Wilson's Disease Support Group – UK

NEWSLETTER

APRIL 2015

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 :

BRITISH LIVER TRUST

RARE DISEASE UK

Genetic Alliance UK
Supporting. Campaigning. Uniting.

EuroWilson

Children's Liver Disease Foundation
fighting childhood liver disease

Climb
Children Living with Inherited Metabolic Diseases

Hello and welcome to this year's newsletter which has turned out to be bigger than ever. Thank you to all our contributors, without whom there would be no news at all! As it is there are articles bringing you up to date with what both the committee and our members have been doing over the past year, as well as the popular and inspirational stories from patients and their families about the effect Wilson's disease has had on their lives. We have included the Food Table again which gives an approximate guide to the copper content of certain foods and which is useful to keep in your shopping basket for when the chocolate aisle beckons at your local supermarket! If anybody is interested in a more comprehensive list, I can recommend McCance & Widdowson's *The Composition of Foods* 6th ed. in which a total of 1,235 foods have been analysed and recorded.

On the medical front there is an *insight* into new medical techniques being developed for monitoring the progress of Wilson's disease through the eyes, an explanation of the meaning of genetic mutations in WD and also the third and final simple guide in the series discussing the function or, in this case, the malfunction of the liver. We hope that there will be plenty to interest everyone.

However, our big news story this year is that the Group is about to launch the first Wilson's Disease Patient Register ever in the UK. Currently, there is no cohesive list documenting how many patients are living with Wilson's disease in the UK nor where they are being treated. It is hoped that by putting together a register, it will encourage and facilitate research, which in turn could lead to better treatments and outcome for all patients living with the disease. Please support us in this endeavour: full details are enclosed.

Our Facebook Group continues to flourish and we now have over 400 members made up of patients and their families from all over the world. In fact, the only continent that still remains unrepresented is Antarctica! I cannot recommend it enough as a way of sharing our experiences and supporting others, particularly those who have just been diagnosed or those who have have never had contact with others with the disease.

This year we are hoping to have a more informal annual meeting, in celebration of the fifteen years that the Group has been running since Caroline and Linda set it up at the beginning of the new Millennium. Do please come if you can. I know that it is a long way for many of you to travel, but we do appreciate your support and without you, there would be no Group, no meetings and nothing to report in our newsletters!

I wish you all a very happy Easter and good health in the year to come.

Valerie

IN THIS ISSUE ...

Chairman's Report for 2014-15

The past year has seen a continuation of our links with like-minded organisations and the ways we communicate with members. But also some new initiatives are coming to fruition. For example, we are seeking ways to sponsor research into Wilson's disease and we are taking advice about establishing a UK register of patients with Wilson's disease. We have also commissioned a new logo for *WDSG-UK* in response to requests from *Rare Disease UK (RDUK)* and others.

WDSG-UK Meetings

The *WDSG-UK* management committee met in Knowle in June 2014 and in Cambridge in November 2014. We held the fourth *WDSG-UK* AGM as part of our annual meeting for members, family and friends in Cambridge on 13 July 2014, and a report of our annual Cambridge meeting appears on *p 4* of this Newsletter.

Donations and Fundraising

I should once again like to thank members, their families, friends and sponsors for the magnificent additional income of **£2,100** that they have raised for the Group through donations and fundraising in **2014-15**. I would particularly like to thank **Belinda Diggles**, **Sylvia Penny**, **Allie Johnston** and **Liz and John Wood** for their fundraising (see *pp 5-7.)* We were very sorry to hear of the deaths last year of Wilson's patients, **Maureen Adams, Ron Shaw** and **Valerie Kingdom**, and thank their families for kindly holding collections for *WDSG-UK* at their funerals, attracting donations totalling **£1,150.** Finally we are grateful to ***Univar*** for their sponsorship in **2014-15** of **£1,000.**

A New Logo for WDSG-UK

This is our new logo. The 'W' in the new design can also be viewed as the symbol for copper, *Cu*, the metal responsible for the symptoms of Wilson's disease. The blue colour is the colour of cupric ions (Cu^{2+}) complexed in aqueous solution with e.g. an amine. Trientine is an amine, and complexing copper with trientine is one way of removing copper from the body by excretion of the copper-trientine complex in urine, and thus treating the disease.

Cu ilson's
Wilson's Disease Support Group – UK
www.wilsonsdisease.org.uk

Facebook Site

In the last year, membership of the *WDSG-UK Facebook* site (a Closed Group) has increased by more than one hundred. Replying to, and keeping track of our **400** members is no easy task, and we owe a big debt to **Valerie** and **Linda**, who deal with the many requests for advice and information received from our *Facebook* members. I should just like to repeat a request that I have made several times before – please could our *Facebook* members who do not belong to *WDSG-UK* consider joining? For a modest subscription, your enrolment would make a big difference to the future of *WDSG-UK*. A membership application form may be found on the *WDSG-UK* website.

Sponsorship of Research into Wilson's Disease

We are pleased to report that Susan Mollan, a Consultant Neuro-Ophthalmologist at University Hospitals Birmingham, has accepted a donation from *WDSG-UK* towards her research on the examination and analysis of *Kayser-Fleischer* rings in Wilson's disease. You will remember that Susan gave a presentation on the techniques she uses in her work at last year's *WDSG-UK* annual meeting in Cambridge (see her synopsis here on *pp 18-19*.)

Wilson's Disease Network – UK

Dr James Dooley, Dr Oliver Bandmann, and colleagues who diagnose and treat patients with Wilson's disease have formed an informal working group – *Wilson's Disease Network – UK* – with the aim of establishing a UK-wide network of specialists who have experience in the diagnosis and treatment of Wilson's disease. *WDSG-UK* committee members Jerry, Valerie and Mary were invited to attend the third meeting of the *Wilson's Disease Network – UK*, which took place at the Queen Elizabeth Hospital Birmingham in June 2014.

Links with British Liver Trust (BLT), Liver Patients' Transplant Consortium (LPTC) & *NHSBT*

Valerie continues to represent the Group at meetings organised by the *BLT* and *LPTC*. In February she attended an *NHS Blood and Transplant (NHSBT)* meeting in London where the proposed new allocation of livers in transplantation was discussed. This was then followed by a BLT & LPTC meeting.

BRITISH LIVER TRUST
LPTC The Liver Patients' Transplant Consortium

Rare Disease UK (*RDUK*)

Over the past year, *WDSG-UK* has continued to support *RDUK* and, in particular, its work in promoting the implementation of the *UK Strategy for Rare Diseases* in the health services of the four home nations. I have attended seven meetings in London organised by *RDUK* over the last year. Brief details of these events are given below.

Europlan II National Conference (24th June 2014): Europlan is an initiative of EU linked organisations with an interest in rare diseases. The second Europlan conference, hosted by Rare Disease UK, included presentations by representatives from England, Wales, Scotland and Northern Ireland on the implementation of the *UK Strategy for Rare Diseases* in their respective countries, and talks by specialists who treat patients with rare diseases. Full details of this conference may be found at http://www.raredisease.org.uk/europlan2.htm

Rare Disease UK Patient Empowerment Group Meetings (6 June and 25 September 2014, 19 January 2015): The *Patient Empowerment Group* was established as part of the work carried out by *RDUK* (and *Genetic Alliance UK*) in monitoring progress of the *UK Strategy for Rare Diseases*. Representatives from about one dozen patients' groups (including *WDSG-UK*) were invited to be members of the Patient Empowerment Group. The three meetings held so far have provided an opportunity to hear directly from officials at the Department of Health about implementing the five main strands of the *Strategy*: (i) empowering those affected by rare diseases; (ii) identifying and preventing rare diseases; (iii) diagnosis and early intervention; (iv) coordination of care; (v) the role of research.

First All Party Parliamentary Group (APPG) Summit on Rare Diseases (19 November 2014): This event, hosted by *RDUK* and some other medical charities, took place at the Palace of Westminster and coincided with the first anniversary of the publication of the *UK Strategy for Rare Diseases*. The meeting, attended by members of the House of Commons and the House of Lords, provided an opportunity for *RDUK* to launch its report on 'Patient experiences of transition between care providers' – the issues faced by patients with rare conditions when they progress from child to adult care and then later to older age care. A pdf of this report is available from the *RDUK* website.

Rare Disease UK AGM (19 January 2015): There were three guest speakers at this year's *RDUK* AGM: (i) Richard Jeavons, Director of Commissioning Specialised Services, NHS England; (ii) Sarah Stevens, National Congenital Anomaly and Rare Disease Registration Service, Public Health England; (iii) Professor Kate Busby, Professor of Neuromuscular Genetics, Newcastle University, who spoke on her experiences of setting up data registries for neuromuscular conditions. Further details of the 2015 AGM may be found from the *RDUK* website at http://www.raredisease.org.uk/governance.htm

Rare Disease Day (25 February 2015): This year's Rare Disease Day in England was recognised with a parliamentary reception at the Palace of Westminster. Earl Howe (Frederick Curzon), a Conservative front bench member of the House of Lords and a Parliamentary Under-Secretary of State at the Department of Health, spoke on the progress in meeting the objectives of the *UK Strategy for Rare Diseases*. Earl Howe emphasised the value of genomics for tackling the diagnosis and ultimately the treatment of rare diseases, and announced the establishment of eleven new *NHS England* Genome Centres. He also referred to a single national registration service for rare diseases, which *Public Health England* is in the process of establishing. For the Labour Party, Liz Kendall MP, Shadow Minister for Care and Older People, confirmed the Labour Party's commitment to the *UK Strategy for Rare Diseases*.

WDSG-UK Annual Meeting and 5th AGM

The 2015 Support Group Meeting has been arranged for **Sunday, 12 July 2015** at the clubhouse of the city of Cambridge's Rugby Union Football Club. During the course of this meeting the **5th *WDSG-UK* AGM** will be convened. An agenda for the AGM is enclosed with this Newsletter. As part of the AGM, the election of officers and members of the *WDSG-UK* Management Committee for the year **2015-2016** will take place. All members of the current committee have submitted their names for re-election to the committee for this period.

I am grateful to members of the *WDSG-UK* management committee for their hard work and enthusiasm during the past year: **Linda Hart, Valerie Wheater, Jerry Tucker, Anne-Marie Le Cheminant** and **Mary Fortune**. We look forward to increasing the influence of *WDSG-UK* over the next year by helping our members in the UK and our very many virtual members worldwide.

