Wilson's Disease Support Group - UK

NEWSLETTER

APRIL 2014

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 Everyone,

I am sure, like me, you will be sorry to hear that Linda lost her partner John in such sad circumstances shortly before Christmas after over thirty happy years together. While she picks up the threads left behind, she has asked me to take her place in introducing this year's newsletter.

So, I am delighted to welcome you to our 19th edition. It is bursting with varied articles, for which I would sincerely like to thank all our contributors. Running through these quickly, we have four ever popular patients' stories, three medical articles including the second one in Alan Stevens' *Simplified Guide* series (if you remember last year he talked about oesophageal varices), news of fundraising events held during the past year (get your pinnies on) and another question and answer session with Dr Walshe. Then there is the customary Chairman's report, an update from Jerry Tucker on the *Rare Disease Strategy—UK*, an article with photographs of last summer's annual meeting in Cambridge, members' news, a rather sparsely furnished calendar of events planned for 2014 (let us know if you have anything lined up for 2015) and finally a notice board advertising two multi-disciplinary clinics for Wilson's patients which are currently run in Birmingham and Sheffield.

Also worth a mention is our well-used *facebook* (*fb*) site, which at 270 members is growing by the day. It quite often proves to be the first port of call for newly diagnosed patients, and for people living in remote parts of the world their first contact ever with others suffering from the same condition. There are lively exchanges, with patients eager to offer the benefit of their experience and give useful advice and encouragement to other patients. If you aren't already a member and are hesitant about joining us, please give it a go. Access to the site is through the home page on our website *www.wilsonsdisease.org.uk* where all you have to do is click on the *fb* icon on the top left-hand side. It takes a minute or so to join (as long as you are on *fb* already), and if you find you don't like it, it's even quicker to leave!

Finally, please don't forget to put the date of our next annual meeting in your diary. It will take place on **Sunday, 13 July 2014** at our usual venue, the Rugby Club in Cambridge, (see details on booking form and AGM agenda enclosed.) It would be lovely to see as many of you there as possible. Meanwhile, membership for **2014-15** is now due and you will be pleased to note from the enclosed renewal form that fees have again remained the same. For the first time, however, our bank details have been included for those preferring to pay electronically.

Have a wonderful summer everybody and hope to see you on 13 July.

Valerie

IN THIS ISSUE ...

Chairman's Report for 2013-2014

The ability of ***WDSG-UK*** to represent its members' interests nationally and to raise awareness of Wilson's disease within the medical community has been further demonstrated in the past year. Our membership of ***Rare Disease UK*** continues to provide us with an insight into government policy on the ***UK Strategy for Rare Diseases***. Arising from this strategy, a new initiative to identify areas of expertise in the UK for the treatment of Wilson's disease has resulted in the participation by *WDSG-UK* in (to date) two meetings of the ***Wilson's Disease Network – UK***. And the past year has seen a steady reliance on our expanding *WDSG-UK* ***facebook*** site for interacting with our members, both here in the UK and abroad.

WDSG-UK Meetings

The *WDSG-UK* management committee met in May and October 2013 at Knowle and Cambridge respectively. We held the third *WDSG-UK* AGM as part of our annual meeting for members, family and friends in Cambridge on 21 July 2013. A report of the annual get-together in Cambridge, which **Valerie** and **Linda** again so ably organised, appears on ***p4*** of this Newsletter.

Donations and Fundraising

I should like to thank members, their families, friends and sponsors for the magnificent additional income of **£3,000** which they brought in through donations and fundraising in **2013-14.** I would particularly like to thank **Univar** for continuing to sponsor us so generously, **Dr David Nicholl** for kindly nominating *WDSG-UK* to receive an honorarium due to him, **Belinda Diggles, Sylvia Penny** and **Allie Johnston** for their tireless fundraising and **Emma Collcott's** family for holding a collection in aid of the Group at a recent family funeral. 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 **£185.00** on gift aided money from the previous financial year (**2012-13**.)

Facebook site

Visitors to the *WDSG-UK facebook* site (a Closed Group) will see that by the end of March 2014 we have approaching 270 members. It is to the credit of Valerie and Linda that the considerable number of queries, concerns, and requests for help arising from our *facebook* page are all answered with skill, sympathy and efficiency. What we would like to see as a consequence of this web activity is for more *facebook* followers to become members of *WDSG-UK*, which involves payment of a small annual subscription. At present the number of paid-up members of *WDSG-UK* for the year 2013-2014 is around 51. As a result, we are too dependent on a commercial sponsor to pay for the Newsletter and our annual meeting in Cambridge. The subscription to join *WDSG-UK* is a modest sum, and even for our many overseas *facebook* members well worth the investment in order to secure the existence of *WDSG-UK*. With an increased financial support we could offer greater support for patients, and even consider sponsoring research into Wilson's disease.

British Liver Trust's *(BLT)* 5th Annual Patient Support Groups' Representatives' Conference

Jerry and Valerie represented *WDSG-UK* at the *BLT's* two-day conference in Birmingham last November. Among the five speakers was Andrew Langford, the Chief Executive of *BLT*, who spoke about his involvement with an All Party Parliamentary committee which is holding an inquiry into the prevention, diagnosis and treatment of patients affected by liver disease.

Rare Disease UK

At the *Rare Disease UK* AGM, held at the Royal College of Paediatrics and Child Health, London, on 22 January 2014, the Chair of *Rare Disease UK*, **Alastair Kent**, summarised the main features of the ***UK Strategy for Rare Diseases***, which was launched in November 2013. "For the first time since the establishment of the *NHS*, patients and families with rare diseases have a clear and strong statement of the commitment of Government to meeting their needs. This is a real breakthrough." On behalf of *WDSG-UK*, our Vice-Chair, **Jerry Tucker**, has been closely following the fine detail of the *Strategy* and how it will affect the treatment of patients with rare metabolic genetic disorders. Jerry provides a summary opposite. Also at the *Rare Disease UK* AGM, there were two presentations on clinical trials (of drugs) and a detailed account from the UK Department of Health on the funding and support for research into rare diseases.

