and her mum, Lesley

The morning's proceedings began with a welcome from **Linda** followed by a summary by **Rupert** of the Group's involvement over the last year with *EuroWilson* and with *Rare Disease UK (RDUK.)* Following Rupert's talk, **Dr Richard Sandford** gave an eloquent presentation on the genetics of Wilson's disease and the role of molecular genetics as a diagnostic tool for detecting mutations in the *ATP7B* gene. He summarises his talk on *pp **16-17*** of this newsletter.

George Mells - deep in thought!

Lunch was followed by a raffle (organised by **Belinda Diggles**) which raised **£87.00** for the Group. And **Linda**, who had spent the winter and spring growing fresh produce, taking cuttings, making jams, pickles, cakes and greeting cards for a stall she had planned to run on our behalf at her local garden fete the previous Saturday (but which was cancelled as the ground was waterlogged) instead sold her produce to us and raised **£50.00** for the Group.

There was a short AGM in which the current Management Committee was unanimously re-elected for the year **2012-2013** and a new committee member and patient, **Mary Fortune**, was co-opted. Univar's generous support of WDSG-UK was re-emphasised by a donation of **£1,000** presented by **Ray Estall**. Then to our great delight, **Allie Johnston** came forward and presented the Group with a cheque for **£500** (see Allie's Story on *pp **10-11***.)

Malin and Camilla from Sweden

WDSG-UK President **Dr John Walshe** completed the day's presentations with an account of the treatment of Wilson's disease. This successful meeting concluded with questions for the medical consultants followed by Group photographs, taken inside the clubhouse this year (by **Barry Diggles**) against the backdrop of the flooded rugby fields outside.

Rupert receiving a cheque from Allie

Group Photo

FUNDRAISING 2012-13

Sponsored Walk

Philip and Tulin Hawkins

Philip - full of enthusiasm!

Philip and Tulin Hawkins, whose daughter was diagnosed with Wilson's Disease at university fourteen years ago, began the year's fundraising events with a walk at Easter along the Thames Path from Oxford to London. In so doing and through the generosity of their sponsors, they raised the fantastic sum of **£410** for the Group. Philip's account of their trip appears overleaf *(p6.)*

Sponsored Skydive

Emma Coombes

Emma - no going back...

On Friday, 6 July, **Emma Coombes** from Torquay took to the clear skies over Honiton in Devon and fulfilled a lifetime's ambition to skydive. Watched from the ground by her mum **Sheila**, sister **Daisy** and son **Tom**, **Emma** who was diagnosed with Wilson's disease seven years ago, said it was the most amazing thing she had ever done.

With the support of her friends, neighbours and family she raised **£180.60** for the Support Group.

Coffee Morning and Bring and Buy

Sylvia Penny

L to R: Emma, Amy (Emma's daughter), Sheila, Tom and Daisy

On Saturday, 18 August our regular fundraiser **Sylvia Penny** from Devon, (**Emma Coombes'** grandmother) held yet another coffee morning at Victoria Park Methodist Church in Plainmore, Torquay, with cake and bric-a-brac stalls and a raffle. She raised **£366.26** for Group funds.

We are very grateful to her for her continued hard work. Sylvia has since had a hip replacement and we send her our best wishes for a full and speedy recovery.

Asda St Helens and Xmas Puddings

Belinda Diggles

Belinda receiving cheque from Asda on behalf of WDSG-UK

In Autumn last year **Belinda Diggles** from St Helens nominated WDSG-UK at her local Asda (where she works) as a cause worthy of consideration for a charitable donation from the Asda Foundation. Three charities were shortlisted and customers voted for their favourite. **WDSG-UK** proved to be the most popular and Belinda was presented with a cheque for **£50.00** on our behalf.

At the same time she was busy making Christmas Puddings, which she sold to friends and family raising a further **£82.00** for the Group.

Lathom Golf Society

Barry Diggles

Barry, in full swing

Meanwhile, Belinda's husband, **Barry Diggles**, was also helping the Support Group out last year. Having been appointed captain of the Lathom Golf Society for the 2012-13 season, he very kindly nominated **WDSG-UK** as his chosen charity for the year.

As a result he was able to send us a colossal cheque for **£840** recently, being the amount he had raised during his captaincy through golf outings for the members and by organising other Society fundraising events.

Walking the Thames - Oxford to London - April 2012 by Philip Hawkins

In April last year Tulin and I are on the Thames Path into our first day of walking from Oxford to London. At this point we meet a runner coming the other way. After the customary greetings, the following conversation ensued.

Runner: "So, what are you doing?"

Us (smugly): "We've just walked fifteen miles from Oxford."

Runner: "Where are you going?"

Us (more smugly): "London."

Runner: "Oh, I did that the other day."

Us (crestfallen): "Oh?"

Runner: "Yes, I ran it in x hours." (Note: can't remember the exact time, but it was much less than twenty-four hours.)

Us: (more crestfallen) "Oh!"

Nevertheless, we continued for the rest of the day, almost speechless. But not really. However, it has to be confessed the challenge we had set ourselves – Oxford to the Thames barrier in four days – was pretty much impossible. On our first night in the lovely small town of Goring and Streatley, our landlady at the B&B, where many walkers have stayed over the last 25 years, told us the most taxing daily average she'd ever heard of was 17 miles a day. I had nonchalantly calculated from the safety of a comfortable armchair in winter that we could do roughly 15 miles in the morning and 15 miles in the afternoon for each of the four days. This adds up, of course, to 30 miles a day, 120 miles in all.

So, I can't say we didn't extend the notion of 'walking' the Thames path by occasionally taking the train along short stretches of line which parallel the path and sort of walking along the train.

More seriously, Tulin got quite ill by the end of the second day and so had to 'retire' from the ordeal, a plight she has quickly recovered from, I hasten to add. I carried on at least until the outskirts of the capital. If any of our sponsors were tempted to query our achievement, then all I can say is that we certainly felt the pain of the experience. But we are grateful to all those who 'believed' in us!

On a more positive (and truthful) note, we enjoyed ourselves immensely. Tulin and I know the path quite well, having walked most of it in (manageable) sections in the past. It rained or drizzled quite a bit but this proved much less of a problem than we had expected. Otherwise, the Thames is never dull and always varied both in terms of landscapes and history. We love it! Certainly, if you want to raise money for WDSG-UK, I can't recommend a better way. Just avoid runners with a clearly superior attitude! (And allow for more time.)

Tulin - taking it all in her stride

The Thames' Blisters arrive

Oh for a bicycle...

David's Story

by David Groome

David around 1986

I was born in Edmonton, Middlesex, in January 1950 (I know that because I was there.) I had an uneventful childhood, moving from Edmonton to Nazeing, Essex, where I lived in a cottage by the River Lee and enjoyed the summers fishing and swimming in the river.