Rupert Purchase
March 2015

Wilson's Disease Support Group Meeting & 4th AGM

Cambridge Rugby Union Football Club, 13 July 2014

The 2014 annual meeting of *WDSG-UK* attracted an encouragingly good turnout on a warm, slightly humid day in Cambridge. **Mary**, **Linda** and **Valerie** greeted members and friends as they made their way to the spacious surroundings of the Cambridge Rugby Union Football Club. We were especially pleased to welcome back **Charlie Watsham** and Biggles, **Emma Colcott** and her mum **Lesley**, and **James Kinnier Wilson**. Also, we were delighted to meet **David Lin** from Taiwan, and **Bongkeun Jeon**, his wife **Sunhae** and their daughter, **Olivia**, whose story about her chance diagnosis of Wilson's disease whilst visiting South Korea appeared in last year's newsletter.

Alastair Kent, Chair of RDUK

Clinicians and scientists with a special interest in Wilson's disease who attended again this year were **Dr John Walshe**, **Kay Gibbs**, **Dr Godfrey Gillett** (Northern General Hospital, Sheffield) and **Dr Bill Griffiths** (Addenbrooke's Hospital, Cambridge.) Also, on his first visit we were pleased to welcome **Prof Aftab Ala**, (Consultant Gastroenterologist and Hepatologist, Frimley Park Hospital, Surrey.)

Before lunch, **Rupert** talked about our links with two other groups: the ***Wilson's Disease Network – UK*** and ***Rare Disease UK.*** *Wilson's Disease Network UK* is an initiative of **Dr James Dooley** and **Prof Oliver Bandmann** and is an informal collaboration of clinicians and scientists with a special interest in Wilson's disease. It has the aim of improving the treatment of Wilson's patients across the UK by the creation of Centres of Excellence. One of the other goals of *The Network* is to establish a UK Register of patients with Wilson's disease, which **Jerry** is undertaking. He briefly described how this work is progressing and urged *WDSG-UK* members to participate in the project. ***Rare Disease UK***, a national alliance for people with rare diseases, was involved with the input, compilation and launch in 2013 by the UK government of the ***UK Strategy for Rare Diseases***. **Alastair Kent OBE** is the Chair of *RDUK* and has a major role in liaising with UK health professionals and UK government in the implementation of the Strategy. We were therefore very pleased that Alastair was able to give us a first-hand account of this work at our meeting.

Fire hazard cake!

After lunch, **Linda** was presented with a cake in recognition of a *special* birthday and for the work she has carried out for *WDSG-UK* since she and **Caroline** founded the Group in 2000. The afternoon's proceedings began with the formalities of the *WDSG-UK* 4th AGM, during which the current management committee was unanimously re-elected for the year **2014-2015.** **Belinda Diggles** then presided over the magnificent raffle she had organised which raised the grand sum of **£150**. The Group's funds were then further enhanced by a donation of **£1,000** from ***Univar Ltd,*** which **Anne-Marie** accepted on our behalf from **Philippa Hoare** and **Sabrina Chowdhury.**

Anne-Marie receiving a cheque for £1,000 from Philippa Hoare and Sabrina Chowdhury of Univar

Susan Mollan, a Consultant Neuro-ophthalmologist at University Hospitals Birmingham, gave the third presentation of the day. Susan and her team are investigating a number of advanced imaging techniques as diagnostic tools in ophthalmology, and in particular the application of optical coherence tomography (OCT.) Susan has been using this technology for the examination of *Kayser-Fleischer* rings (an ophthalmic manifestation of Wilson's disease) and she gives us a review of this work in her article on ***pp 18 & 19*** of this newsletter.

Questions from the audience had been addressed to the two speakers during the day, and these continued with further questions to all the medical specialists present before the meeting concluded with the customary Group photographs taken by **Barry Diggles**.

Patients' Group Photograph

Fundraising 2014-15

by Belinda Diggles

For a number of years I have made Christmas puddings and in doing so raised funds for *WDSG-UK*. At the same time I have baked, made jam, marmalade and chutneys for other charities such as *Children in Need, Comic Relief* and *Tickled Pink,* selling all my home made produce to family and friends.

Last July I decided to concentrate all my fundraising efforts on *WDSG-UK*. I set myself a target of raising **£400** by our annual meeting in July 2015. With this in mind, I knew I needed to get started soon, so one of my first jobs was to enrol my husband Barry into helping me to pick the fruit. On a wet Sunday morning in August we drove up to Milnthorpe in The Lake District to a *Pick Your Own* fruit farm, where we gathered 5kg strawberries, 8kg blackcurrants, 1.24kg redcurrants and 627g raspberries. Luckily the weather improved and having spent time with our son Peter and his fiancée who live in that area, we returned home stopping at *Asda* on the way to buy sugar and the pectin needed to help the jam set.

Barry - Picking Blackcurrants

1:

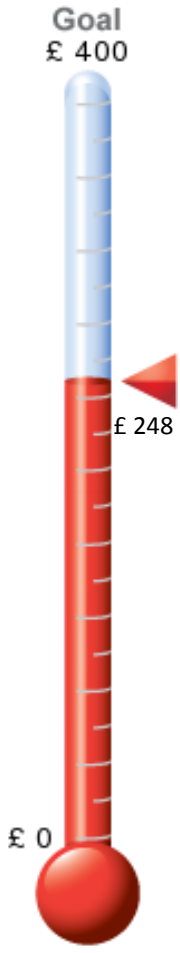
The fruits of our Labour

2:

Hubble Bubble

3:

8 lbs of Strawberry Jam



My next fundraising idea was to make chutney as I had been given lots of pears and apples by friends who had had bumper harvests. In fact I had so many pears I started making jam again and anything else I could think of that has them as an ingredient. However, by now I was running out of jam jars so the call went out to my friends to help, which luckily they did. And once word got around that I had been making jams and chutneys, orders started pouring in, giving me a good start towards my £400 target.

Fruit Chutney

October soon arrived and so I drew up a list of ingredients I needed to make my Christmas puddings. I then set off to buy 6½ kg dried fruit, chopped nuts, mixed peel, plain flour and mixed spice, 2 kg brown sugar, 3½ kg breadcrumbs and shredded suet, 32 eggs, 8 apples, 8 lemons and 48 tablespoons of brandy! Four days later I had baked the first batch of 7 large puddings which left me, I am pleased to say, with only a further 2 large, 13 small and 4 extra small (in ramekin dishes) puddings to bake.

Christmas Puddings

I could now rest until January when the Seville oranges would be in the shops for me to make marmalade. However, this nearly didn't happen due to an accident I had at work, when I fell and fractured my right elbow which then required surgery. Unfortunately, I have only been able to make a few jars this year, but at least I have made some.

But I am determined to try and reach my target of **£400**. As I sit and write this at the end of February my total stands at **£248.02** leaving me with **£150** still to raise. I will be attending the meeting in Cambridge in July and will bring any surplus jars of jam, marmalade and chutney down with me to sell at a price of **£1.50** each, together with some small Christmas puddings at **£6.00** each.

Fundraising 2014 - 15

Sylvia's Summer Bring & Buy Sale

by Valerie

Emma Coombes was diagnosed with Wilson's disease at the beginning of 2007 (see her story in last year's newsletter) and her grandmother **Sylvia Penny** has been regularly organising fundraising events for *WDSG-UK* ever since. She holds a bring and buy sale, coffee morning and raffle every August in the Victoria Park Methodist Chapel near to where she lives in Torquay. Having surprised them all by turning up to the event unannounced in 2013, I surprised myself this year by turning up again! It was a spur of the moment decision made at 4.00 o'clock in the morning and meant a ten hour round trip to get there and back in the day. I arrived just as the doors were opening and was able to enjoy a fun morning with the family and all their supporters catching up on everybody's news.

L-R : Sylvia, Tom, Emma, Geraldine, Daisy and Sheila with Geraldine's 6 year old grandson Ashton

This is the eighth such event that Sylvia has organised and this year she raised another staggering **£472** for the Group, bringing her total over the years to **£3,500**. Our one disappointment is that she and Em have never been to any of our meetings, but we hope that this year they might finally manage it. A very big thank-you to them all for continuing to support us in this way.

* * * * *

Allie's Handicraft Sales & Christmas Raffle

Allie has spent the whole year tirelessly raising funds for the Group, finding assorted outlets in and around Edinburgh in which to sell her hand made greetings cards. Customers included her physical trainer **Donnie**, her nephew **Michael**, staff at **St Jude's Laundry**, staff and pupils from the Community Learning Centre that she attends, **Sir Tom Farmer** a former business associate and his work colleagues, and most recently from fellow patients at her dental practice and medical centre. She also collected a generous donation of **£100** from her former employer, **Capital Solutions**.

In addition she has been attending cookery classes at evening school and was encouraged in November to start making a Christmas cake. The finished result was amazing and as certain of the ingredients in a Christmas cake contain copper, Allie decided to raffle hers. Her class tutor, **Belle Bale**, came out in sympathy and raffled hers too! Between them they raised **£140.00** for the Group. This brings the total collected by Allie this year to **£398.00** for which we should like to thank her and all her supporters.

Mixing the cake at home

Allie and her mum Rita are hoping to get down to Cambridge for our meeting in the summer and plan to set up a stall selling her latest stock of cards. For those of you who are also intending to join us, be sure to bring your cheque books!

Before…

and After

John's London Triathlon - August 2014

by John Wood

I was in the bar after playing tennis when I had my arm twisted to join in the London Triathlon – not something I had even thought of doing before, although I ran reasonably often. I thought I would do it for charity to motivate me through the training and, having previously run a half marathon for a local hospice, my wife Liz had the brilliant idea of doing it for ***WDSG.***

The event was based at the ExCeL Exhibition Centre in London. I chose to take part in the *Sprint Triathlon,* which is exactly half the distance they do at the Olympics and involves swimming **750m** round the adjacent Royal Victoria Dock, cycling **20km** around the Docklands area and then running **5km.**

I had always found swimming front crawl extremely difficult, so I decided to have some lessons. My teacher was a little bemused, as despite being very fit and able to swim 30 x 25metre lengths breast stroke without any difficulty, I was exhausted after just one length of front crawl! I ended up having to stick to breast stroke, which I swam faster than many of those using front crawl! Practising the swim in a wetsuit in the sea at Southend took a bit of adjustment, because to start with I was so buoyant I couldn't keep my legs in the water. I was left floating on the surface and kicking in the air!