Wilson's Disease Network – UK

Jerry mentions in his summary the development of specialised services for the treatment of rare diseases. As a consequence of the proposals outlined in *UK Strategy for Rare Diseases*, and the current reorganisation within the *NHS*, **Dr James Dooley**, **Prof Oliver Bandmann** and colleagues who treat patients with Wilson's disease have formed an informal working group with the aim of establishing a UK-wide network of specialists who have experience in the diagnosis and treatment of Wilson's disease. As well as physicians, this working group ***Wilson's Disease Network–UK*** also includes clinical scientists who carry out laboratory tests for the diagnosis of Wilson's disease. Members of the *WDSG-UK* management committee were invited to attend two meetings of the *Wilson's Disease Network–UK* held in 2013 in London. A third meeting will take place in Birmingham in June this year.

WDSG-UK Annual Meeting and 4th AGM

The 2014 Support Group Meeting has been arranged for **Sunday, 13 July 2014** at the clubhouse of the city of Cambridge's Rugby Union Football Club. During the course of this meeting the **4th *WDSG-UK* AGM** will be convened. An agenda for the AGM is included with this Newsletter. As part of the AGM, the election of officers and members of the *WDSG-UK* Management Committee for the year **2014-2015** will take place. All members of the current committee have submitted their names for re-election for this period.

Once again I thank the members of the *WDSG-UK* management committee, **Linda Hart, Valerie Wheater, Jerry Tucker, Anne-Marie Le Cheminant and Mary Fortune** for their hard work and enthusiasm during the past year. We are confident that the changes now taking place within the *NHS* will benefit Wilson's disease patients.

Rupert Purchase
March 2014

Update on Rare Disease Strategy in the UK

by Jerry Tucker

Rare diseases are increasing as diagnostic procedures improve and in the UK more than 3 million people have a rare disease. Over 80% of rare diseases have a genetic cause and 50% of all new cases are children. Consequently rare diseases as a group are not uncommon and the UK already uses significant resources within the *NHS* in treating patients. In the future, treatment for rare diseases will need to be delivered with greater efficiency and productivity whilst striving to provide an improved service for patients and their families.

In **2009** the European Union called on each EU State to have in place a documented ***Rare Disease Strategy*** by the end of **2013** to meet the goal of the strategy being implemented by **2020**. All four UK Countries have worked together to meet the 2013 deadline and have produced a report ***The UK Strategy for Rare Diseases*** (www.gov.uk/government/publications/rare-diseases-strategy) which outlines their approach to improving the lives of all who are affected by rare diseases. The strategy covers diagnosis and early intervention, identification and prevention, coordination of care, research and the empowerment of patients affected by rare diseases. A strong theme throughout the report is the importance placed on working with patient organisations and listening to the issues and problems patients face living with their illnesses, and incorporating their needs into the provision of lifelong care. All four UK countries have agreed to individually address the 51 commitments identified in the strategy document through their own unique national plan, but will continue to work closely with one another. The ***Rare Diseases' Stakeholder Forum*** will monitor the implementation of each country's strategy.

In England, ***NHS England*** is the group which has responsibility for commissioning specialised services in England, and is currently developing a detailed plan for Rare Disease services over the next five years. A national team within the Medical Directorate will document requirements in a *Rare Disease Annex* and coordinate the 75 Clinical Reference Groups who will provide advice on commissioning specialised services. *NHS England* will work with the *Rare Diseases Advisory Group*, the *Rare Diseases' Stakeholder Forum* and other health institutions to deliver the *Rare Disease Strategy.*

An integrated strategy across the UK promises an improved and consistent medical service across the UK. Currently *WDSG-UK* is a member of a group within the *NHS* which is already reviewing how we can develop improved services for Wilson's disease patients. *WDSG-UK* is well placed to be the patient organisation assisting in the definition of services required in the future.

Wilson's Disease Support Group Meeting & 3rd AGM

Cambridge Rugby Union Football Club, 21 July 2013

It was good to see such a large turnout of *WDSG-UK* members for our annual meeting at the Cambridge Rugby Union Football Club in their tranquil grounds on the west side of the city. As well as our regular supporters, we were pleased to welcome **Bianca Klimsa** (from the German patient support group, Verein Morbus Wilson e.V.), **Ron Shaw** (a patient diagnosed in the 1950s) and **Charlie Watsham,** who together with her dog, Biggles, joined us from Lichfield.

Professor Niall Quinn

A very special guest last year was the distinguished Assyriologist **James Kinnier Wilson**, who is the son of the neurologist, Samuel Alexander Kinnier Wilson, the doctor who first recognised the disease. Many clinicians and scientists who have a special interest in Wilson's disease also travelled to Cambridge – **Dr John Walshe** (accompanied by his daughter **Susan**), **Kay Gibbs**, medical geneticists **Dr Richard Sandford** (*Addenbrooke's Hospital, Cambridge)* and **Prof Oliver Bandmann** *(Royal Hallamshire Hospital, Sheffield),* the Group's medical advisers, **Dr Godfrey Gillett** and **Dr James Dooley**, and from the *National Hospital for Neurology and Neurosurgery, Queen Square, London*, the neurologist **Prof Niall Quinn**.

James Kinnier Wilson

The morning's proceedings began with a welcome from **Linda,** a resumé of the Group's activities by Rupert Purchase and presentation from our Vice-Chair **Jerry Tucker** on the opportunities offered by the current reorganisation of the NHS for the development of specialised services for rare diseases. Jerry outlined the type of service that WDSG-UK would like to see commissioned by NHS England for the treatment of Wilson's disease and explained that the patients' voice and patients' support groups can play a key role in bringing these proposals to fruition.

Mary receiving a cheque for £1,000 from Graeme Manley of Univar

Professor Quinn then spoke to us about the early career of Samuel Alexander Kinnier Wilson at the National Hospital, Queen Square, London, in the 1900s and gave a description of some of the patients reported by Wilson in his landmark 1912 paper published in the journal *Brain*. Finally, James Kinnier Wilson gave a short address about his personal memories of his late father.

Lunch was an opportunity for guests to mingle and also buy some of **Allie Johnston's** beautiful handmade cards that she and her mother, **Rita**, had brought down to the meeting from Edinburgh. The proceeds (**£60.35**) from the sale of the cards were generously donated by Allie to the Group.

Linda, Charlie, Belinda & Anusha

The afternoon began with the formalities of the WDSG-UK 3rd AGM, during which the current management committee was unanimously re-elected for the year **2013-2014**. Some splendid prizes were on offer for this year's raffle, again organised by **Belinda Diggles**, which raised **£120.00**, and the Group's funds were then further enhanced by a donation from Univar Ltd of **£1000**, which **Mary Fortune** accepted on our behalf from **Graeme Manley** of Univar.