Fast forward a number of years to 1986, to be exact, when I was married with children. I was working for a leading electrical wholesaler, when I had my first *attack*. I was on the 'phone and suddenly couldn't talk or move, although I knew what was happening around me. I was rushed to Romford Hospital where the medical staff diagnosed me with having *epilepsy*.

All was fine for a number of weeks. I was taking medication and had no further *attacks*. But then the *attacks* returned one after another, sometimes up to ten times a day, which left me exhausted. It obviously wasn't *epilepsy*, so I was referred to another specialist who went through extensive tests before diagnosing me with *Benign Essential Tremor* (now known as *Essential Tremor*) for which I was treated with a course of beta blockers. The *attacks* became more frequent and my employers not being at all sympathetic, politely informed me that I was no longer needed. My condition was worsening and now I was also out of work.

My GP then referred me to the National Hospital for Nervous Diseases and I underwent far more extensive testing. My eyes were checked with a slit lamp after which everybody became very excited. *Kayser Fleischer* rings had been seen which meant that I probably had *Wilson's Disease* with excessive copper in my body. I thought to myself, "How on earth could copper cause all these problems?"

At least I had got a formal diagnosis and wasn't suffering from either *epilepsy* or *Benign Essential Tremor*. However, I was told that I would have to take medication for *Wilson's disease* for the rest of my life and was prescribed *penicillamine* to chelate the copper. It later transpired that the dose was too high for me as I was bruising very easily and if I cut myself it took a long time to heal. After an accident once I attended the A&E department at Homerton Hospital, Hackney and was found to have a seriously low platelet count. I was given a transfusion of platelets before having my medication swapped from *penicillamine* to *trientine* (which I stayed on until 2009.)

In late August 1988 my health had improved so much that I applied to be an Administrative Assistant working for HM's Government in the Home Office. As a civil servant I held various positions before settling in the Immigration and Nationality Department, where I stayed until retirement in March 2010.

My American Adventure

On 21 March 2010 my wife, Diana, and I embarked on our American adventure. The flight was uneventful and the weather was gorgeous. We could see New York City and Philadelphia clearly from the plane. Landing at Baltimore airport, I found myself a stranger in a strange land where people drive on the wrong side of the road and speak a language which is similar to ours, although the spellings are often different. Everything here is on a grand scale because the people love big things, particularly the cars, trucks (known as pick-ups) and portion sizes of food! But I do like it over here and have a part-time job as a cashier at Walmart, which is a story in itself.

David today

I miss the *National Health Service* as health care here appears to be a lottery, which I still don't fully understand and I've been here nearly three years. It seems to be based on how much or how little insurance you have whether or not you get the best possible health care. If your employer is willing to pay full costs towards your health insurance then you have it made, but if you have an insurance with some kind of deductible e.g. the company pays a certain percentage and you have to find the rest, then it can be expensive.

I also miss my family (although I am in regular contact with them on *Skype*) and of course, I miss you guys. I enjoy following you on *facebook* and hope that your annual get together goes well in the summer. Don't forget to embrace modern technology so that I can be there with you.

A rather large truck, but some are even larger than this one

I could have and would have written more, but perhaps I'll leave that for my book!

Oesophageal Varices - A Simplified Guide

A.Stevens

What are Oesophageal Varices?

Oesophageal varices (pronounced "varry-sees") are abnormally dilated veins mainly confined to the bottom end of the oesophagus, where it empties into the stomach. For simplicity the diagram shows only three, but there may be many more. These veins lie just underneath the internal lining (***mucosa***) of the oesophagus (*see* ***Diagram 1(b)***) and when enlarged into varices they bulge into the space (***lumen***) of the oesophagus, as shown in ***Diagram 1(c)***.

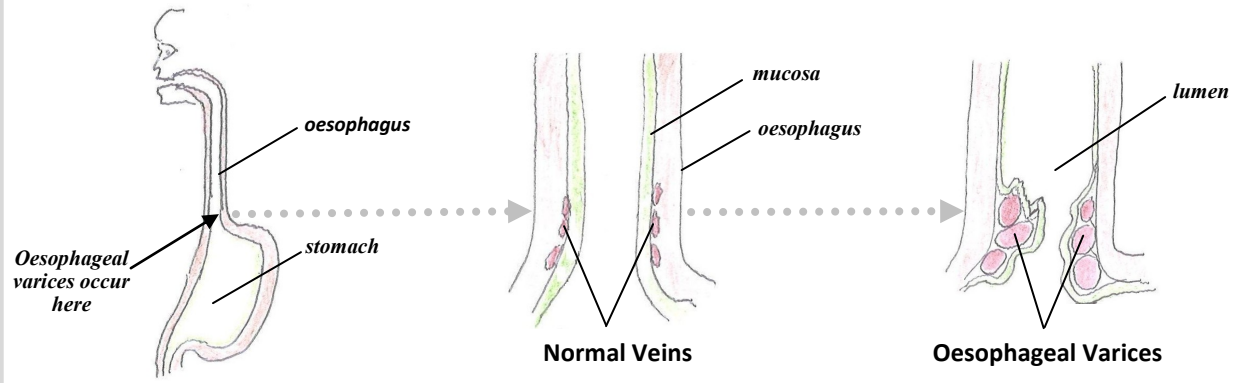


Diagram 1

Why Do the Veins become so Big?

The veins are part of a special closed system of veins called the **hepatic portal vein system**, which runs between the gut and the liver with a branch joining from the spleen. The hepatic portal veins contain blood which is rich in vital chemicals such as amino acids, fatty acids and simple sugars, derived from the breakdown of food in the gut; they also contain vital proteins and other substances from the breakdown of old red blood cells in the spleen. The veins carry these chemicals from the absorptive part of the gut (stomach, small and large intestines) and spleen to the liver for processing. The blood pressure in these veins is normally low, but oesophageal varices can occur when the blood pressure rises to higher levels. High blood pressure in the hepatic portal vein system is called **portal hypertension**.

Would my Doctor spot Portal Hypertension when I have my Blood Pressure Checked?

No. The blood pressure your doctor measures is the blood pressure in the ***arteries***, not veins. The blood pressure in the arteries is much higher than in veins because it is pushed along by the powerful pumping action of the heart, whereas the low blood pressure in veins is largely created by contraction of the thin layers of muscle in the vein wall, much weaker than the force provided by contraction of the powerful heart muscle.

What Causes Portal Hypertension?