In some ways I was extraordinarily happy with my time of 2 hrs 1 min 15 secs, but there is just that little niggle that I spent over 10 minutes "in transition" (changing from one event to the next) whereas lots of people did it less than 5 minutes. If I had spent a little less time drying between my toes or eating that banana, I might have broken the 2 hour mark! Maybe next time…

I had a great time and also raised **£475.00** (plus Gift Aid) for *WDSG-UK,* who thanked all my sponsors and sent them more information about WD. This must have worked because only yesterday I met a colleague who had sponsored me and he mentioned Wilson's disease unprompted, so that is at least one person whose awareness has increased!

John (2nd left) - No 4281

Swimming in the Royal Victoria Dock

20 km Cycle Ride

We would like to thank everybody who continues to make donations to the Group with their subscriptions and also our fundraisers who have raised over **£8,000** for ***WDSG-UK*** over the past ten years.

In particular we are grateful to **Sylvia Penny** and family from Torquay, **Barry** and **Belinda Diggles** from St Helen's**, Liz** and **John Wood** from Southend**, Rita** and **Allie Johnston** and **Stephanie** and **David Reid** from Edinburgh**, Lesley Galloway** and family from Leighton Buzzard and **Philip** and **Tulin Hawkins** from Oxford. Together with committee members, they have run coffee mornings and bring and buy sales, undertaken sponsored walks, cycle rides, swimming events and sky dives, run quiz nights, marathons and triathlons, put on concerts and golfing events, baked puddings and cakes and made jams and chutneys, sold handicrafts and held raffles, and most unusual of all have taken part in Loony Dooks (a Scottish tradition involving getting very cold and wet on New Year's Day!)

If **you** have a special talent or interest and feel that you would like to use it to raise funds for the Group, then we would be delighted to hear from you. Sponsorship and Gift Aid forms are available on request. We have a Diary of Events section at the back of the newsletter and currently only Sylvia from Torquay ever advertises in it. We would love to hear from anybody else who has ideas for fundraising.

Cirrhosis - A Simplified Outline

by A. Stevens

What is Cirrhosis?

Cirrhosis is a chronic disease of the liver which has many causes, and may eventually lead to liver failure and oesophageal varices. A chronic disease is one which begins insidiously and usually progresses very slowly, usually over a period of many years. By contrast, an acute disease starts suddenly and progresses quickly, usually over a few days or even hours. Appendicitis and pneumonia are examples of acute diseases. The liver may also have acute diseases: for example some viruses can cause acute viral hepatitis.

What are the Causes of Cirrhosis?

Many diseases can lead to cirrhosis. The most common is alcoholic liver disease, but some hepatitis viruses lead to cirrhosis, as do some autoimmune diseases. The common feature is that the liver cells are damaged and destroyed a few at a time, but repeatedly, and over a long time period, rather than in acute liver disease where all the millions of liver cells are damaged or destroyed at the same time. Wilson's disease and the iron storage disorder (haemochromatosis) are rare causes of cirrhosis.

What happens in the Liver in Cirrhosis?

There are four main changes in cirrhosis, which occur in the following sequence:-

- groups of liver cells (hepatocytes) are killed by the causative disease
- scars form in the space formerly occupied by the dead liver cells
- the scars can disrupt the architecture of the liver, interfering with the input and output systems
- the surviving liver cells in the region multiply to try to compensate for the liver cells which have been killed. These form so-called "regeneration nodules."

Why do These Changes Interfere with the Function of the Liver

In the 2014 edition of the Newsletter, I illustrated (on ***pp 8 & 9***) how the functions of the liver as a sort of chemical factory depended on the perfect interrelationship between:-

- the **input system,** bringing raw materials (via the hepatic artery and hepatic portal vein) to the liver to be worked on by
- the **work units (the liver cells, called hepatocytes)** which carry out the chemical processes and then pass the products into
- the **output system** which carries the products (via the central and hepatic veins) into the blood circulation to be transported to the parts of the body where they are needed.

Diagram 1, which also appeared in my previous article in the **2014 Newsletter**, shows in greatly simplified form how this system works normally.

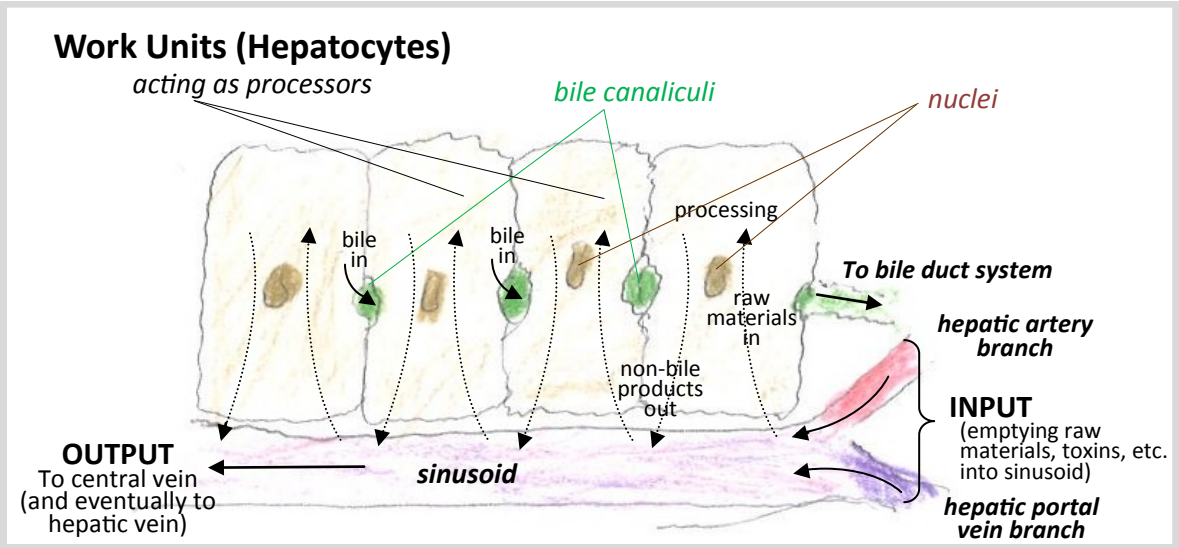
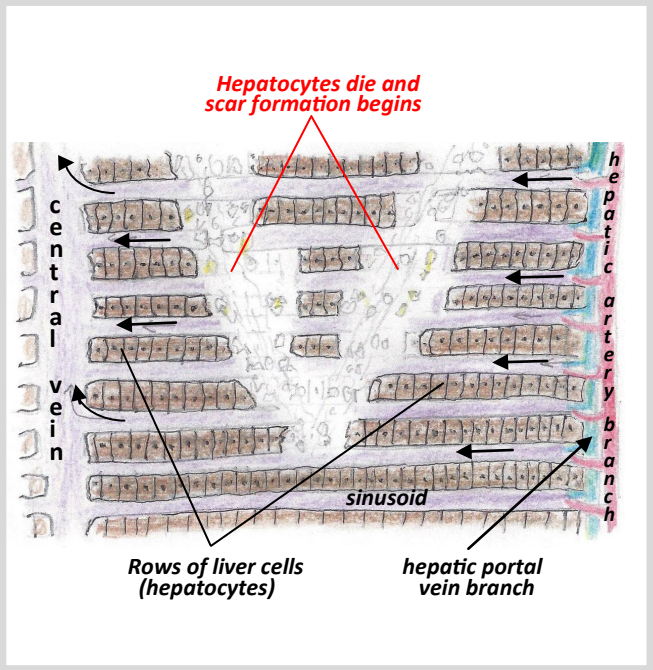
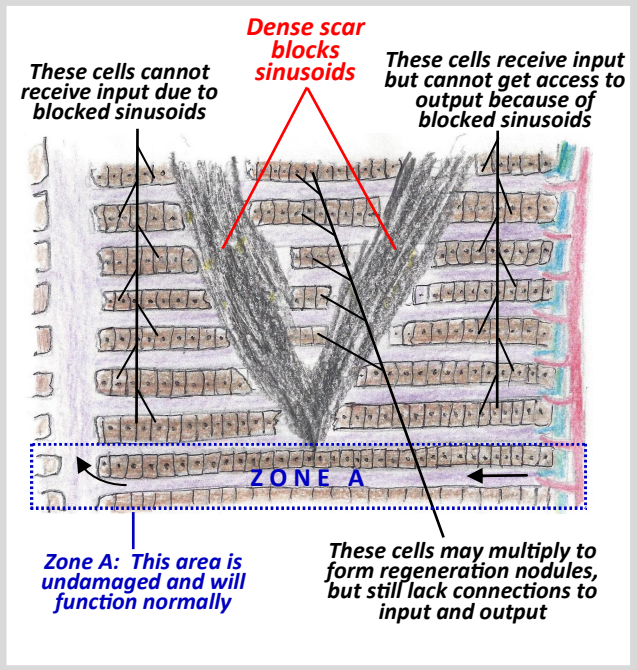


Diagram 1

Diagrams 2a and 2b show (also in simplified form) what happens in cirrhosis. In **Diagram 2a** two groups of liver cells have been destroyed by disease, and thin scars are beginning to form at the sites of the dead liver cells.



2a



2b

In **Diagram 2b** the scar is fully established and is interfering with the input and output systems. In this tiny area of the liver therefore, there are some hepatocytes which are receiving raw materials and processing them but cannot pass the products into the output system, and there are other hepatocytes which have access to the output system but are cut off from the input system and therefore cannot receive the raw materials to work on. The effects on liver function on a single episode at one site in the liver like this is, of course, minimal, but over the years it occurs in many thousands of places in the liver, and eventually the normal liver functions outlined in my last article will become impaired.

The impairment of liver function is the result of a combination of great reduction in functioning hepatocytes, and the distortion of the input and output systems by the extensive scarring. Reduction of the numbers of hepatocytes means that there are fewer to carry out the vital chemical functions of synthesis and detoxification (see my **2014** article), and the obstructions to input and output flow has a number of consequences (see below.)