In 2013 another landmark paper on Wilson's disease was published in *Brain* – '**A Genetic Study of Wilson's disease in the United Kingdom**,' *Brain*, 2013, **136**, 1476-1487 – and we were particularly pleased to hear first hand from one of the co-authors of that paper, Prof Oliver Bandmann, about the findings and conclusions of this important study (*see* ***p14.****)*

Patients at the Meeting, with James Kinnier Wilson

Insightful questions were addressed to the 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.**

Fundraising - Christmas 2013

Belinda's Christmas Puddings

Once again one of our chief fundraisers Belinda Diggles took up wooden spoons, put on her Christmas apron, retreated to her kitchen and started the unenviable task of mixing and cooking **23** Christmas puddings for friends, relatives and Wilson's patients from all over the world. In so doing, she raised a total of **£125.00** for Support Group funds.

Belinda - Slaving over a Hot Stove...

She is not too sure how all this started, but for those who wonder what quantities of ingredients she uses, Belinda has given us her recipe below. She chooses to stir the mixture in a washing-up bowl because it is the biggest bowl that she owns. However, she assures us that it has never been used for washing up in its life!

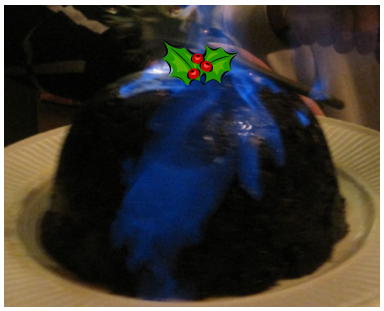
My *Secret* Xmas Pudding Recipe by Belinda Diggles

Makes:

7	**LARGE**
10	**Small**
6	**Very small (specials only)**

Ingredients:

1.350kg SULTANAS	1.500kg MUSCOVADO SUGAR
1.350 kg CURRANTS	1.350kg SHREDDED SUET
1.350 kg RAISINS	300g CHOPPED NUTS,
1.350 kg FRESH BREADCRUMBS,	300g MIXED PEEL
6 APPLES,	300g PLAIN FLOUR
6 LEMONS grated rind and juice,	6 TABLESPOONS OF MIXED SPICE
24 EGGS	½ BOTTLE BRANDY



The Proof of the Pudding...

Method:

1.

Weigh out the Ingredients...

2.

Put into a Washing-Up Bowl...

3.

Stir for Britain...

4.

Prepare to Steam...

That`s the easy bit! It takes a long time to steam 23 puddings and about 2 weeks for the steam to leave my kitchen! Will I do it again? With orders last year from Charlie Watsham and Valerie, and a repeat order already from Valerie this year, I will no doubt find myself back in the kitchen in October.

Anybody wishing to order and collect a pudding at this year's annual meeting (large £10; small £6), please let Linda or Valerie know by the ***END*** *of* ***MAY*** *(linda@wilsonsdisease.org.uk) or (val@wilsonsdisease.org.uk.)*

Full Steam Ahead

Fundraising - August 2013

Sylvia's Summer Bring & Buy Sale and Coffee Morning by Valerie

Emma Coombes was diagnosed with Wilson's disease at the end of 2006 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. As another of our chief fundraisers, Sylvia has raised over £3,000 during the past seven years. I have been wanting to show my support in person for some time, so last year I decided to book a week's holiday in Ashburton on the edge of the Dartmoor National Park and from there drive over to Torquay early on the Saturday morning to pay Sylvia and her family a surprise visit. They were setting up the stalls when I arrived and couldn't believe it when I told them who I was!

Sylvia

Being the head of a very close-knit family, Sylvia is able to rely upon the help of her daughter Sheila, granddaughters Emma and Daisy, great grandson and great granddaughter Tom and Amy and cousins Geraldine and Marjorie, together with Emma's other grandmother Hazel. Between them they manage the stalls, entertain the customers, advertise the Support Group and sell tickets for the raffle. I learnt from Sylvia that collecting goods for the stalls and prizes for the raffle is an ongoing task as people are making donations all year round. Such is the support of the local community that there were over forty raffle prizes on offer including a £60 voucher for a family of four to have a day's outing on the steam railway between Paignton and Dartmouth. Now I wouldn't have minded winning that!

Iris with Hansel & Gretel

Meanwhile, there were plenty of bargains to be found on the bric-a-brac stall which was run by Sheila, Emma, Tom and Amy and on the other side of the room Daisy was quickly selling out of her delicious home baked produce on the cake stall. Friends Mavis and Ann were in charge of refreshments in the kitchen and ninety-one year old Iris had little difficulty finding a new home for Sylvia's lovely hand knitted dolls Hansel and Gretel. The event was so successful that the total raised amounted to **£549** and on behalf of the Group I should like to thank everybody involved for their hard work, generosity and enthusiasm.

Mavis and Ann

In all it proved to be a very enjoyable occasion with a regular stream of people dropping in all morning including the Assistant Pastor Irene Cochran, who turned up to lend her support. I was pleased to have the opportunity to talk to everybody about Wilson's disease and the work of the Support Group. And for anybody planning a holiday in the south-west this summer, I can heartily recommend you look no further than a visit to Torquay on the morning of **23 August** between the hours of **10.00** and **12.00** o'clock (*see events board* p19.)

Selling Raffle Tickets

L-R : Geraldine, Sylvia, Daisy, Tom, Hazel, Valerie, Emma, Amy and Sheila

Emma, her daughter Amy and mum Sheila

Enjoying the morning...

Emma's Story

by Em Coombes

Hi! I'm Em. I'm Sylvia's granddaughter and the mother of two lovely children, Amy and Tom aged 15 and 12. I was born in Torquay at the end of **1980** and have been here ever since. It is a wonderful place to live on the south coast of Devon. In the summer the town is bustling with holiday makers and because of its setting and favourable climate it is often referred to as *The English Riviera.* Torquay is also the birthplace of the popular crime writer, Agatha Christie, who was born here in **1890** and used many of the nearby places as settings for her plots. Gran once had the pleasure of meeting her at *Greenway,* her home in Torquay, when she was asked to assist with some upholstery repairs that were needed in the house.