High blood pressure in the portal vein system is due almost always to chronic liver disease. Here is how it happens (*see* ***Diagram 2***)

Diseases which cause damage to the liver over a long period of time lead to a pattern of scarring called **cirrhosis**. Normally, the portal veins empty their blood into channels throughout the liver, but when there is cirrhotic scarring many of these channels get blocked or distorted and the portal veins cannot empty their blood efficiently. As a result, there is back-damming of blood and the pressure in the portal vein tributaries rises. At the lower end of the oesophagus, this persistently high pressure can manifest as **oesophageal varices**.

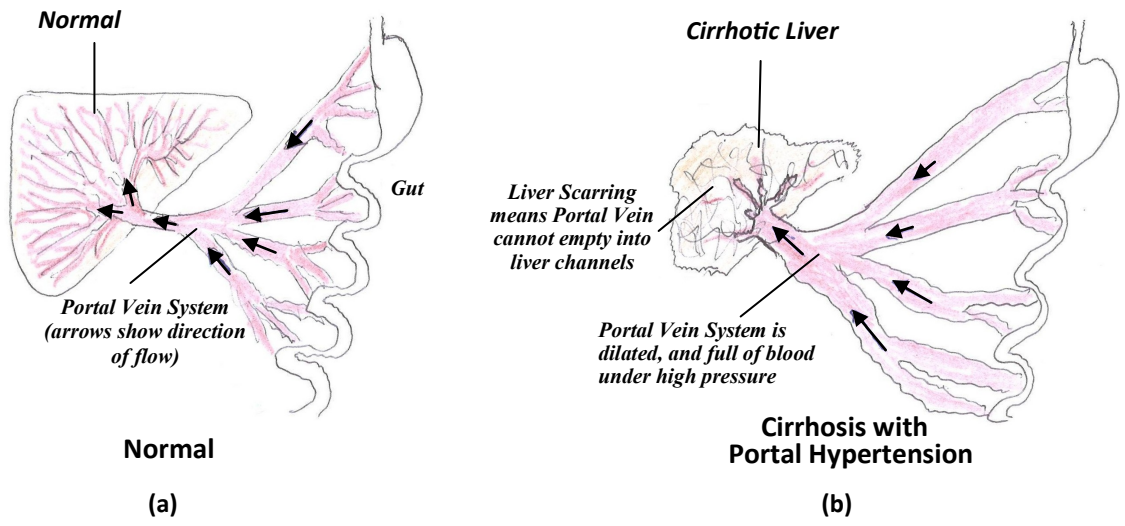


Diagram 2

What has all this got to do with Wilson's Disease?

Any disease which can cause **cirrhosis of the liver** can cause oesophageal varices and Wilson's disease is no exception. However, it remains a rare cause, alcoholic cirrhosis of the liver is the most common cause in the UK.

So What is the Problem with Oesophageal Varices?

The main danger is that they can rupture, leading to bleeding which can be difficult to control. One of the possible factors leading to rupture is the destruction of the overlying mucosa by stomach acid during acid regurgitation into the oesophagus.

How can Oesophageal Varices be Prevented and Treated?

Once the pressure in the portal vein system rises above a certain level, oesophageal varices cannot be prevented. The main aims of treatment are to prevent them bleeding in the first place and, if they have already bled once, to prevent them bleeding again. There are three main treatments:-

1. **Beta blocker drugs** (such as *propanalol*) can be given to lower the pressure in the portal vein system and reduce the chances of bleeding;

2. The oesophageal varices can be ligated (tied off) by special elastic bands **(banding.)** This is done via an endoscope which is passed down the oesophagus. **Banding** can be used to treat oesophageal varices which are already bleeding, and to prevent further bleeding episodes;

3. **Liver transplantation** cures oesophageal varices. The open channels in the transplanted liver allow the hepatic portal vein system to empty its blood normally without obstruction, so that the portal vein pressure falls to a normal level and the varices collapse. Transplantation is not performed just for this reason though. One type of emergency treatment is a 'TIPS' (transjugular intrahepatic portosystemic shunt) which is a tube placed, under radiological control, across the liver to divert blood from the portal vein into the main venous circulation away from the varices.

Any patient with **cirrhosis** should have an **endoscopy** to look for **oesophageal varices**. Small varices do not need treating but larger ones do - beta blockers are tried first, with banding left in reserve as a second line method as it is more invasive, with more potential complications.

An episode of bleeding of varices can be precipitated by alcohol ingestion (particularly binge drinking) and vomiting, **so best avoided**!

Allie's Story

by Allie Johnston

I was born in Edinburgh in **April 1970** and have a sister, Margaret, who is four-and-a-half years older than me. We get on really well together. I had a happy childhood, although when I was nine years old my father fell and broke his neck and was in hospital for almost a year. As my sister and I were both at school, during term time we could only get to visit him in the evenings and at weekends.

My sister, Margaret and I in 2008

I enjoyed school but was not particularly academic. I had lots of friends and loved sport, playing tennis and basketball. I have two claims to fame - one is swimming in a charity event with David Wilkie (British 200m Breast Stroke Gold Medallist in the 1976 Montreal Olympics) and the other was appearing on television with Rolf Harris, when he came to visit our school.

Pre-Diagnosis - 2009

I left school after O'levels and went to work for a firm of solicitors attending court and delivering offers on properties. In my next job with a copier company I was responsible for looking after the records of its 200 employees in their BSI library. And up until my illness I was working for a builder letting and selling units and houses. I have always been a loyal employee and was very seldom ill. I drove a car, owned my own flat, enjoyed socialising after work and took various holidays. I also loved cooking and entertaining. When I was twenty-seven I married my boyfriend of nine years and had a traditional white wedding with four bridesmaids. Unfortunately, the marriage didn't last and we ended up going our separate ways.

Zip Wire - not for the faint-hearted

By early **2009** I had become really unwell. My mother, Rita, thought a cruise to the Caribbean might do me good. Apart from flying over a tropical rain forest in Colombia along a zip wire (which was brilliant), I was getting no better and on our return to Scotland I visited the G.P. who sent me to hospital for blood tests. My liver was found to be enlarged. I was by now having difficulty walking, talking and swallowing and on top of that I was drooling incessantly and was doubly incontinent. I had trouble getting myself dressed and I just wanted to sleep all the time. I had gone from being an extrovert to a pathetic woman who needed to get around in a wheelchair.