What problems arise because scarring affects the input and output systems?

There are three main problems:-

- The flow of raw materials in and synthesised products out is impaired, reducing the efficiency of the hepatocytes;
- The physical obstruction to the outflow of bile from the liver to the bile duct system means that the bile gets stuck in the liver, and some can pass into the blood stream to cause **jaundice;**
- The obstruction to flow of the branches of the hepatic portal vein within the liver leads to a great increase in the blood pressure in the hepatic portal vein, and this leads to the formation of **oesophageal varices**, (the subject of my first article in the **2013 Newsletter**.)

Why don't the new hepatocytes in the regeneration nodules take over the functions of the dead hepatocytes?

They do to an extent. However, the problem is that the cells in the regeneration nodules may not have proper connections to the input and output systems, and therefore not all of them can work efficiently, if at all.

The loss of output from the remaining hepatocytes leads to severe metabolic upset in the rest of the body, the features of biochemical liver failure (perhaps the subject of another article in the future.)

Rita's Story

by Rita Johnston

My name is Rita Johnston and I wanted to write an article about the effect that Wilson's disease has had on my daughter, Allie. For those of you who have not read Allie's story in the **2013** newsletter, let me give you an overview of the challenges she has faced. Wilson's reared its ugly head in **2009** and on her 40th birthday in **2010** Allie was admitted to hospital, very ill, to have a peg fitted so that she could be fed through her stomach. **2010** was not a good year for her: she deteriorated so fast that she ended up in a wheelchair. She was mute, incontinent and had a 19cm deep pressure ulcer on her back. To round things off the side effects of the drug that she was on made her throat close and her tongue swell such that she had to have her lower teeth removed to allow her tongue to deflate.

Enjoying an ice cream with Allie in 2008 before she became ill

I made up my mind that things couldn't get any worse and I had to try to move forward and do what I could for her. I hired a physical trainer, an acupuncturist, a psychotherapist, a hypnotherapist and a speech therapist using the funds she receives from her *Employment & Support Allowance* (*ESA*). I went a little over the top with therapies, but my daughter needed help and it fell upon me to arrange it for her. As a result we were able first to get rid of the wheelchair, then the lounger she had been lying on in the sitting room (which she wasn't at all happy about), but the idea was that she would have no alternative but to get up and keep moving without these things around her. She was not happy at my dispensing with her comfortable lounger, but my mind was made up, it had to go!!

Allie and I December 2014

We still have our physical trainer because Allie needs to keep agile and she is not good at doing exercises by herself. At one time we had some gym equipment in the house, a treadmill, etc., but they all had to go too. Instead she now has to make the effort to walk to the gym, where her trainer puts her through all kinds of exercises. And recently she has started taking swimming lessons, swimming being something she loved to do before Wilson's disease affected her. After swimming she rewards herself by using the steam room to relax. I once went with her, but two minutes in the steam room was quite enough for me!

Unfortunately, Allie has been left with involuntary movements, incoherent speech, and an inability to chew her food properly. When she is out in public people stare and are rude to her and when shopping and visiting the library she often has difficulty making herself understood. It is very upsetting for her, but one of the saddest consequences of the illness is that her former friends have found her abnormalities difficult to deal with and no longer come to visit. However, Allie has decided not to allow the loss of these *so called* friends to further ruin her life and has taken up arts and crafts which she looks forward to every week. She has made (with a little help) many greetings cards and sells them to raise funds for the Group. She plans to bring some with her to the ***WDSG-UK*** meeting in the summer to display and sell there.

One day in **2012** a package came through the post, which was a bit of a surprise as I hadn't ordered anything. It turned out to be from *Motability*, offering Allie a car to be paid for from her *Disability Living Allowance (DLA*.) How wonderful! Allie hadn't driven for 4 years so we immediately enlisted driving instructors and although there were lots of tears and doubts like, "I can't do it, I'll never be able to drive again", and "I hate Wilson's", we persevered as I wasn't prepared to let her give up. It cost a lot of money but in September **2013** she went through a driving appraisal with the *DVLA* and she passed!! There is a shadow though, because Wilson's is so rare *DVLA* wants her to sit another appraisal in **2016**! Already Allie is worried as it would be absolutely devastating if she were to fail, but our plan is that nearer the time we'll employ instructors again to set her mock tests, perhaps once a week, so that when the actual test day comes she will be more confident. I have total belief she will pass.

Life can be very cruel but with help and determination we have to remain positive. We cannot cure Wilson's, but we will push it aside as far as possible.

Never Take the Little Things for Granted by Sana Latif

Nothing really prepares you for what's to come next when you are diagnosed with such a disease; or any disease for that matter. It was seven years ago, in February **2008**, when I was first diagnosed with Wilson Disease. I was **23** and in my final year at university. I was popular, with many friends and a good social life. But lying in that hospital bed, I had lost it all. My symptoms had become really bad; severe tremor, my speech had completely gone, I couldn't swallow even a sip of water and I was unable to stand up.

This had all started a year and a half earlier. My speech was the first thing that began to decline. It became extremely slurred. I would get terrible cramps in my legs and feet during the day and night. Slowly, I found it hard to swallow my food. I'd go to see my GP at least once a week as I knew that there was something terribly wrong happening to me. But no-one believed me; my GP would sometimes say that it was a nutrition malfunction or that I was iron deficient or even that it was psychological. I felt terrible and completely helpless. It came to a point where I started to fall frequently and I was walking as if I were drunk.

It was only when I was assessed by a psychotherapist that he concluded that there was nothing psychologically wrong with me. He could clearly see that it was a physical crisis. So with my permission, he called an ambulance and took me to hospital. I was seriously relieved at this point that someone had finally had the sense to do something logical. The next morning I was seen by a neurologist, after which it was a pretty quick diagnosis. He asked about my symptoms, took one look at my eyes and saw very visible Kayser Fleischer rings, then told me that I probably had Wilson's disease. Those were the four words that changed my life.

I felt so alone, but couldn't express it. I texted a few of my friends and told them what had happened. A couple of them even came to visit me in hospital during the three months that I was there. Mum used to pop by every day after work, which was just about the only thing I had to look forward to.

When they let me go back home, the realisation hit me. I couldn't finish my studies which meant I couldn't graduate and all my friends had suddenly disappeared. All of them. They wouldn't talk to me even when I tried. All of a sudden I felt quite lonely, something which I wasn't prepared for. My family only saw the physical struggle I was going through, but nobody really could see that I had become deeply depressed. The once happy life I had enjoyed had slipped away from my grasp in moments.

This is something which is difficult to deal with, and even more difficult to explain. Every time I went to see my doctors, I only ever had one question to ask them, "When would I get better?" – to which I never got an answer. It was an extremely gradual process for me to be able to stand up again. Even today, I struggle tremendously to walk. My tremors are still there. And worst of all, I feel as lonely as ever. What makes it all worse is the cynical remarks people make, which really undermines a person's self esteem. This is the sole reason why I avoid large gatherings. I hardly ever go out as half of the time I'm unable to. But what I really need the most is friends. Somebody I can talk to. What I've found over the past six years is that nobody really wants to know somebody who's unwell all the time. Harsh as that may sound, it's quite true.

But who doesn't fall ill? Good health is something a lot of people take for granted. Having good health, friendship, companionship, being able to walk and having the ability to talk well may all seem like little things to some, but to others it means the ability to appreciate life. Because, even the best fall sometimes. And its only when you fall, you realise how difficult it is to get back up.

One good piece of news, however, is that I have been secretly studying and although I had to start all over again, I have recently gained a degree in Law (LLB) and surprised everyone!

IMPORTANT NEWS

The Launch of The Wilson's Disease Patient Register - UK

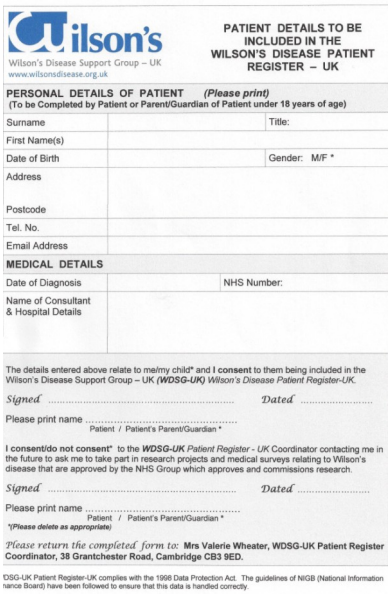
One of the aims of **WDSG-UK** ***(WDSG)*** is to support any initiative particularly clinical research which aims to improve the lives of patients living with Wilson's disease. To this end and with the backing of the medical community, we are about to launch the ***Wilson's Disease Patient Register - UK*** *(The Register)* which will be funded and managed by the Group. Research into rare diseases is one of the core principles of the UK's *Rare Disease Strategy* and *The Register* will complement and support the implementation of this *Strategy* by 2020. It will also undoubtedly raise the profile of *WDSG* within the *NHS*.

Over the last few months, I have sought extensive advice about running a Patient Register, and ours will be managed from a central location within *WDSG*. Confidentiality of patient information is our prime concern and *WDSG* will strictly comply with guidelines issued under the *1988 Data Protection Act*. Anybody living in the UK who was born with Wilson's disease is eligible to have their details entered on *The Register,* including those people who have subsequently received liver transplants. *The Register* is open to both adults and children, with children (up to age 18) being registered by their parents or guardians on their behalf. Only the following information about patients will be kept on *The Register*:

- Name, address and contact details
- Gender
- Date of birth
- Date of diagnosis
- Name of consultant and hospital details

Agreeing to be on *The Register* is one way in which we can potentially assist in clinical research which we hope might lead to improvements in diagnosis and a more consistent approach to treatment of Wilson's disease within the UK. Researchers will be invited to approach the Register Coordinator outlining the details of any proposed research and requesting to be put in touch with any suitable patients. The Coordinator will in turn write to such patients giving details of the research and asking if the patients would like to take part. If the patient gives his/her written consent, then the details held on *The Register* will be forwarded to the researchers who will then separately contact the patient. For each new research project, separate written consent will need to be obtained from the patient. A patient can withdraw their details from *The Register* at any time.