Emma

We were pleased to meet Valerie in Torquay in the summer and she mentioned it would be nice if I wrote an article for the next newsletter about my experience of Wilson's disease. First of all, let me say that I never had any health issues while I was growing up. I was very happy and popular among my friends. I met my partner David, who is the father of my two children, soon after I left school and we started living together at the end of the 1990s. It wasn't until I was twenty-five that my health began to deteriorate. In the summer of **2006** I noticed that I was starting to lose control of my arms, and my feet weren't working properly either. Things got no better and so I made an appointment to see my G.P. a few weeks later. He arranged for me to attend our local hospital in Torbay, where I was seen by a neurologist who performed various tests but could offer no explanation as to why it was happening.

Then one week at the end of **2006** I became so ill that I had to take to my bed. I was feverish, with a high temperature and rash all over my body. Later I became so delirious that David sent for the doctor who came out and immediately arranged for me to be admitted as an emergency to Derriford Hospital in Plymouth, which is some thirty miles away. I was treated successfully and discharged one week later. While I had recovered from the fever my neurological symptoms were getting gradually worse. It was now the beginning of **2007** and I was starting to drool and have difficulty swallowing, eating and talking. I was losing my balance and falling over. I returned to the G.P., who referred me back to Torbay to see a visiting neurologist from Derriford called Dr Edwards. More tests were carried out, but still everybody was baffled.

Then one day Gran was talking to her friend about me and the friend's husband, Carl Penn, went on to his computer and entered a list of my symptoms. Two conditions seemed to fit the description: Huntington's disease and Wilson's disease. He printed out the details of both and gave them to Gran and she in turn gave them to my mum Sheila, who showed them to my G.P. I was referred for a third time to Torbay Hospital and again I was seen by Dr Edwards who ran yet more tests. This time Kayser-Fleischer rings were noted and a 24 hour urine collection confirmed that I did indeed have Wilson's. Dr Edwards said that I had Gran to thank for finding out about Wilson's. I was prescribed penicillamine and given a list of foods high in copper that I was told to avoid.

The drugs worked well and after only a couple of months there was a marked improvement. However, later in **2007** the penicillamine presented problems, so I had to stop taking it and change to trientine. I was still having difficulty talking, so I went to see a speech therapist. Eating also was a challenge but I was lucky that mum would come round to feed me, cook me special meals, help in the house and look after Amy and Tom taking them to school and back each day.

But, I am a fighter and fiercely independent and by the middle of **2008** I was so much better. My speech had improved and my walking was becoming easier. I still had difficulty feeding myself, not least because the fingers on my right hand were permanently cramped and touching my palm making it very awkward to hold cutlery. I had an operation on them in **2010** and although there was a slight improvement they have never returned to normal. Otherwise, seven years later I am back to my old self. I no longer need help doing things and try to live life to the full. In **2012** I fulfilled a lifetime's ambition to do a sky dive which was a great achievement. Who knows what challenge I will take on next?

Torquay harbour

The Normal Liver - What does it do? - A Simplified Guide A.Stevens

The liver is a complicated structure which can be regarded as a vast chemical factory that imports many raw materials, processes them and then exports the finished products. Like any efficient factory, the liver needs:-

- an efficient and constant **input system**, bringing raw materials in;
- an efficient **output system**, taking the finished products out to the sites where they are needed; and
- multiple **work units** capable of working 24 hours a day.

The Input System

The Input system comprises two blood vessels:-

a) the **hepatic artery,** which brings oxygenated blood and some of the raw materials into the liver, as well as potentially toxic substances resulting from the breakdown of body cells, requiring detoxification in the liver, and

b) the **hepatic portal vein** which is a closed vein system bringing blood directly from the intestine and spleen. The blood is rich in raw materials such as sugars, amino acids and fatty acids produced in the intestine by breakdown of carbohydrates, proteins and fats in food. It also contains some of the breakdown products of red blood cells in the spleen for processing in the liver and converting to **bile.**

These two blood vessels are shown in ***Diagram 1***. They enter the liver at the back, whereupon both vessels branch extensively, taking blood to all parts of the liver and bringing the blood into contact with the rows of **work units** by thin walled channels called **sinusoids**, which are abutted to the work units (see ***Diagram 2***.)

The Output System

The Output system has two components:-

a) the **hepatic vein** which is a large vein taking blood from the liver back to the heart for redistribution throughout the body, taking the products of the liver to the sites where they are needed; and

b) the **bile duct system** which takes the bile made in the liver to the **gall bladder,** where it is concentrated and stored until it is needed in the small intestine to assist in the digestion of food materials, particularly fats.

These output systems are also shown in ***Diagram 1*** opposite.

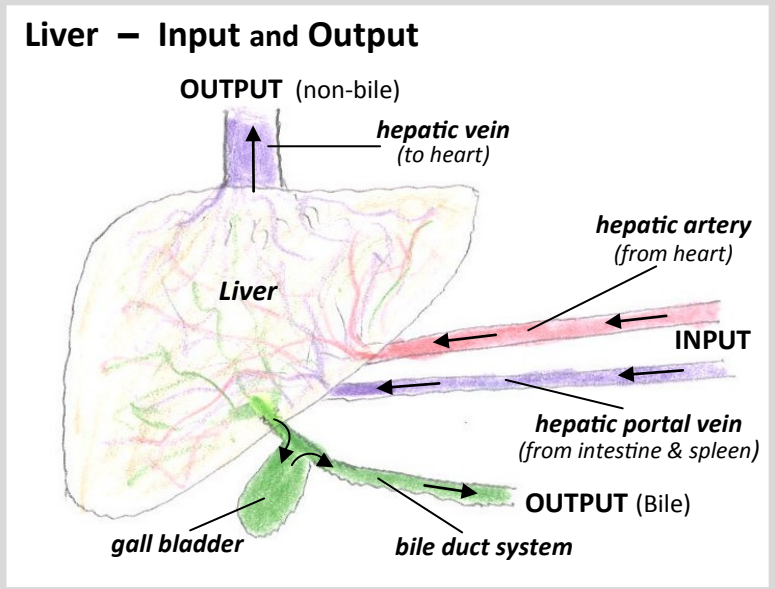


Diagram 1

The Work Units

The work units of the liver are cells called **hepatocytes** and there are many millions of them arranged in columns and sheets. The hepatocytes carry out many chemical functions:-

They make ("synthesize") lots of important substances essential for the body's normal functioning

Some of the things they synthesize include:-

a) many proteins, particularly those which are carried in blood plasma, such as **albumin** and the many proteins which enable blood to clot to prevent blood loss. Another important pair of substances made in the liver are ***transferrin*** and ***caeruloplasmin*** which are the substances which bind to **iron** and **copper** respectively, and transport them around the body;

b) important lipid ("fatty") substances, particularly cholesterol; and

c) glucose and other carbohydrates from non-carbohydrate sources such as fats and amino acids.