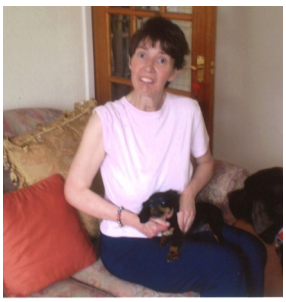
At the end of **2009** and the beginning of **2010** I had been seen at different hospitals by various doctors including a gastroenterologist who did a biopsy, an ENT specialist who noted that although I couldn't speak my voice box was in tact, a neurologist and finally an ophthalmologist. When the ophthalmologist found *Kayser-Fleischer* rings in my eyes, I was given the diagnosis of *Wilson's disease*. But instead of things getting better thereafter, they only got worse. I had patches on my neck to curb the drooling and this led to me being catheterised. By **April 2010** I was unable to eat and was admitted to hospital to have a peg inserted into my stomach, so that I could be fed at night through a pump. I remained in hospital for one month and by the time I left I had a weeping wound on my coccyx from a bed sore, which has only just cleared up three years later.

The consultant wanted me to be admitted to a Rehabilitation Centre for people with head injuries, but my mother just wanted to take me home to nurse me herself. Although I couldn't speak, I communicated by pointing to letters on a board and said, "Please don't send me to another hospital." I was allowed home on condition that I attended the centre as a day patient from Monday to Friday. I was taken to and from the centre by taxi which was a long and uncomfortable process. It was the most depressing place I had ever been. The other patients were badly brain damaged and I felt very isolated. The physiotherapist, speech and language tutor and psychologist all seemed to think I was being deliberately unco-operative, but I had neurological *Wilson's* which had affected all my muscles and it seemed that nobody had had experience of dealing with it before. I will be grateful to my mother until the day that I die for refusing to allow me to be an inpatient there.

Three weeks later the ulcer on my bottom became severely infected and I was readmitted to hospital as an emergency. I ended up staying there for the next six months until **December 2010**, when the wound was finally brought under control. During this time my treatment included *penicillamine*, then *dimercaprol* injections (which are unbelievably painful), and finally *trientine*. I was still unable to eat normally, but now my feeds were being given to me through eight syringes a day which had the advantage of my mother being able to administer them. However, one day when looking at the content of the boluses I was being fed, my mother was dismayed to discover that they contained copper. She heard about a Southampton biochemist who had developed a copper free diet for another *Wilson's* patient, and she contacted her for information about the formula.

By December 2010 I still could not speak or eat and my head continually lashed backwards and forwards, but I had started to walk again with the aid of a *zimmer frame* and thankfully I was allowed to go home. The entire country was blanketed in snow, but it was wonderful to be back in my own cosy surroundings. However, my relief was short-lived as I began to have frightening episodes when my teeth clamped down on my tongue and I was having difficulty breathing. On the second such occasion I was rushed to hospital and treated under anaesthetic, but on the third occasion my tongue had swollen so much that I had to have my bottom teeth removed to release it. I was in hospital for a further two weeks and was put back on *penicillamine*. On discharge my mother made a promise that from then on there would be no more nurses, hospitals or ambulances in our lives.

For my birthday in **April 2011** I was given a wee puppy that was two months old. It was a black and tan *Cavalier* and it was absolutely adorable. However, exercising it proved too difficult for us in the end and my sister very kindly found a new home for him. Shortly afterwards my mum and I were sitting in the garden one day, when she popped back into the house and absentmindedly locked the door behind her. I couldn't get back in and despite hammering on the door, she couldn't hear me. When she later realised what she had done she rushed out and apologised. I replied, "You numpty, locking me out!" It was the first time I had spoken in eighteen months and we were both overjoyed.



The black and tan puppy

Now I had my voice back I could at least speak to my mother and tell her how I was feeling. Before that she had relied on the expressions on my face to gauge how I was. In **July 2011** we travelled to Cambridge to attend our first Support Group gathering. We met **Dr Walshe** in person, my mother and sister having only spoken to him by telephone before then, and it was lovely to meet other patients there, too. In **December** that year our community dietician finally made the copper free diet recommended to us by the Southampton dietician twelve months earlier and I was pleased I was able to prepare it myself. In **May 2012** I finally had the peg removed, which meant proper food again and no more syringes, hooray!

We travelled to Cambridge for our second **WDSG** meeting in **July 2012**. My employer had kept my job open throughout my illness, but in the summer of **2012** I was made medically redundant. I received compensation and through the proceeds was delighted to present a cheque to the Group. I rehearsed a short speech, but unfortunately fluffed my lines which really upset me. However, I really enjoyed the meeting and in particular savouring the taste of the strawberries and cream which I had missed the previous year when I still had my peg in!

Since I have been home my mum has arranged for various therapists to come to the house including an acupuncturist, physiotherapist and speech therapist. I also have a support worker from *Action Group* who helped me get a volunteer's job in a charity shop. Through her and my own efforts I currently work four shifts a week in two different shops. Last Christmas my mother also arranged to take me and my sister and her husband and two children (aged 16 and 12) to London for a short break. It was her way of saying *thank you* to my sister for all the help she has given us in the past three years. It was a very tiring but enjoyable holiday. The last time I was in London many years ago, I had been travelling on a bus when I looked out of the window and saw Princess Diana driving an open topped car, which had pulled up alongside us. She had given me a wave and a lovely smile, which I shall never forget.

Margaret and I in 2011

Wilson's has attacked all the muscles in my body and the ones in my mouth in particular, making it still difficult for me to eat, swallow and speak, and I still have an uncontrollable tremor in my head, which is absolutely exhausting. However, as far as my other neurological problems are concerned, it's a case of wait and see. My right hand is turned in like a claw, but the *physio* works on it each week and I am glad to say that I can now operate the computer slowly. After three long years of recurrent infected wounds on my bottom, they have at last healed and I have been told recently that I can start swimming again. Look out David Wilkie!

My final piece of good news is that I have started to drive again. I have been out with an instructor, who praised my driving skills highly. He said I would be able to drive on my own in next to no time. I am absolutely over the moon and am looking forward to driving my niece and nephew to the swimming pool soon and taking them on holiday again with my mum. One of the last holidays we had with them was in **2006** when we stayed on a house boat on the shore of Lake Windermere and it would be nice to do something like that with them again.

The House Boat on Lake Windermere

But now I am looking forward to my trip to Cambridge on **21 July**, when I hope to see you all again at this year's **WDSG-UK** meeting.

At Home With Dr John Walshe

by Valerie

Dr Walshe has devoted his professional life to Wilson's disease and has looked after over three hundred patients during his career. He discovered *penicillamine* in the 1950s, which was the first effective oral treatment to be prescribed to chelate copper. As a nonagenarian he still takes an active interest in the management of patients and in the running of the Support Group, of which he is President.

Dr Walshe – At Home

In last year's newsletter he told us of the importance of taking *penicillamine* and *trientine* in **divided doses** during the day around **half an hour** before **eating**. Following on from this advice, I recently called by and asked him about the importance of **diet** in the management of Wilson's disease.