Recruiting new patients to *The Register* is an area of high priority for *WDSG.* According to *Public Health England*, 75% of rare diseases, such as Wilson's disease, affects children. The majority of patients with rare diseases are diagnosed when they are children or in adolescence, and Wilson's disease follows this trend. Consequently, recruiting in this group is an area we are focussing our attention on. We are currently producing a new *WDSG* contact leaflet for medical staff to issue at clinics, and it is hoped that newly diagnosed and existing patients who are not members of the Group will be encouraged to join *The Register*. Finally, we are in the process of updating our website with information about *The Register* and when we have finished, we will officially announce it on our *Facebook* page.



Wilson's Disease Support Group – UK
www.wilsonsdisease.org.uk

PATIENT DETAILS TO BE INCLUDED IN THE WILSON'S DISEASE PATIENT REGISTER – UK

PERSONAL DETAILS OF PATIENT *(Please print)*
(To be Completed by Patient or Parent/Guardian of Patient under 18 years of age)

Surname		Title:
First Name(s)		
Date of Birth		Gender: M/F *
Address		
Postcode		
Tel. No.		
Email Address		
MEDICAL DETAILS		
Date of Diagnosis		NHS Number:
Name of Consultant & Hospital Details		

The details entered above relate to me/my child* and **I consent** to them being included in the Wilson's Disease Support Group – UK ***(WDSG-UK)*** *Wilson's Disease Patient Register-UK.*

Signed ..... *Dated* .....

Please print name .....
Patient / Patient's Parent/Guardian *

I consent/do not consent* to the ***WDSG-UK*** *Patient Register - UK* Coordinator contacting me in the future to ask me to take part in research projects and medical surveys relating to Wilson's disease that are approved by the NHS Group which approves and commissions research.

Signed ..... *Dated* .....

Please print name .....
Patient / Patient's Parent/Guardian *
*(*Please delete as appropriate)*

Please return the completed form to: **Mrs Valerie Wheater, WDSG-UK Patient Register Coordinator, 38 Grantchester Road, Cambridge CB3 9ED.**

WDSG-UK Patient Register-UK complies with the 1998 Data Protection Act. The guidelines of NIGB (National Information Governance Board) have been followed to ensure that this data is handled correctly.

A sample registration form for the new Patient Register

Meanwhile, for all members of the Group who are eligible to join *The Register,* may I please encourage you to do so. An information sheet and registration form are enclosed with this newsletter. Please consider completing the form and returning it to Valerie, the *Patient Register Coordinator*, in the attached sae.

Jerry Tucker
March 2015

Genetic Mutations in Wilson's Disease **by Mary Fortune**

Like many of you, I have had genetic testing for Wilson's disease. In my case this was done in 2007 after my transplant. The report states that I am "*a compound heterozygote* for the p.Q260fs, c.778dupC mutation in *exon 2,* and the p.G1221E, c.3662G>A mutation in *exon 17* of the *ATP7B* gene." But what does this mean?

Wilson's disease is caused by the inability to produce functional *ATP7B* proteins, which are required for copper transport, leading to an accumulation of copper in the body. Generally, people have two functional copies of the *ATP7B* gene; one inherited from their mother, and one from their father. Wilson's disease is a recessive genetic disorder - in order to have the disease, both your copies of the gene must be malfunctioning. People with only one malfunctioning gene are known as *carriers* since they are able to pass the mutation on to their children; they themselves may have mild copper metabolism abnormalities, but are not likely to have significant problems as a result. I am a *compound heterozygote*; I have two Wilson's disease causing mutations, one from each of my parents. However, they are not the same mutation ("*hetero"* comes from the Greek word for different - someone with two identical copies of the gene would be called a *homozygote*). Currently, over **300** different mutations in the *ATP7B* gene leading to Wilson's disease have been recorded.

Your genetic instructions are contained within the DNA in your cells. There are four possible DNA bases; **adenine (A), cytosine (C), guanine (G) and thymine (T)**. These bases **A, C, G** and **T** (think of them as *letters*) within the *ATP7B* gene can be thought of as making up a *book,* giving the cell instructions on how to make the *ATP7B* protein. Every three of these bases makes up a *word* within this *book,* which instructs the cell which amino acid to make; these amino acids form chains which then make up more complex proteins. Mutations in the *ATP7B* gene correspond to spelling mistakes within the *words.* The first of **my** mutations is known as a duplication - at a certain position within the gene, I have **two** copies of the ***C*** base, rather than **one** (which is normal.) Now the extra base has been inserted, the *words*, which are still read three letters at a time, are not the same three *letters* as previously, and so incorrect amino acids are produced from this point on. In my second mutation a *word* has been spelt wrong, and instead of the standard *G* base, I have an *A*. This causes my cells to make the wrong amino acid here, and I produce a Glutamic acid instead of the Glycine which the *ATP7B* protein requires to function correctly in order to eliminate copper.

Genetic testing services such as 23andMe offer to provide a personal genomics test. If you were to get yourself tested using this, I expect you would find you had far more than two mutations in your *ATP7B* gene. In fact, some mutations are so common that almost as many people have the mutation as don't. So why don't more people have Wilson's disease?

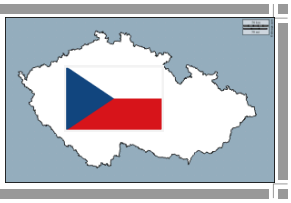
Both of my mutations are in the *exons* of the *ATP7B* gene - these are the sections of the gene which are put together to make the protein. However, between the *exons* are long stretches of DNA which are discarded when the gene is *read* (although we now know that these stretches may have subtler functions.) In general, mutations in the *ATP7B* gene which are not in the *exons* are unlikely to have a significant effect.

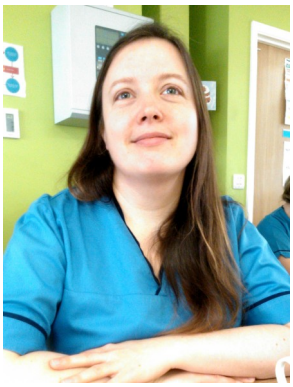
However, even if they are in the *exons* of the gene, some mutations don't have any effect on the protein produced. If I am reading a book, I can still understand a sentence regardless of whether the author writes *colour* or *color*. Similarly, there is more than one way to *spell* an amino acid. Glycine, the amino acid which one of my mutations spells wrongly, can be spelt *GGA, GGC, GGG or GGT.* So, if I had been fortunate enough to have the mutation in the third letter of this *word* instead of in the second, as mine is, I would still have produced the correct protein, and would never have known that Wilson's disease even exists. Alternatively, a mutation may change the protein, but in a way which allows it to still function, although possibly in a different way. It may be that someone has a mutation which changes the *ATP7B* protein, but which doesn't interfere with its ability to transport copper.

The *ATP7B* protein is made within the liver - so technically, all that matters is whether your liver has the Wilson's disease causing mutations. This is why a liver transplant is a cure for Wilson's Disease, provided the donor liver is able to make functional *ATP7B* (although this is a good example of a case where the cure is often worse than the disease!)

Lenka's Story

by Lenka Jonasova





Hello, my name is Lenka and I am 33 years old. I was born in Prague in the Czech Republic, although I am currently living in Edinburgh where I moved in 2002. Some of you may know me from the *WDSG-U*K Facebook page, which I joined eighteen months ago. I have been asked to write about my experience of Wilson's disease.

It all started in **1993** when I was about 12 years old. I was always very tired and was sick a lot, so I was taken to the doctor's where I had some blood tests done which showed elevated liver enzymes. I remember the hospital ringing on a Sunday morning to say that I needed to see them for more blood tests as they thought that I had *hepatitis B*. This proved to be negative and my paediatrician started me on some medication to improve my liver function. During the next two years there was no improvement and I was referred to another doctor who did CT and ultrasound scans and also took more blood. I was given new tablets which improved my liver function briefly.

Meanwhile, my uncle told us that he had spoken about me to a doctor friend of his and as a result I was invited back to the hospital for yet more blood tests. Shortly afterwards I was called back in and was immediately admitted to the ward where more tests were carried out. I had my eyes checked for *Kayser Fleischer* rings (but there weren't any) I had to collect urine, I was given an *echo (echocardiogram*) and on the fourth day I had a liver biopsy. I was now aged 14 and after two years of being unwell, I was at last given a diagnosis of Wilson's disease.

Neither my parents nor I had ever heard of Wilson's disease as it hadn't affected anybody else in the family. We were basically told what it was and how important it was to take the medications as prescribed. Two weeks later I returned to school with a bag full of tablets and instructions when to take them, which was difficult to remember. By the age of 16-17 it all started to get to me and my consultant agreed to reduce the number of tablets I was taking (between 12-15 a day) so that I was just taking penicillamine and Vit. B supplements. I was interested in *WD* and started to do my own research, although sadly I don't think my parents ever fully understood it. I never felt I was given the support that I needed and even now they ask if I still have to take my medications every day!

Wilson's disease never really stopped me doing what I wanted to do in my life, although I had been told not to play sport at school which I had always loved. I had no neurological symptoms and didn't mind having to give up chocolate and other foods that were high in copper. I came to Scotland in **2002** when I was 21 years old and worked as an au pair, before joining the *NHS* as a clinical support worker. In **October 2010**, in the middle of my final year at university where I was taking a nursing degree, I gave birth to my beautiful son, Jamie. My pregnancy went smoothly with no problems at all. On the advice of my consultant in Prague, I reduced my usual dose of penicillamine by half to 500mg a day and stayed on this dose while breastfeeding until Jamie was one.

In **Spring 2011** I started to have problems with my hips when lifting Jamie up or walking upstairs. I did nothing about it and after 3 months the pain went away. However, later my friends and I started to notice a difference in my movements. As I wasn't in pain, I didn't mention it to my GI consultant until **August 2013**. He did a blood test and found that my muscle enzymes were very high (2,800 or so.) I was told to stop taking penicillamine immediately. I had no alternative medication until **January 2014** when I was put on trientine (1200mg /day.) In the meantime I was referred to rheumatology and had a chest X-ray, *echo* and *EMG* (*electromyogram*, which measures the electrical activity of muscles at rest and during contraction.) Everything was within normal limits. An emergency muscle biopsy was ordered which I had in **May 2014**. Finally, I was given an MRI of my shoulders, hips and thighs which showed nothing abnormal. My muscle enzymes are still out of range, while the muscle biopsy came back showing borderline abnormalities.