They neutralise toxic substances, rendering them harmless to the body.

The body often ingests substances which are toxic, and the liver cells are able to neutralise them. The most well known ingested toxin is alcohol, which the liver is able to break down into harmless substances. This mechanism can fail when too much alcohol is ingested in too short a time for the liver cells to detoxify it, and the unmodified alcohol passes into the main blood circulation and into the brain where it damages brain cells, leading to neurological malfunction (drunkenness), and if a big enough dose of alcohol gets through, death by acute alcohol poisoning may ensue.

In addition to external toxins, the liver also has to handle internal toxins. When the body's cells die, which they do in great numbers every day, some of the breakdown products of cell death are toxic. In particular, the breakdown of proteins (the main structural components of cells) releases substances which contain nitrogen, many of which can be highly toxic (e.g. ammonia) and require detoxifying by the liver cells. Most of the nitrogen-containing substances produced by cell breakdown are converted in the liver to substances called **urea** and **creatinine** which are passed via the blood circulation to the kidneys where they are excreted harmlessly in the urine.

They can hold and store substances until they are needed.

The most important stored substances are glycogen (which can be converted quickly into glucose when needed e.g. when the blood sugar level falls), vitamins A, D and B12, and iron (in the form of ferritin.)

They produce bile.

When red blood cells reach the end of their useful lifespan (about 120 days), the old cells are trapped in the spleen where they are broken down, and the breakdown products of the haemoglobin they contain are carried to the liver in the **hepatic portal vein** (see ***Diagram 1***); these products (**bilirubin)** are insoluble and have to be bonded to albumin for transport to the liver. The hepatocytes in the liver link the bilirubin to glucuronic acid, and the compound formed is soluble, forming the major component of bile**.**

Although most of the substances produced or detoxified by the hepatocytes pass back into the sinusoids for transport to the central and hepatic veins, the bile (and some of the cholesterol) pass into tiny canals **(bile canaliculi)** between adjacent hepatocytes, and from these into the bile duct system (see ***Diagram 2.***)

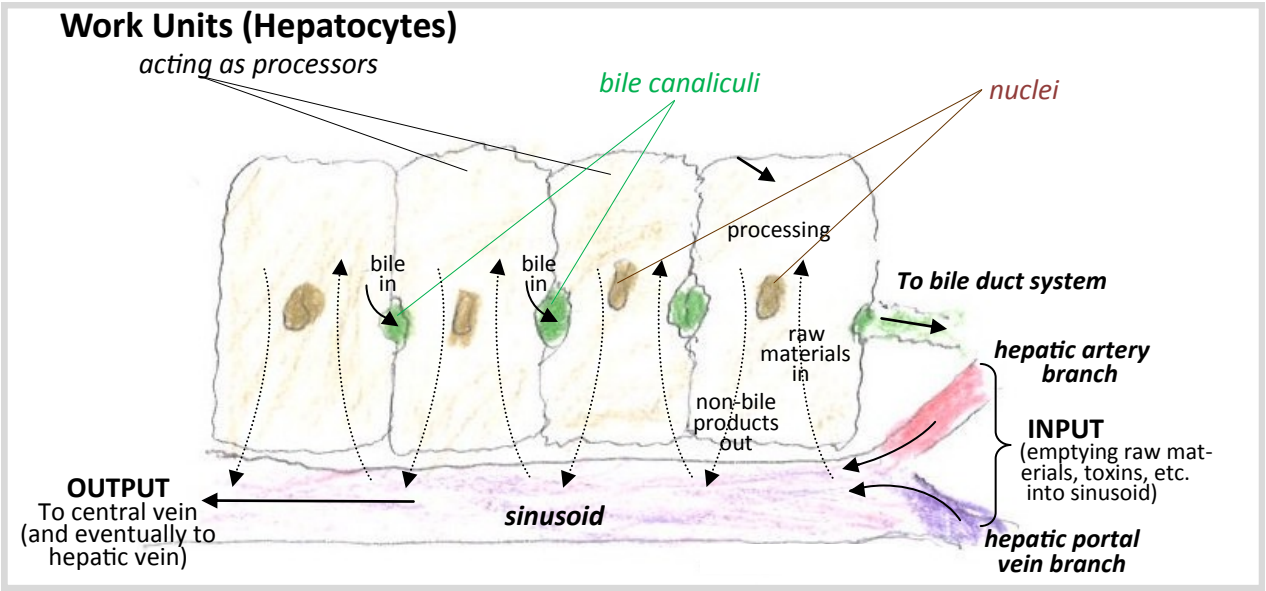


Diagram 2

Summary

- For the liver to work efficiently, input, work units and output systems all need to be working properly in a fully integrated way.

In my next *Simple Guide*, I will explain how, in **cirrhosis of the liver**, disruption of the input and output systems interferes with normal functioning of the liver, even though many of the hepatocytes are capable of functioning normally.

Olivia's Story

by Bongkeun Jeon

I would like to say a big *thank you* to the UK Support Group for inviting me to write this article for your newsletter about our daughter Olivia's Wilson's disease, how it has affected my wife and me and how it has been treated in a different part of the world in the past and here in the UK now.

Olivia

The encouragement my wife and I have received from WD patients, doctors, nurses and friends has been huge. Our English *family* in London have been supporting in many ways and giving more attention to our loving daughter Olivia. These made us so special feeling that we are not alone. We still have to battle with potential factors that might affect Olivia's future health, but we always appreciate that we have our lovely daughter, Olivia.

Let me introduce myself briefly. I am Olivia's father, Bongkeun Jeon, age 41. My wife, age 39, and I have been living in England for about 12 years and I have been working in a Further Education College as a lecturer in hairdressing for about 9 years. Olivia was born in October, 2009 in St. Mary Hospital, Paddington, and is a special child for us as we waited ten years for her to arrive. I sometimes found my job difficult dealing with sensitive teenagers and speaking English for Second language. After Olivia was born I requested a two year sabbatical leave from my college. My request was accepted and my wife and I and Olivia returned to our home country of South Korea between August 2011 and August 2013. Both of our parents live in a village called Mun-San near the border of North Korea. Mun-San is about 1-2 hours away from the capital city of Seoul.