Is it Important for Wilson's Patients to avoid Foods with Copper?

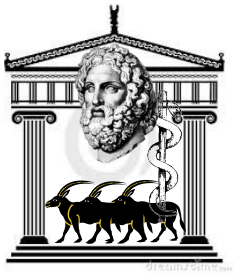
All things in moderation! It is clearly unwise to eat a lot of food with very high copper content, such as **liver, shellfish, cocoa products, nuts** and **seeds**. With **chocolate** it is the **cocoa bean** that contains the copper. The higher the **cocoa** content of the chocolate (**up to 99%**), the greater the amount of copper. White chocolate is probably safer for those who can't avoid it altogether.

Do Long Established Patients need to be as Careful?

If you are **newly** diagnosed it is wise to observe and avoid the foods with **high/very high copper** content wherever possible. Once your treatment is **well established**, you can be more relaxed about what you eat, but try to avoid eating any of the **high/very high** copper foods **in excess**.

Should I be trying to Avoid Copper in the Diet altogether?

Remember that **copper** is a trace element **essential for life** and it is neither possible nor desirable to live on a copper free diet. Long term de-coppering can, but does not always, result in the skin losing its elasticity, because it removes the copper from the enzyme which forms the cross linkages in collagen and elastin.



Can Copper be absorbed through the Skin?

I know of no evidence to that effect. There certainly are no reports of copper miners suffering from copper poisoning.

So Why do People wear copper bracelets to relieve rheumatism?

Why did the Ancient Greeks sacrifice goats to Aes**cu**lapius?

Is it OK to drink alcohol when you have Wilson's Disease?

Alcohol damages the liver so it is very unwise to add further insult to that caused by copper. The combination of **alcohol** and **copper** on the liver is **DANGEROUS.**

Summary

- **All things in moderation, and try to avoid eating high copper foods in excess**
- **Copper is essential for Life. It is neither possible nor desirable to live on a copper free diet**
- **The combination of alcohol and copper on the liver is DANGEROUS.**

An Approximate Guide

FOOD TABLE: Copper (Cu) Content in mg per 100g Portion

LOW <0.2 mg Cu/100g	mg	MEDIUM 0.2–1.0 mg Cu/100g	mg	HIGH 1.0–3.0 mg Cu/100g	mg	VERY HIGH ≥ 3 mg Cu/100g	mg
Meat		**Offal:**		Liver Pâté (depending on animal...) →		**Liver:**	
All fresh, frozen & tinned meat;		Brain – calves'	0.42			Calves'	12.00
Poultry (dark meat higher in cu)		Heart – lambs'	0.52			Lambs'	8.70
Fish - White/Oily		Kidney – pigs'	0.81			Cows'	6.40
		Meat: Duck	0.30			Pigs'	3.75
		Goose/rabbit	0.50				
Dairy		**Nuts:**		**Nuts:**			
Milk: Cows'	0.02	Peanut Butter	0.70	Cashews	2.20		
Goats'	0.05			Brazil	1.74		
Butter	0.03			Pine Nuts	1.30		
Cream: Single	0.20			Pecan	1.20		
Double	0.13			Hazelnuts	1.76		
Cheese: Cheddar	0.03			Walnuts	1.60		
Yogurt: Natural	0.04			Pistachios	1.32		
Flavoured	0.08			Almonds	1.20		
Eggs:		**Dried Fruits:**		**Shellfish**		**Shellfish:**	
Whole	0.10	Peaches	0.63	Crayfish	2.00	Scallops	10.00
Yolk	0.30	Currants	0.48	Calamari	2.10	Whelks	7.00
White	0.05	Sultanas	0.35	Prawns	0.70	Oysters	7.60
Oils/Fats		Apricots	0.27	Shrimps	0.80	Crab	4.80
Cooking Oils	*Trace*	Raisins	0.24	Mussels	0.48	Lobster	2.90
Butter/ Margarine	0.04	Figs	0.24	Cockles	0.28	Clams	5.00
		Dates	0.21				
Fruits: Fresh		**Fruits:** Fresh		**Candied Fruit**			
Berries (average)	0.13	Olives	0.23	Glace Cherries	1.28		
Prunes	0.16	Avocados	0.21				
Pears	0.10	Banana	0.21	**Seeds**		**Seeds**	
Grapes	0.10	Kiwi	0.30	Sunflower	2.00	Sesame	4.10
Oranges	0.07	Lemons	0.26	Pumpkin/Squash	1.40		
Vegetables: Fresh		**Vegetables:** Fresh		**Vegetables:**			
		Mushrooms	0.78	Beans -Butter (raw)	1.22		
Root Vegetables	0.08 -0.20	Asparagus	0.20	Soy (Edamame)	1.10		
		Beans - Broad	0.43	Tomatoes			
Greens	≥0.06	Haricot (raw)	0.61	(Sun Dried)	1.40		
		Mung (raw)	0.97	(Tomato Puree)	0.63		
Salads		Red Kidney (raw)	0.61	(Tomato Ketchup)	0.40		
Lettuce, peppers	≥0.05	Baked	0.21	Brown Sauce	0.33		
		Peas (raw)	0.23	*(Bovril)*	0.45	Bakers' Yeast	
Pulses		Chick Peas (cooked)	0.33	*(Marmite)*	0.30	(dried)	5.00
Lentils, split boiled	0.19	Chips	0.27	(Stock Cubes)	0.71		
		Crisps	0.22				
		Spinach (boiled)	0.26	**Herbs:** Pepper	1.13		
		Parsley	0.52	Basil (dried)	1.40		
Pasta - dry	0.20	**Preserves/Cakes**		**Cereals**			
Semolina		Treacle (black)	0.43	Bran	1.34		
White Rice (cooked)	0.10	Jams (berries)	0.23	*All Bran*	1.20		
White Bread	0.12	Mincemeat & fruit cake	0.20	*Shredded Wheat*	*0.40*		
		Xmas Pudding	0.25	*Weetabix*	*0.54*		
Pastries		**Confectionery**		**Sweets/Chocolate**		**Chocolate Bar**	
Cakes		Liquorice	0.39	Fruit Gums	1.43	(≥70%)	≥4.00
Ice Cream		Mars Bar	0.31			Chocolate Bar (<70%)	<4.00
(without chocolate)		Bounty Bar	0.47	Drinking chocolate (sweetened)	1.10	Cocoa Powder (unsweetened)	3.90

Rosie's Story

by Razwana Faruok

Rosie

Hi. My name is Razwana but people also know me as Rosie. I'm 23 and from Bolton in Lancashire.