I had a recent appointment with a genetics/neurology consultant in Edinburgh who requested more tests on my muscle, a review of my MRI and a repeat of my lung function tests. Sadly, I have started to notice tremors in my hands and sometimes I think I have slurred speech as well. I strongly believe this could be stress related but want to speak to my GI consultant about it. I would also like to have an MRI of my brain, just in case.

Well this is my story so far! I completed my nursing training in January 2012 and although my doctor says that I really shouldn't work, I continue to do so; so I guess it's not too bad. I love nursing and find that work helps me to get by, keeps me moving and to be honest keeps me sane!

Copper Levels in Food: *An Approximate Guide*

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 - HIGH		**Liver:**	
All fresh, frozen & tinned meat;	L	Brain – calves'	0.42	(varies according to animal - consult list on right...)		Calves'	23.86
Poultry (dark meat higher in cu)	O	Heart – lambs'	0.66			Lambs'	13.54
		Kidney – pigs'	0.84			Cows'	6.40
Fish - White/Oily	W	**Meat:** Duck/goose	0.34			Pigs'	3.75
Salmon/Tuna	0.04	Venison	0.36			Chicken	0.52
		Stock Cubes (Beef)	0.70				
Dairy		**Nuts:**		**Nuts:**			
Milk: Cows'	0.02	Peanut Butter	0.70	Cashews	2.20		
Goats'	0.05			Brazil	1.76		
Butter	0.03			Pine Nuts	1.32		
Cream: Single	0.20			Pecan	1.07		
Double	0.13			Hazelnuts	1.23		
Cheese: Cheddar	0.03			Walnuts	1.34		
Yogurt: Natural	0.04			Pistachios	1.00		
Flavoured	0.08			Almonds	1.00		
Eggs:		**Dried Fruits:**		**Shellfish**		**Shellfish:**	
Whole	0.10	Peaches	0.63	Crayfish	2.00	Scallops	10.00
Yolk	0.30	Currants	0.48	Calamari (squid)	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	0.38	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	**Seeds**			
Pears	0.10	Banana	0.21	Sesame	1.46		
Grapes	0.10	Kiwi	0.30	Sunflower	2.27		
Oranges	0.07	Lemons	0.26	Pumpkin/Squash	1.40		
Vegetables: Fresh		**Vegetables:** Fresh		**Vegetables:**			
Root Vegetables	0.08 - 0.20	Mushrooms	0.78	Beans -Butter (raw)	1.22		
		Asparagus	0.20	Soy (Edamame)	1.10		
Greens	≥0.06	Beans - Broad	0.43	Tomatoes			
		Haricot (raw)	0.61	(Sun Dried)	1.40		
Salads		Mung (raw)	0.97	(Tomato Puree)	0.53		
Lettuce, peppers	≥0.05	Red Kidney (raw)	0.61	(Tomato Ketchup)	0.40		
		Baked	0.21	Brown Sauce	0.33		
Pulses		Peas (raw)	0.23	*(Bovril)*	0.45	Bakers' Yeast (dried)	5.00
Lentils, split boiled	0.19	Chick Peas (cooked)	0.33	*(Marmite)*	0.30		
		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/crisps**		**Sweets/Chocolate**		**Chocolate Bar (≥70%)**	≥4.00
Cakes	L	Liquorice	0.39	Fruit Gums	1.43	**Chocolate Bar (<70%)**	<4.00
Ice Cream	O	Mars Bar	0.31			**Cocoa Powder (unsweetened)**	3.90
(NOT CONTAINING CHOCOLATE)	W	Bounty Bar	0.47	Drinking chocolate (sweetened)	1.10		
		Liquorice Allsorts	0.34				
		Bombay Mix	0.62				
		Twiglets	0.32				

Wild Carpathia

by Linda

Last year I had the opportunity to visit Romania; I've never really been much of a traveller so seized the chance! My niece Suzanne and her partner Robin *upped sticks* and emigrated there three years ago. I think it was an immensely courageous and life changing step to take. They sold up, bought a huge shiny *Chevrolet Starcraft*, loaded what belongings they wanted to take and set off to drive there.

The Chevrolet Starcraft ready to go...

Their adventure started in Sibiu, Transylvania, where they spent quite a while house hunting. However, they eventually bought a property in Jugur, which is over the other side of the Southern Carpathian mountains from Sibiu. Sue had started to learn Romanian several months before they left England, but had little chance to use it as everyone in Romania wanted to practise their English! The Romanians are such warm friendly and hospitable people.

overland to Romania

My eldest brother Ray (Sue's father) and I set out on our trip from Luton on a very misty September morning. We had all manner of things for Sue that she couldn't buy in Romania; bath bombs seemed to be the most important as she now had a 'proper' bathroom…long story! We were to be met in Bucharest by their friend, Radu. He was a professional driver, he told us on our first meeting. He then proceeded to take us on the most hair raising night time drive we had ever had! I shall never forget speeding on to the main road from the airport, when Radu suddenly slammed on his brakes and did a lot of cursing. In front of us was a convoy of horses and carts laden with tree length logs, on top of which sat women and several children. As we passed them the driver of one of the carts was chatting away on a mobile phone! It was the most bizarre sight: Health and Safety in this country would be apoplectic!

Robin, Suzanne & I

Next morning we had our first proper sight of the village. It was a clear sparkling morning and it was like stepping back in time; horses and carts, sheep & cattle bells tinkling, wild flowers, and most of all it was quiet and peaceful with no roaring traffic or sirens or anything! It was unbelievably beautiful. Almost all of the houses have a well on their property and for most this is their only source of water. So as you can imagine, life, particularly in winter months, can be hard. Sue and Robin have a well, which was their source of water for several months, but they are now connected to the mains. The well water is used for watering their garden, which became my task one evening. Once I got the hang of drawing the water, I enjoyed it immensely. One thing I really liked is the way life is so geared to the seasons; the sowing of crops, then the collecting and drying of herbs and medicinal plants in the spring, later on the preparation for winter, cutting and storing wood and preserving food: such a natural existence.

Ray & I at the well – wishing!

We spent several idyllic days exploring the area. We *the English people* were quite a novelty. Sue had told us the words for "Hello" or "Good day", which is ***Bu**nă se**a**ra*. Another charming greeting is *Saru' mana,* which is a respectful greeting to a woman (though not a very young woman) and roughly translated means "I kiss your hand." My niece is now really beginning to feel settled, though I'm sure there have been some trying times. She and Robin are both well liked and respected by their friends and neighbours. It seems the locals thought that they'd never settle, but they have proved them wrong. They are fairly self-sufficient, have learnt the local customs and involve themselves in the community. I must admit to feeling a tad envious of her way of life!

Doro and Viorica who live in the village

The village of Jugur is very picturesque being in the mountains, and with forests all around the air is so clean and fresh. I felt so much healthier for it! When Ray and I took a walk around the village on the first morning, we did notice a strong smell of alcohol every now and then. It turns out a lot of the villagers have a still on their property in which they distil there own *tuica.* *Tuica* is a national drink made from plums, and pretty good stuff it is too! On a visit to Campulung, the nearest town of any size, we noticed home brewed *tuica* being sold in HUGE plastic bottles at the fruit and vegetable market, which also sold fresh bread, pies, jars of pickles and jams.

A copper still for distilling tuica

I think my most outstanding memory is the trip up the *Transfagarasan Highway*, often referred to as Ceausescu's folly. The road was built between **1970-74** mainly by military forces, at a high financial and human cost. It connects the historic regions of Transylvania and Wallachia, and the cities of Sibiu and Pitești. It rises up and over Transylvania's Fagaras range, past spiky peaks and jagged waterfalls, as it climbs to a ridge between the country's highest mountains; Moldoveanu at **2,543m** and Negoiu at **2,535m**. The road runs through forests with notices telling you to beware of bears and also along the side of the beautiful Lake Vidraru, a lake created in **1966** by a huge dam on the Arges river.

A market stall in Campulung

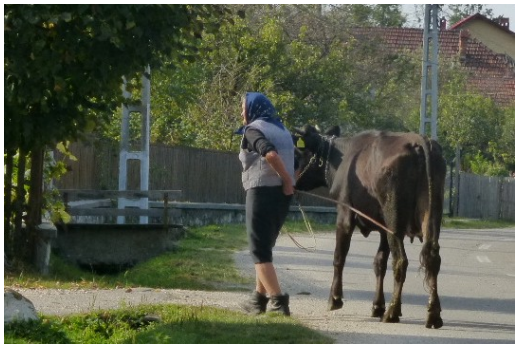
As you get above the tree-line the *fun* really starts; crazy, zig-zagging switchback roads clinging on to the side of dizzyingly high ledges. When we reached the top, we were in cloud but occasionally it cleared enough to give us spectacular views. In the winter an ice hotel is built up there with blocks of ice cut from Lake Balea at the summit and reached by a cable car bringing guests up from Sibiu. The route we took up there is closed as soon as the first snows come, as it is much too dangerous. I understand it closed the week after we left, which was late September, and won't re-open until at least May or June. On the way up we passed Poienari Fortress, the castle which served as the residence of Vlad the Impaler, the prince who inspired Bram Stoker's Dracula character, and "OH BOY", does it look scary!!

The Transfagarasan Highway

Another wonderful memory is sitting on the front porch in the early evening with a cold beer and watching the cows and sheep being brought back from pasture, bells a-clanking. Everyone waves a cheery greeting (not the sheep and cows of course!) In the past, times have been very hard for people in Romania. Whilst I was there I saw very few, if any, unhappy or stressed faces. There seems to be an enthusiasm and zest for life which is contagious; people were happy with what they had and contentment seemed to be the order of the day...bliss!

Poienari Fortress of Vlad the Impaler

All too soon our time there came to an end and I've never been sorrier to leave a place. Radu's help was once again enlisted to drive us to Bucharest, although this time he promised to go on better roads and drive slower. At least it was daylight, so we could see the many different small towns and villages through which we drove. The landscape was fascinating, changing from mountains and forests to fertile land full of orchards. There were apple sellers at the sides of the roads and Radu stopped several times and bought us different varieties of apples to try; it was a splendid journey!



The cows coming home!