Olivia and her family in Mun-San celebrating the Korean New Year

I regretted once just after arriving in Korea that it was too long off my UK job, but then in terms of discovering and treating my daughter's WD, it was one of the great decisions I had made in my whole life.

One evening in 2012 when we were at home in Korea, Olivia was diagnosed with croup and we took her to A&E in our village, but the doctor referred us to a bigger hospital which was a slightly far away from home. We took Olivia to the hospital called Baek Hospital on that night. We met a senior specialist the next day. The doctor commented that having croup was a common symptom to any child in their early age and recommended a couple of more treatments. The doctor gave an option to choose either staying in the hospital or coming back the next day to check her symptoms again. We decided to stay in the hospital for a couple of nights for just making sure Olivia's full and prompt recovery. We genuinely didn't think that Olivia's symptom was any serious condition and neither did the doctors in that hospital.

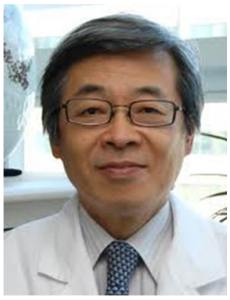
It was an unexpected policy that any inpatient had to have blood tests in that hospital. At the beginning, we were not happy to carry out some blood tests for a little girl, but we allowed it and the doctor told us that some of the results were abnormal. The senior doctor asked a lot questions like whether or not Olivia took Chinese herbal medicine recently, had had any sleeping problem or eating disorder etc. The doctor and we expected that in a few days the results would be normal, but the liver level remained the same.

Prof. Kim

The senior doctor who was in charge of children in the hospital promised that she would organise the child liver specialist to look at Olivia. We stayed in hospital a few more days for monitoring her liver level. When the liver levels dropped down a bit, we were told we could go, but we were unhappy with the fact that we were not able to see the liver specialist. My wife eventually insisted that we see the liver specialist to discuss Olivia's symptoms and in the end a Dr Kim, who was a young professor, was sent to see us. We call Dr Kim our life saver. He said that there were various reasons why the liver level might be elevated and that sometimes there was no obvious answer.

Olivia was in hospital for 10 days and my wife and I stayed with her throughout. We met Professor Kim a few more times to check Olivia's symptoms and he suggested having the DNA test and 24 hours urine test as he was suspicious about Olivia might be having WD. By then Olivia's blood results were improving, so we strongly believed that Olivia would be fine.

Later in **April 2012**, we met Dr. Kim again. He didn't say much and kept typing on the computer. He then said that from the 24 hours urine test, it resulted as normal. When we heard, we were so pleased but he said that he was waiting for the result of DNA test. He looked at us once and stared at the computer screen again. He sat down in front of us and said, "We found that Olivia has WD." We couldn't believe what he said and asked checking again. He nodded several times and quietly said, "Please not to worry. Olivia is very lucky girl being found this disease in her early age." My wife and I had a massive shock and came across with a great fear as we already researched about WD on the web. We really felt that it was the end of the world. He recommended that Olivia go to the Wilson's Disease Special Clinic of the Seoul National University Children's Hospital.



Prof. Seo

Later we found that young professor Dr. Kim was once a trainee doctor of WD Professor Dr. Seo in Seoul National University Children's Hospital. That was the reason why Dr. Kim went more deep into the investigation of Olivia's abnormal liver results. We met Professor Seo on **4th July 2012** for a first appointment. At that time, Olivia was two-and-half years old. It had been heartbreaking at the beginning, but later we gradually accepted Olivia's disease and were pleased to find it. Olivia had full DNA test and 24 urine collection and blood tests again in Seoul National University Children's Hospital and being confirmed she had WD. We found that Olivia's caeruloplasmin level was very low 3.6 and Wilsons's disease was confirmed with the detection of missense mutations.

We had seen Professor Seo for about one year. Professor Seo was a great doctor and an internationally recognised Professor for WD and wrote a lot of reports about children's WD. Dr. Seo was very caring and kind doctor for Olivia. He is recognised as the one who has the majority numbers of children WD patients in the world, so it meant that we were happy as Olivia was in a right hand. Dr. Seo told us that Olivia was one of the youngest patients among about one hundred child WD patients in his clinic.

Olivia has taken zinc (wilizin) acetate since that time. The doctor said it was very rare case that he found a patient who has WD in that young age and he can hardly find any medical report how to treat that young age patient like Olivia. We have had some difficult moments as she had mild vomiting and nausea which were particularly intense in the morning (she was advised to take a morning medication 1 hour before the breakfast) and then another small dose in the afternoon. After taking a little dose of zinc, Olivia has complained a consistent tummy ache. The doctor was great that he closely monitored her symptoms and reduce/increase her medicine after checking her blood test, 24 hours urine sample and scanning her tummy in a regular basis.

I found it hard to travel to meet the doctor every 1-3months in Korea. Olivia had to pass her 24 hours urine pack to the hospital and had to have a blood test one week before meeting the doctor, when we got the result. We have been very proud of Olivia that she was brave for the blood test and never complained or avoided taking medicine and never made a hard time not to eat certain food e.g. chocolate or nuts which Olivia loved.

My wife and I were also checked for WD through DNA, but both of us were being confirmed as normal. Olivia has been a bit less complaining of her abdominal pain but we hope that it will be better. Just before we left Korea, Olivia's liver level AST and ALT got back to normal, but her pancreas level went up slightly and she had some scans for that. The result was fine. Professor Seo mentioned that Olivia might need to take zinc for her rest of life if the only side effect of tummy ache was settled, but we still have to monitor her closely.

We joined the Korean WD patient group which was great. Whenever I asked some questions regarding WD for Olivia, there was always someone answered my questions. We had to leave South Korea in August, **2013** for returning to my job. Professor Seo recommended us to meet Dr. Darwan at King's College Hospital, London and we have met him a couple of times and waiting for another appointment soon. We found the NHS system is a bit slower compared to South Korea, but all the members of staff in the hospital are extremely kind and supportive. Dr Darwan and his medical team monitor Olivia's symptoms and tummy ache now. We worry how Olivia will cope with her disease, but we hope that she accepts her disease and fights it in a clever way through her life.

Thank you for your reading my article and thank you to the UK Group for your tremendous support and efforts for all patients and family and for improving awareness of dealing with WD. I hope all patients respond well to treatment and enjoy a normal life as like all other people around.