It has been over thirteen years since I was diagnosed with *Wilson's disease*. It has been very challenging, both physically and mentally, but if there is one thing I can say about *WD*, it is the fact that I have never really felt angry towards it. I've known people to really despise this disease and I can understand why if their condition is worse than mine, but I have never found myself thinking so strongly about it. Ok, so I may have been a little angry in the past, when I first got diagnosed, but I think that is only natural. Knowing you were getting ill, but not knowing why or if you would get better, was scary.

It was hard being around people and not being able to eat and drink what everyone else was eating and drinking. I wasn't allowed to do small, simple things like put salt on my chips, or drink hot chocolate or have mushrooms on my pizza. While my cousins were playing outside, I'd sit inside because I didn't have enough energy to play. To make matters worse, I'd have to drink a horrible medicine to keep my liver working (great way to kill an appetite.) It wasn't really so bad, but it wasn't pleasant being different from other children.

As many people with *WD* know, *Wilson's* doesn't just affect the body, it affects the mind too. It challenges you daily, physically, mentally and emotionally. I can go from feeling super happy and super active to feeling sad and apathetic. I like to think of it as a roller coaster ride and personally, I hate roller coasters!

So how did I find out that I had *WD*? I remember when I was 10 waking up one morning with a very sharp pain in my stomach. I visited my doctor and was sent to my local hospital. After two days of baffling the doctors, I was transferred to a Children's Hospital in Manchester. I spent a month there undergoing many tests until finally I was diagnosed with my lovely rare disease. It came to the point where I was having four blood tests done every day. Eventually nurses had to take blood from my feet, which was not a pleasant experience!

I never really fully understood what *WD* was or how I should manage it, until my late teens. I was bad at taking my medication. Reading about the effects of *Wilson's disease* terrified me, so I never tried to learn more about it. I didn't know anyone else who had it and my doctors were just as clueless as I was. I was under the care of a doctor who specialised in rare diseases but, nice as he was, he was learning about my condition at the same time as I was.

I was often depressed. I became very unsociable and didn't like leaving my home. I had lost all confidence due to missing a lot of school. I was just plain angry in my late teens and I didn't even know why. It wasn't the fact that I was suffering from a disease, but more that I didn't understand how to manage it properly or ask for the help that I needed. I come from a BIG family and as lovely and supportive as they were, they could never truly understand what I was going through. I feel *WD* has affected me more mentally than physically. As a teenager, I didn't think it was possible to have depression and thought I was just being silly.

However, one day when I was twenty, I had had enough of feeling the way I did. Family and friends were great but I needed somebody who understood how I was feeling. I went on to *facebook* and typed in "*Wilson's disease*" in the search box and hey presto, I came across some groups and pages dedicated to *WD*. After messaging a few fellow sufferers, I came to know how they were dealing with it in their lives. It was interesting reading their stories and comparing them with mine.

I discovered ***WDSG-UK*** not long after that. It was more active than the previous groups and pages and the support that was given on it was amazing. I had learnt more about my condition through talking to others in the Group than I had from years of talking to my doctors. The advice, support and friendly chats from other members were just great. I couldn't have asked for nicer friends. Learning more about how other people had managed to control this disease and still lead a normal life was very inspiring and had made me accept my life the way it was. I saw that I was very lucky compared to most people. I had learnt to deal with my illness better and had even had the courage to talk to my doctor about my depression.

I've never considered myself a particularly religious person but I do believe everything happens for a reason. I have been through a lot from a young age, but my experience has turned me into a stronger person today. I appreciate the smaller things in life and thanks to modern medicine and the support of friends and family my illness is manageable and I am grateful to it for making me the person that I am today.

Thanks for reading my story.

Linda's Woes

Linda's Woes

After more than thirty years of being a smoker I finally gave up last year on July 28th, the date is etched on my brain for all time! I had tried the *cold turkey* business when I went down to Cambridge earlier in the month for the Support Group meeting. I was quite successful, surprisingly enough, and after three days wasn't particularly missing cigarettes, but the problems arose back in Nottingham when I was around friends and family who still smoked. I thought, "I'll have *just the one*," but of course it never is *JUST the one*. I was so angry with myself, but smoking being the addiction that it is, it didn't stop me. A friend suggested I try the *New Leaf* service to help me quit, and also managed to convince me that there was no shame in seeking help. It worked and I've never looked back, plus I made a new friend in the *New Leaf* counsellor.

Four weeks post op. and down to one crutch! First trip out to Attenborough Nature Reserve.

It does seem odd to me though that after stopping I seemed to be plagued with irritating health problems; abnormal liver scans, a peculiar stomach bacteria that took barrow loads of antibiotics to kill and then of course the problems that large amounts of antibiotics cause! But the real *biggy* was having a total knee replacement in early February. Arthritis runs in our family and I have had problems with my knees for more years than I care to remember. I think the first twinges were at least twenty years ago. I've tried everything to avoid having surgery, (yoga, tai chi, heat treatment, osteopathy and acupuncture) but when walking became just too painful I finally bit the bullet and agreed to have it done.

The date duly arrived and I turned up at the hospital at the appointed hour – 7.00 a.m. Operations start at 8.00 o'clock and I was to be either the first or second on the list. Well, that was the plan anyway! But then there was a hitch, when somebody pointed out that I had Wilson's disease and somebody else suggested that I should have a blood test done to check that I didn't have any clotting problems. So I ended up instead going to theatre at noon, by which time my stomach thought that my throat had been cut and I was starting to plot my escape.

Looking back, it was quite funny really as it reminded me of ages ago when I was in a similar situation. I had gone back into hospital a year or two after I'd broken my leg quite badly in a motorbike accident, to have a metal plate and pins removed. Again, I'd arrived at the hospital very early only to be told the operation would be delayed until late morning. I settled down to wait and got chatting to an elderly lady who was trundling around the ward on a *zimmer frame,* on the front of which she had a little bag with bits of metal in it. When I asked what the matter was she said that she *had been fine* but then they had taken the plate and pins out of her leg and now she was having to learn to walk again! I LEFT very quickly and still have the metalwork in my leg to this day!

But I digress...back to the knee replacement. I'd elected to have a spinal rather than a general anaesthetic, the main reason being I was told it provided longer lasting pain relief after surgery. The injections were fairly painless and I was also going to be lightly sedated during the operation. It was the strangest dreamlike sensation; I remember coming to a little and seeing a big green sheet in front of me and then hearing the most awful noise coming from outside. "Roadworks," I thought, "how annoying for everyone!" I mentioned this to the surgeon the next day and he laughed and said, "That was the sound of the operation going on. It's VERY noisy!" He then accused me of having blunted his *Black and Decker*. Had I been able to get out of bed, there would have been trouble!