I left Romania with lovely memories and very much look forward to returning.

Wilson's Disease seen in the Eyes

Most people who are diagnosed with Wilson's disease (*WD*) see an eye doctor (ophthalmologist) only once, usually when they are being diagnosed. Often this is when they are young, so some may not even remember visiting one. The ophthalmologist is there to tell the other doctors if the person has a *Kayser-Fleischer* (*KF*) ring (*see* ***figure 1.***) The *KF* ring is useful in helping to diagnose Wilson's disease in anyone who has unexplained neurology or liver problems, although not all people with *WD* have a *KF* ring.

What is a *KF* ring?

The *KF* ring is copper collecting within the thin layer of the cornea (window of the eye). This ring does not affect the vision or the eye. In the majority of people the obvious ring that we can see when shining a light on the eye disappears when they are on treatment. When ophthalmologists look at eyes with our specialist eye equipment (slit-lamp) we can sometimes see traces of the *KF* ring, even if the patient is well treated. So in Birmingham this made us think about whether we could use the ring to monitor the long-term response to treatment.

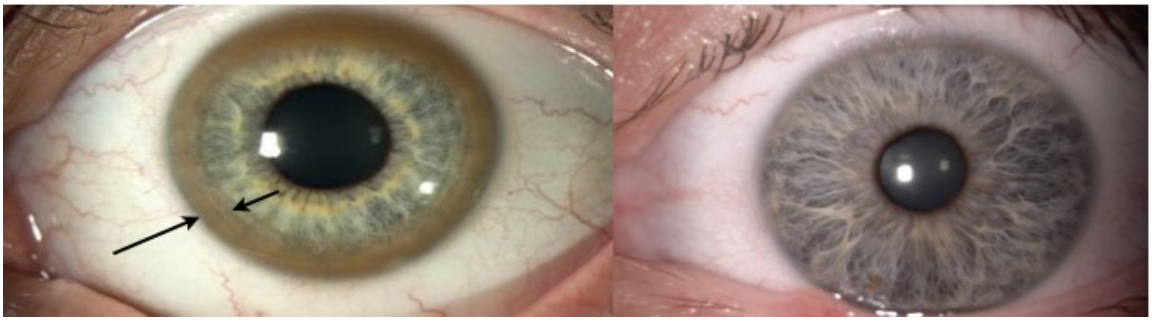


Figure 1: ***Eye (l) - WD patient with typical KF ring which is a brownish band seen in between the two black arrows*** ***Eye (r) - WD patient with no KF ring***

What's new in the eye department?

In the last 5-10 years, a new machine called an *OCT* (Optical Coherence Tomography) is helping manage lots of different eye conditions (from retinal problems to age related macular degeneration). It is what we call "non-invasive", as it does not touch the patient or cause any discomfort or problems when the pictures are taken. Every hospital eye department in the UK (and beyond) will have one of these machines because of the treatments that are now available for other eye conditions. The machine (*see* ***figure 2***) produces an instant detailed picture of the back of the eye, showing the various layers of the retina (*see* ***figure 3***)

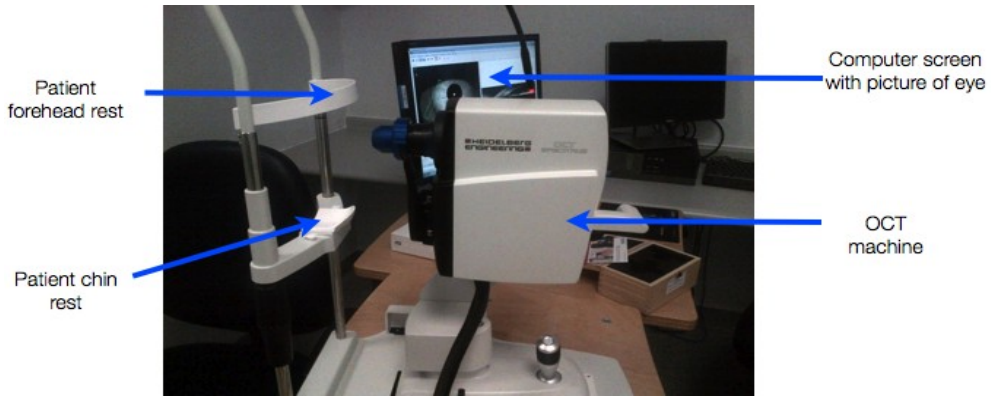


Figure 2: ***the OCT machine with the special attachment to take pictures of the front of the eye***

Since 2005 there has been a special attachment to the *OCT* machine to take pictures of the front of the eye, also called the anterior segment. This is not used widely in the UK, but some hospital departments have this attachment. ***Figure 4*** shows the picture we get from the anterior segment *OCT* (*ASOCT*): it is a cross-section through the cornea.

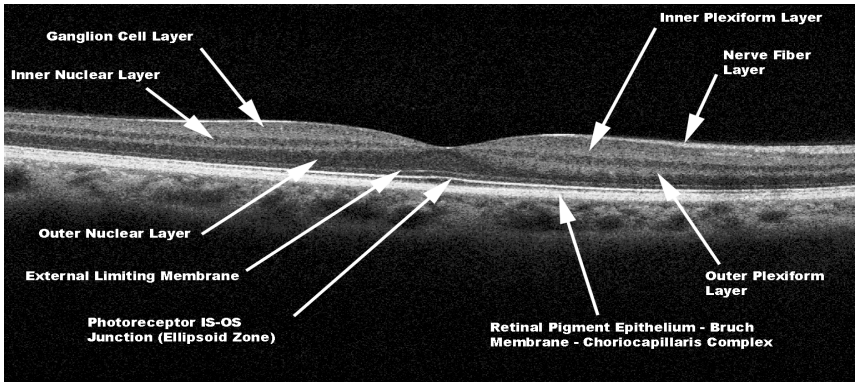


Figure 3: A very detailed OCT picture of the back of a normal eye (retina)

Changes seen on the *OCT* in Wilson's Disease

In Wilson's disease, if there is a KF ring present, the *ASOCT* picks up a highly bright band (*see* ***figure 5***). We have been able to scientifically prove that this brightness seen is different between people with *WD* and those without.

It should also be mentioned that copper deposition in *WD* occurs not only in the front of the eye, but also in the back of the eye (in the retina). This, at the present time, does not seem to cause a problem with how the eye sees, but it may be having a small effect that we have not yet been able to detect in clinic. One recent study, using *OCT*, has demonstrated changes and this requires more detailed investigation.

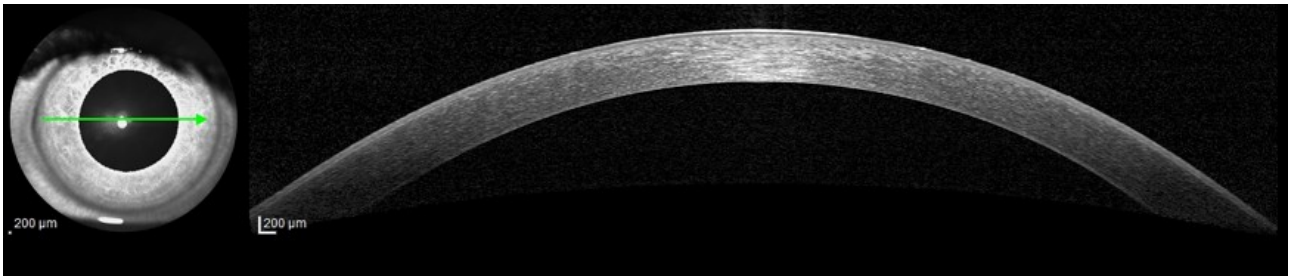


Figure 4: Normal eye picture imaging the layers of the cornea

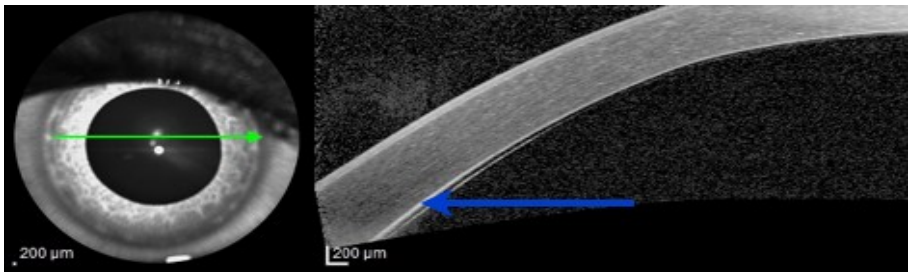


Figure 5: This picture shows the bright band (see blue arrow) seen in Wilson's disease with a KF ring

Future work

We now need to collect more information about whether this bright band can be matched to the levels of copper in the blood and urine. We also need to assess the change in the brightness over time, and in response to treatment.

We hope that these new pictures will possibly lead to a new way to diagnose and monitor *WD*.

If you would like any further information on the research that we are doing in Birmingham, then please contact me at:

Susan Mollan
Consultant Ophthalmologist, University Hospital Birmingham, Birmingham
susan.mollan@uhb.nhs.uk

Members' News

Our President **Dr John Walshe** follows the Group's activities with interest and is always willing to offer medical advice, when asked. Having devoted his professional life to the treatment and management of over three hundred Wilson's disease patients worldwide, he has a wealth of experience from which to draw. He will be ninety-five at the end of April and plans to celebrate his birthday with his two daughters Susan and Clare and their three grown-up children.

Dr Walshe – At Home March 2015

He continues to enjoy the surroundings of his beautiful home and garden in Hemingford Grey and to take holidays with the family in the UK. Last year Susan and her husband took him away for a few days to the picturesque village of Helmsley in North Yorkshire and this year they plan to go even further afield, visiting the Roman remains along Hadrian's Wall and the wilds of the Northumberland countryside.

He has pencilled in the date of this year's annual meeting in Cambridge (**12 July 2015**) and is very much looking forward to joining us again. He hopes that many of his old patients, together with new, will be there to share it with him.