At Home With Dr John Walshe

by Valerie

Dr Walshe – At Home March 2014

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. Now in his ninety-fourth year and still writing papers and delivering lectures, he takes an active interest in the management of patients and in the running of the Support Group, of which he is President.

In the last two years he has told us about the importance of taking *penicillamine* and *trientine* in **divided doses** during the day around **half an hour** before **eating,** and the importance of **diet** in the management of Wilson's disease. This year I want to know how important it is for patients to be monitored by their specialists on a regular basis and what tests should a Wilson's patient expect.

Is it important for Wilson's patients to be seen regularly?

Yes. Patients need to be checked regularly to make sure that they are getting enough treatment and that their treatment is not causing any side effects.

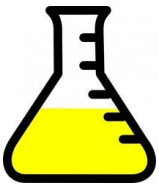
So what tests should patients expect?

Two important tests for patients are measuring the serum copper in their blood and doing a 24 hour urine collection for copper estimation.

Why test the patient's serum copper levels and what results would the doctor be looking for?

By testing the patient's serum copper levels, the doctor would be checking that the patient is getting enough treatment. The serum copper should be stable – about the same level on every visit.

And why do a 24 hour urine collection? What is the purpose of that?



By testing the patient's urine over 24 hours, the doctor is making sure that the urine copper is normal. In a well treated patient the excreted urine copper should be less than 0.5 µmol (micromoles per litre.)

What is a micromole when it is *At Home*?

1 micromole = $\mathbf{10^6}$ of a mole = **1/1,000,000** of a mole = **1/1,000,000** of $\mathbf{6.022 \times 10^{23}}$ molecules or atoms...*I knew I shouldn't have asked...*

How does the doctor check for side effects of the drug treatment?

If a patient has no complaints, a simple screen for side effects is the erythrocyte sedimentation rate (ESR.) This is normally only a few mm/hour. If it is raised, it means that there is an inflammatory process which needs further investigation. Another screen for side effects is to look for protein in the urine. If there is protein in the urine, then more kidney function tests should be done.

So what other tests might patients expect?

Other tests might include Liver Function Tests (LFTs) including alpha feto-protein levels. It is also wise to do an ultrasound scan of the liver at regular intervals to look for liver damage and the early signs of cancer of the liver, a rare complication of Wilson's disease. For patients with neurological signs, a CT or MRI brain scan might be requested.

Priya's Story

by Priya Joshi

Hi. I am Priya and I am a member of the Verein Morbus Wilson Schweiz, the Swiss Wilson's Disease Support Group. I met Valerie in May last year in Heidelberg, where the German Support Group had their meeting. Afterwards, I joined the WDSG -UK facebook site, which I enjoy following when I have time. Valerie has asked me to write about my experience of Wilson's disease to remind patients of the importance of taking their medication properly.

Priya in Heidelberg in May 2013

I was born in Zurich in April **1982** and had a perfectly normal childhood. When I was eleven years old I went to see my mother's ophthalmologist. While doing my eye examination, the ophthalmologist found what he described as *golden brown rings* around my irises. He advised my mother to take me to see a paediatrician straight away. I attended the local Children's Hospital and after undergoing detailed tests, I was given the diagnosis of Wilson's disease. Nobody in the family had ever heard of it before. I was prescribed penicillamine and was told that I should avoid foods with a high copper content, like chocolate for example. That was going to be difficult. After all, I am Swiss and I love chocolate!

The rest of my adolescence was unremarkable. I was in good health and continued to be seen regularly by my physician. However, in **2000**, shortly before my 18th birthday, I was told that I needed to change doctors as I was considered too old to continue being seen at the Children's Hospital. My medication was changed to zinc and I was referred to a gastro-enterologist, who supported the change of my drug therapy.

Between **2002** and **2005** I was in Hamburg, Germany, studying at the Stage School of Music, Dance and Drama. It was a great but hard time for me as the training was demanding and student life was fairly hectic. The new drugs had begun to make me feel nauseous and I became less attentive in remembering either to take them or to take them at the appropriate time in relation to my eating.

Unknown to me, my copper levels had started to rise. In **2005** I returned to Zurich to begin a degree in media and communication science. I was studying hard, working to supplement my income and I even managed to find the time to appear in a musical. I didn't recognise that my extreme fatigue, weight loss, trembling and deteriorating handwriting were anything other than physical exhaustion. I was perceived by those around me as being drunk and indeed that was exactly how I felt in my head. By **2008** I was having difficulty walking and at this point my mother insisted on taking me to the University Hospital, where the doctors immediately recognised that my medication was not working as it should and that the build up of copper in my brain had now caused me neurological damage. Because I was diagnosed with Wilson's disease virtually pre-symptomatically, I was unaware of the symptoms of neurological Wilson's and therefore didn't recognise them in myself. My drugs were immediately changed to trientine and I'm pleased to say that they are now keeping my copper levels in check.

Unfortunately, it all came too late. The excess copper had left me with ataxia, slurred speech, a stiffened gait and muscle cramps. I had lost my independence and during the most difficult time of my life I was even having problems taking a shower alone. However, I was determined to do all that I could to minimise the effects and embarked on a strenuous course of physiotherapy, speech therapy and muscle training, which I continue today. I have had to fight hard for every improvement that I make, but I will not give up.

Today, my muscles are still cramped and I have good days and bad days. Everything takes me so much longer to do. However, I am living in my own apartment and am studying for a master's degree. I have just completed my thesis and am now preparing for my final exams. I have a homecare worker one morning a week and have a student job the same afternoon. I miss my independence more than anything else, but I count myself lucky to have the support of such a loving family and wonderful friends. I am determined never to give up and my current ambition is to be able to walk downstairs again unaided. I know that I can do this.

And my advice to any patients reading this article is to be sure to take your tablets regularly, be seen regularly by your hospital consultant and if you experience problems with any of your medications, do seek medical advice immediately. Thank you...

Genetics of Wilson Disease – an update by Prof. Oliver Bandmann

Oliver Bandmann

I'm sure many of you know a lot about the genetics of Wilson's disease (WD) already. Let me just remind you of two key aspects here:

- WD is inherited in so-called autosomal recessive fashion: We all have two copies of a relevant gene (one inherited from mum and one inherited from dad.) If a disease is recessively inherited, then this means that an individual has ended up with two damaged copies of a particular gene.
- The WD gene is called ATP7B; it is involved in the regulation of copper excretion and is located on chromosome 13.