My recovery has been slow but sure. One of the biggest problems I found was *cabin fever*. It drove me absolutely mad not being able to get out of the house. With the heavy snowfalls that we had, even walking down the garden path on my crutches was out of the question. After the first week or two the pain gradually got less and I was able to start cutting down on the painkillers and spend less time *away with the faeries*. I've progressed to one walking stick and at last I can see the light at the end of the tunnel (let's hope it's not a train heading this way!)

Hopefully Val and I might even manage a few days' holiday again this year.

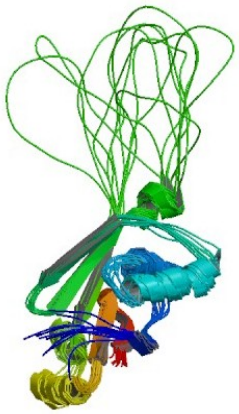
Wilson's Disease is a Genetic Condition…

Dr Richard Sandford

"Wilson's disease is a genetic condition" is a statement that will be very familiar to most people reading this newsletter. But what does *genetic* mean? *Genetic* is often assumed to mean the same as *inherited* and *inherited* to mean *runs in families*. But *genetic* in the context of Wilson's disease tells us something more. It tells us about the cause of the disease, why it is rare, why it can be so variable between different individuals as well as why it can affect several members of the same family. In my work as a clinical geneticist, I hear many families express the same concerns following the diagnosis of a genetic disease. These commonly include, "How will the disease affect me?" "Is genetic testing available?" and "Are other family members including children at risk of developing the same condition?" Understanding the genetic basis of Wilson's disease can help us answer these and other important questions.

Wilson's disease is caused by mutations, or genetic alterations, in a specific part of our genetic make-up or genome. This part is a gene called ***ATP7B***. But what do we mean by genome and genes? Our genome is made up of **DNA**, which is packaged into chromosomes that can be compared to a set of encyclopaedias (remember those!?) It contains all the instructions needed to make a healthy person. Each one of these instructions or genes, (we have about 20,000 of them), has a unique role. This set of encyclopaedias (our genome) contains 23 volumes (the chromosomes.) Most human cells have two copies of each chromosome, i.e. they exist in pairs and therefore we have two copies of each gene. One copy is inherited from each parent. Chromosomes **1-22** are called autosomes (this will become important later) and the other two, the **X** and **Y** chromosome, are called sex chromosomes. We all have all the autosomes, whilst women have two **X** chromosomes and men one **X** and one **Y** chromosome.

Our **DNA** is made up of four letters or bases: **A, C, G** and **T**. In total our genome contains 3 billion of these letters with a chromosome containing from 46 to 250 million letters depending on its size. A gene, or set of instructions, can therefore be thought of as one page in one volume of the encyclopedia containing thousands of letters. The gene functions normally when all the letters on the page are *read* correctly. When a letter is changed or is missing (think of a spelling mistake) then the instructions cannot be followed properly. This spelling mistake, or genetic alteration, is called a mutation. If it occurs in a critical place in a critical gene it may result in disease. This is the case for Wilson's disease. It is caused by mutations in ***ATP7B***. For a person to develop Wilson's disease, however, a mutation is required in both copies of the gene. One mutation will have been inherited from each parent as each parent only passes on one copy of each gene to their children (see ***figure 1***.) Parents who have a mutation in one copy of a gene (whilst the other one is normal) are known as carriers and are perfectly healthy. They will not know they are carriers until someone in the family is diagnosed with Wilson's disease.



The Wilson's disease gene, ***ATP7B,*** is found on chromosome **13**. By *reading* all the letters in this gene, over **300** different mutations have been found in different individuals with Wilson's disease. A new study in the United Kingdom has shown that ***ATP7B*** is the only gene associated with Wilson's disease and that genetic testing is able to identity virtually all mutations (***A genetic study of Wilson's disease in the United Kingdom****: Brain 2013*.) The ***ATP7B*** gene controls how the body handles or metabolises copper. It is involved in copper absorption from the bowel and in the liver, links copper to a protein called caeruloplasmin, as well as removing excess copper into bile. If ***ATP7B*** doesn't work properly, because both copies have a mutation, then copper builds up in the body particularly in the liver, brain and eyes. This causes organ damage and the features of Wilson's disease that I am sure I do not need to describe here. Whilst having mutations in both ***ATP7B*** genes causes Wilson's disease, it doesn't fully explain why the disease is so variable. This may be due to the position of the mutation in the gene, or the presence of other genetic variants elsewhere in our genomes that so far have not been identified.

As we know where in our genomes ***ATP7B*** is, we can now test it directly in an individual with Wilson's disease to see if we can identify the mutations that are causing disease (*genetic testing.) Genetic testing* is routinely available in the UK and can be requested by your specialist. So when should genetic testing for Wilson's disease be carried out?

The main reason for carrying out genetic testing is to confirm a diagnosis in someone suspected of having Wilson's disease. This is particularly important, whether or not *Kayser-Fleischer* rings (deposits of copper around the iris of the eye) are present, when blood tests for reduced copper or caeruloplasmin levels or urine tests for increased copper levels do not provide definitive results and therefore the diagnosis cannot be made with confidence. Other clinical and diagnostic tests, such as liver biopsy may also be carried out, so genetic testing is complementary to these. The use of these tests together will typically allow a diagnosis of Wilson's disease to be made with confidence. Your liver specialist will be able to tell you your results and whether genetic testing is required. Genetic tests can take many weeks to be performed and so blood or biochemical tests will typically be performed first.

So how does someone with Wilson's disease have two faulty copies of the ***ATP7B*** gene? The answer is that a small number of healthy people in the population carry one copy of the ***ATP7B*** containing a mutation. These *carriers* show no signs of Wilson's disease. Until recently it was thought that about **1:90** people were carriers, but the recent UK study mentioned above suggests that it may be as high as **1:42**. When two carriers have children there is a one in four chance that each of their children will inherit both copies of the ***ATP7B*** gene containing a mutation (see ***figure 1***). This is because each parent has a one in two chance of passing on the abnormal gene (**1/2** x **1/2** = **1/4**.) This is called autosomal recessive inheritance. In autosomal recessive diseases we therefore typically see the disease only in siblings, brothers and sisters. With a carrier frequency of **1:42** we would expect to see Wilson's disease in **~1:7000** of the population! Current estimates suggest it occurs in **1:30,000** people. This could mean that not all mutations cause disease or the disease is not always recognised. More research is clearly needed.