* * * * *

A Brief Update

by Caroline

It is lovely to hear from **Caroline Simms**, who founded **WDSG-UK** with Linda Hart back in 2000. A research chemist by occupation, working in the pharmaceutical industry doing patent searching and analysis, she lives with her partner of seven years, Eamon, in Long Eaton in Nottinghamshire. Her hobbies include nature, wildlife and creative cookery and she has recently been commandeered into calling the numbers for bingo at her nearby social club, where she can also be found organizing quiz nights for local charities! She writes:

Caroline

"I cannot believe it is practically 11 years since I had my liver transplant at Queen Elizabeth Hospital, Birmingham. The call came through at 11.15 at night and I was told to get my bag packed and be ready for the ambulance to arrive in fifteen minutes and take me the 70 miles from my parents' house to the hospital. Sometimes it seems like it was just yesterday, other times it seems like a distant memory.

Diagnosed with Wilson's disease in 1987, when I was 14 years old (you can work out my age if you like), life was difficult. I presented with neurological symptoms initially, which did improve with trientine. However, I also had chronic cirrhosis of the liver and seventeen years later I was lucky enough to be given a successful transplant. It was Easter 2004 and I guess that it was a better gift than any chocolate egg could have been! It changed my life completely.

After a lengthy recovery my health, and in particular my neurological symptoms, were greatly improved. Currently I remain well and apart from taking immunosuppressant drugs, I live a pretty normal life. I would be happy to share my experience with any patients reading this who are considering having a transplant or are currently on an organ waiting list. Please get in touch with me via Linda or Valerie and I will try and answer your questions."

Wedding

Congratulations to **Liz Morell** on her marriage to **John Wood** on 30 December 2014 at Southend Register Office. They were attended by their four children and Liz's parents. A reception was held at *The Roslin Hotel* and a honeymoon taken over Valentine's Day weekend in the romantic setting of Lavenham in Suffolk. We wish them both much happiness in the future.

* * * * *

Allie Johnston writes to bring us up to date with her news. Unfortunately she and her mum had to cancel their planned trip to Turkey last year as they decided that the practicalities of sailing in a gulet would have proved too difficult. Instead Allie arranged two alternative breaks which she tells us about now.

First of all in June I went off to stay with my friends John and Alison, two lovely people that I met on my honeymoon in Kenya in May 1997. They have a fourteen year old daughter called Anya, who is my Goddaughter.

I hadn't seen the family since I was diagnosed with Wilson's disease five years ago and neither had I been away by myself since then. However, I took the train from Edinburgh to Chesterfield and they came to meet me from the station and drive me back to their house in the little village of Tibshelf. It was so nice to see one another after all this time. Alison and I stayed up talking until midnight as so much had happened since last time we'd met. It was a lovely weekend. We went to Anya's school fete where I won a foot spa on the tombola! And the weather was so warm that we sat out in the garden and John cooked for us on the barbecue both days. But it was all too short and with tears we parted arranging to meet up again for Anya's birthday later in the year.

Ten days later and I was back on the train, but this time with mum and my thirteen year old niece, Suzanne, who we took to Blackpool for a five day break. We had an action packed holiday staying in the *Grand Metropole Hotel* and visiting the *Blackpool Tower Circus*, the water slide flumes at *Sandcastle Water Park*, the dodgems on *Central Pier,* and the magical Tower Ballroom where we had tea and danced. We spent the last day at the Pleasure Beach taking in all the rides before returning home exhausted and needing another holiday!

Alison, Anya and Allie in Tibshelf

Rita, Suzanne and Allie in the Ballroom at Blackpool

Update Story for Olivia

by Bongkeun Jeon

Olivia with her Gingerbread jar

Last year this time, through the email conversation with Valerie, I was encouraged to write Olivia's story for ***WDSG*** newsletter. Still thinking that my writing was not particularly well written, with Valerie's support it was published. It was just appreciated as it was my first public writing.

My daughter, **Olivia**, was diagnosed with Wilson's disease in South Korea when she was two and we were visiting on a two year Sabbatical. Returning to the UK was a bit scary not knowing much of WD treatment here. However, we are having good treatment and meeting lots of kind doctors and nurses at *King's College Hospital*. Last summer my wife, Sunhae, Olivia and I came to the *WDSG-UK* meeting in Cambridge. Olivia won a raffle prize of a gingerbread biscuit jar donated by Charlie. It was such a great opportunity to meet committee members, doctors and Univar representatives. Everybody supported and encouraged us so much.

We have had some difficult times as Olivia's liver and pancreas levels were/are still higher than normal. Her tummy ache has been ongoing problem. How bad feeling my wife and I cannot do much support for Olivia's consistent tummy aches unless rubbing her tummy whenever she complains. Doctors at *King's* told us that it was very strange her pancreas level was still high. Olivia had some tests including MRI and ultrasound scans. Fortunately, they turned out to be normal but it is worrying that her levels are still high. *King's* doctors suggested contacting the professor of *WD* in Korea, but sadly Dr Seo retired last year. Always up and down circumstances with Olivia; we had non-stop battling getting the medicine on time, but we cannot complain as we use all the *NHS* system.

I have read some of articles and messages on *WD facebook* site, which are often upsetting. Apart from some difficulties with Olivia's health, Olivia has been doing well in her school (now she is five years old and in her reception) and she has made lots of friends. I have moved my work to Chichester College last year, so only able to see her and Sunhae during the weekend which is sad, but this is the best option for a while.

Thank you so much for Valerie and all other chair members, doctors, other patients and family who encouraged Olivia. Our family hopes that God blesses for all *WD* patients and family. Looking forward to meeting all members and doctors again at this year's *WD* patients' meeting in July.

* * * * *

Ashok

Ashok Pandit, a Wilson's patient from Nepal whose story featured in our **2012** newsletter and whose contributions to our fb site and newsletter are always widely appreciated, writes the following:

"So far this year I didn't have any health problems, which made it a wonderful year for me. During the middle of year, I had to face some problems regarding my medication due to its shortage, but that didn't hamper me because I had enough medicine in stock.

I have finished my studies, but as you all know I can write but not as normal people do and I arranged a writer for the examination but he didn't respond my call on the day and so I couldn't do my exam. I have worked so hard last year on my studies so when the writer didn't turn up it was so depressing. I then thought that college degrees don't determine my knowledge so I have left my formal studies but have been learning many stuff on my own.

Moving on to my career you can call me anything like webmaster, SEO expert, SEO consultant. Yes, finally I am there looking to make my career in SEO (Search Engine Optimization). I can now help you earn more profit from your business by taking your business to next level, by promoting your business online, through your official website. You can contact me through my website: www.seoconsultant.com if you think I can help you. I am also working hard for content writing, these days; hopefully I would get success on it also.

As WD patients we all need to have a hope that one day our every problem would be solved. Finally my financial problem is going to be solved. So far I am so happy because from now onwards I don't need to rely on anyone for funding my medicine. As from now onwards I will be earning myself. Along these years Dr. Alex Dalzell and Mr. John Ross were with me helping me get my medicine. I couldn't remain without saying thank you to these two wonderful souls who were with me in my endeavour. I also want to thank *WDSG-UK* who has given this little space to put my words in this year's newsletter."

WDSG-UK 2015-16 EVENTS

Date	Time	Event
Jun 20	**0900 - 1830**	Morbus Wilson e.V. Annual Symposium - University of **Heidelberg,** Germany
July 12	**1100 - 1530**	**WDSG-UK Meeting and 5th AGM** – Cambridge Rugby Union Football Club Grantchester Road **Cambridge** CB3 9ED.
August 22	**1000 - 1200**	Sylvia Penny warmly invites you to **a Coffee Morning, Bring and Buy and Raffle** in aid of **WDSG-UK** at Victoria Park Methodist Church, St Marychurch Road, Claymore, **Torquay.**

WILSON'S DISEASE MULTIDISCIPLINARY CLINICS

The Birmingham WD Clinic

Dr Gideon Hirschfield (Consultant Hepatologist) and **Dr David Nicholl** (Consultant Neurologist) hold a one-stop Wilson's disease clinic at **University Hospital Birmingham** on a Friday morning four times a year. This clinic offers patients the opportunity to have their management reviewed by a hepatologist and a neurologist at the same time and is intended to supplement otherwise established care. Referrals must come from the hospital physician looking after the patient and should be addressed to *Dr Hirschfield* at *Queen Elizabeth Hospital, Mindelsohn Way, Edgbaston, Birmingham, B15 2WB.*

The Sheffield WD Clinic

The Sheffield clinic is jointly run by **Dr Godfrey Gillett** (specialist in inherited metabolic diseases and adviser to WDSG-UK), **Prof Oliver Bandmann** (Consultant Neurologist) and **Dr Mohammed Karadjeh** (Consultant Hepatologist.) Clinics take place every six months at the **Royal Hallamshire Hospital, Sheffield** on a Tuesday afternoon. Referrals should be addressed to *Prof Oliver Bandmann, Dept Neurology, Royal Hallamshire Hospital, Glossop Rd, Sheffield, South Yorkshire S10 2JF*. Both GPs and hospital specialists can refer to this Sheffield WD clinic.

IN MEMORIAM

We were very sad to hear that **Maureen Adams** had died in Suffolk on 14 August 2014 aged 64 years. Maureen had been a patient member of WDSG-UK since the Group was formed in 2000 and had attended nearly every gathering since. Her presence at future meetings will be sadly missed and our deepest sympathy goes to her husband Fred and sister Anne, who kindly chose to hold a collection for us at her funeral raising the sum of £513.00.

* * * * *

We were also saddened to learn of the death of **Ron Shaw** from Berkshire on 21 November 2014 at the age of 80. He and his wife, Sue, had celebrated their 40th wedding anniversary only a few days before. Ron was one of the first patients in the 1950s ever to be diagnosed with Wilson's disease and we were pleased to meet him and Sue at our meeting in Cambridge in 2013. We send our condolences to Sue, and thank her for holding a collection for WDSG-UK at Ron's funeral which raised £537.00.

Wilson's

Wilson's Disease Support Group – UK

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Dr John Walshe Honorary President, World Authority on Wilson's disease
Dr Godfrey Gillett Group Adviser, Honorary Member
Dr James Dooley Group Adviser, Honorary Member
Dr Caroline Simms Group Co-Founder

WDSG-UK

Tell others about WDSG-UK

Please encourage anybody else that you know with Wilson's disease to join **WDSG-UK**

Inform your family, friends, consultant physicians, general practitioners and local MPs about the work of **WDSG-UK.**

The more people who know about **WDSG-UK**, 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