Both Parents are *Carriers*

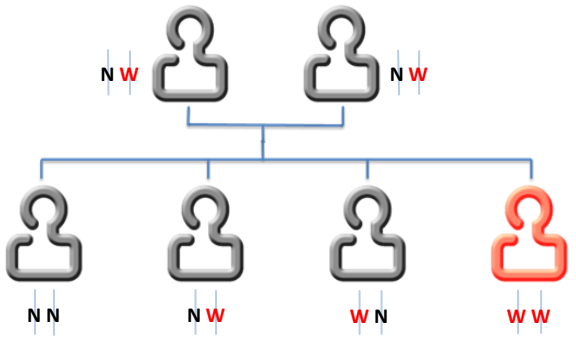


Fig 1.

If both parents are carriers for a Wilson's disease mutation (W) they have a 1:4 or 25% chance of having an affected child with two mutations (WW.) Children with one mutation are carriers and unaffected. The vertical lines represent the ATP7B gene - N is the normal gene.

One Parent with *Wilson's Disease*: the Other *Normal*

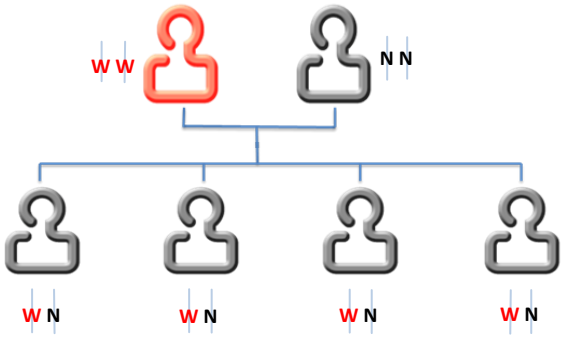


Fig 2.

ALL the children of an individual with Wilson's disease will be carriers. The chances of them also having Wilson's disease depends on the likelihood of the other parent being a carrier.

So once Wilson's disease has been diagnosed in an individual and this may be at ANY age, it is essential that all full siblings are screened. Once the mutations in the ***ATP7B*** gene are known in the family, this information can be used to offer genetic testing to other family members. Again it would typically be used with the blood tests mentioned above. This genetic information can also be used in other ways such as in prenatal diagnosis and these issues should be discussed with a GP, liver specialist or after referral to a genetics' specialist.

For an individual with Wilson's disease, ***figure 2*** shows that all their children will be carriers. The risk of them being affected is determined by the likelihood of the other parent being a carrier. If this risk is **1:90** then the risk of having an affected child is **1:180**. If the risk of being a carrier is **1:42** then the risk is **1:84**. A few families are known where a child of an affected individual is also affected but this is very uncommon. Currently genetic testing of unaffected partners is not routinely offered but this situation may change and if you are concerned you should discuss this with your own doctors.

So understanding the genetics of Wilson's disease has helped us more fully understand the condition and how it affects people. It also tells us what are the risks to other family members and who should be offered testing. Your own doctor should be able to answer most of your questions about Wilson's disease but occasionally referral to a genetics specialist may be required.

Wedding

We are delighted to announce that our committee member **Anne-Marie Styles** (who was diagnosed with Wilson's disease in **1977**) recently married the very lucky **Steve le Cheminant** in the beautiful surroundings of **Austwick Hall** in the Yorkshire Dales. The happy occasion was shared with close family and friends, after which they jetted off to **Venice** for a magical honeymoon.

Outside Austwick Hall

Signing the Register

Honeymooning in Venice

Caption Competition Winner

The winner of the caption competition set by Dr Walshe's daughter, Susan, in last year's newsletter is

Caroline Barr of Edinburgh

Congratulations to Caroline who will shortly be receiving a cheque for £20.00

Panto time

Kay and Dr Walshe at the 2012 meeting

With Joan Smith, daughter & grandson

WDSG-UK 2013-14 EVENTS

Date	Time	Event
Jun 8	**0900 - 1830**	Morbus Wilson e.V. Annual Symposium - University of **Heidelberg,** Germany
July 21	**1100 - 1530**	**WDSG-UK Meeting and 3rd AGM** – Cambridge Rugby Union Football Club Grantchester Road **Cambridge** CB3 9ED.
August 24	**1000 - 1200**	Sylvia Penny warmly invites you to **a Coffee Morning, Bring and Buy and Raffle** in aid of **WDSG-UK** at Victoria Park Church, Claymore, **Torquay.**
Sept 21-22		**Wilson Patient Foreningen - Annual meeting, Denmark**

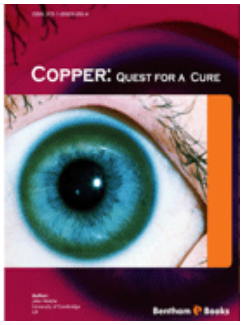
COPPER: QUEST FOR A CURE

Bentham Science Publishers

DOI: 10.2174/97816080506041090101
eISBN: 978-1-60805-060-4, 2009
ISBN: 978-1-60805-657-6

This autobiographical e-book, written by **Dr Walshe** in 2009, describes how Wilson's disease was conquered by a series of individual discoveries, leading to highly effective treatments. It also describes the difficulties which had to be overcome to achieve this in the face of government and institutional bureaucracy. It should be of great interest to patients, those involved in medical research and doctors in general. It is a book for the general reader which avoids technical detail.

Editor:
John Walshe
University of Cambridge
UK



It is available in an electronic format and
can be downloaded from the publishers' website:

http://www.benthamscience.com/ebooks/9781608050604/index.htm. **Price: US$ 35.00**

IN MEMORIAM

Joan Smith, who has been a member of the Group since it was formed in 2000, has very kindly sent us a personal cheque of £50 in memory of both her aunt Joan Drew, who died on Christmas Day aged 95, and her cousin John Warwick, who died on New Year's Eve, 2012. We thank Joan for marking their passing in this way.

* * *

We send our condolences to Emma Collcott and her family for the sad loss of her grandfather, Charles Galloway, who passed away on 13 February 2013. We were pleased to have met him and had his support at several of our Cambridge meetings. We are also most grateful to his widow, Emma's grandmother Elizabeth Galloway, for choosing to hold a collection for WDSG-UK at his funeral, which raised the very generous sum of £335.

Wilsons Disease Support Group UK

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Dr Godfrey Gillett	Group Adviser, Honorary Member
Dr James Dooley	Group Adviser, Honorary Member
Dr Caroline Simms	Group Founder

W D S G - U K

Tell others about WDSG-UK

Please tell others you know with **WD**, who might benefit from the Support Group and what we are doing.

Inform your family, friends, consultant physicians, GP surgery, local MPs about **WDSG-UK.**

The more people who know about us, the more we can promote a better awareness of **Wilson's disease** within the community and the better the chance of early diagnosis.

If more copies of this newsletter or patients & families' correspondence lists are required, please contact:

Linda Hart

We're on the web
www.wilsonsdisease.org.uk