At last year's meeting of WDSG-UK, I had the pleasure of talking about a recent large study on genetic aspects of WD which we completed in early 2013.***** Our study addressed two questions:

1. How useful is analysis of the WD gene ATP7B for the diagnosis of WD?

   Short answer – extremely useful. Modern techniques allow us to nail the diagnosis genetically in 98% of all WD patients. This does NOT mean that genetic testing should replace other, more conventional blood tests such as the measurement of caeruloplasmin or copper which can be done much more quickly and are also cheaper. However, our results confirm that genetic testing can be extremely helpful in those patients where the biochemical investigations are difficult to interpret. It's important to understand the limitations of genetics as well – the precise gene defects (or "mutations") will only ever tell you whether you've got WD or not. They will not tell you which symptoms you are likely to develop in the future, whether you will respond well to medication or not or anything else. Genetic testing is getting cheaper and cheaper – it is quite likely that more and more patients with definite or possible WD will be tested genetically in the future.

2. How common are ATP7B mutations in the UK?

   Old studies dating back to the early eighties suggested that there is one WD patient for every **30,000** people and that there is one ATP7B mutation carrier for every **90** people in the general population. However, these figures were at least partially based on guesses and predated the discovery of ATP7B as the WD gene. We therefore decided to analyse the entire ATP7B gene in 1000 controls to get a better idea of how common ATP7B mutations really are. Which controls did we choose? We used a little bit of dried blood spots from new-born babies. How did we do this? All new-born babies have a tiny little bit of blood taken so that certain illnesses can be detected and treated as early as possible.

   The results of our study actually suggest that WD may be as common as **1:7000** – much higher than the previous figure of **1:30,000,** and the incidence of carriers of about 1:40 rather than previous estimates of 1:90. However, it's really important to point out two limitations of our study:

   - It is likely that there is so-called reduced penetrance for at least some ATP7B mutations. This means that not everybody who carries two ATP7B mutations may develop WD.
   - It cost us a lot of money and took us several years of hard work to analyse ATP7B in 1000 controls. Nobody has ever attempted this before. However, it's still important to point out that 1000 controls is a comparatively small sample size when it comes to working out how common disease mutations such as ATP7B are in the general population. Just like any other study, our work needs to be repeated by others, ideally in many more controls still.

***** '**A Genetic Study of Wilson's disease in the United Kingdom'** *Brain*, 2013 May; 136(Pt 5): 1476-87. doi: 10.1093/brain/awt035. Epub 2013 Mar 21. Coffey AJ, Durkie M, Hague S, McLay K, Emmerson J, Lo C, Klaffke S, Joyce CJ, Dhawan A, Hadzic N, Mieli-Vergani G, Kirk R, Elizabeth Allen K, Nicholl D, Wong S, Griffiths W, Smithson S, Giffin N, Taha A, Connolly S, Gillett GT, Tanner S, Bonham J, Sharrack B, Palotie A, Rattray M, Dalton A, Bandmann O.

Genetic Sequencing and Wilson's Disease

by Mary Fortune

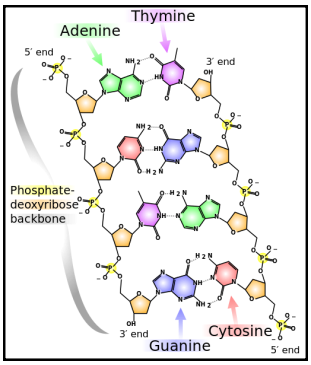
I am a PhD student in the Cambridge Institute for Medical Research, working on the development of statistical techniques to uncover information in genetic datasets, with a focus on Type 1 Diabetes and other autoimmune conditions. I have been asked to write about the techniques which are employed for testing people's genetics for the presence of Wilson's disease mutations. I have Wilsons disease myself, and am a member of the WDSG-UK Committee.

Mary

In the UK, genetic testing for Wilson's Disease is done in Sheffield, where Prof. Bandmann is based, using the Sanger Sequencing method on an ABI 3730 machine. The rest of this article will concentrate on this technique, although many alternatives exist. If you are interested in learning more, I would particularly recommend <www.yourgenome.org> as a good introduction to the basics.

Most DNA molecules in the human body take the form of a double stranded helix, with phosphate backbones being held together by bases attached in pairs. There are four possible bases; adenine (A), cytosine (C), guanine (G) and thymine (T), and an error in even one of these at the wrong position can be the cause of a genetic disease. These DNA molecules can be very large; the human genome contains approximately three billion base-pairs, and even ATP7B, the gene whose dysfunction is responsible for Wilsons Disease, is over 100,000 base pairs long. This creates difficulties for sequencing, since in practice we can only sequence short segments. Hence, we first cut the segment of DNA we are interested in into short overlapping sections, using special enzymes. We will sequence each segment individually, and at the end use mathematical techniques to piece them all together like a giant jigsaw puzzle.

Even though we might start the sequencing process with only a single copy of the DNA segment we are interested in, we will need many more in order to infer the sequence of all the bases. Hence, our first step is to use a technique known as polymerase chain reaction (PCR) to generate thousands of copies. This uses repeated cycles of heating and cooling, along with the same enzymes used to replicate DNA inside a human cell, in order to double the number of copies present at each cycle. Great care has to be taken not to contaminate the sample at this step, since even a small contaminant will also be amplified by this process.



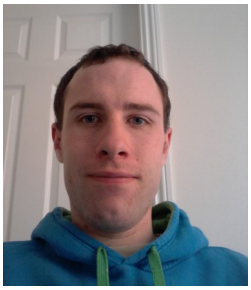
DNA Chemical Structure

Next, we proceed to the true sequencing stage. First we need to separate the two strands of DNA. A primer (that is, a strand which serves as a starting point for DNA synthesis) binds to this template, and then DNA bases bind to the primer, to begin the process of synthesis. Free bases are present, and these attach to the strand. However, in addition to the presence of standard bases in the reaction, we also add special chain terminating bases. At some random point in the process, one of these will be incorporated in the place of a standard base, terminating the strand. The chain terminating bases are also fluorescently labelled, with a different colour for each of the four bases, and so their presence and nature can be detected by a laser. Since this process occurs on so many copies of the sequence, chain terminating bases should be present at any given position on at least one copy, and hence strands of all possible lengths should exist. Capillary electrophoresis is used to separate these strands out by size; longer fragments will take longer to pass through the capillary. Hence we read off the final sequence by the presence of the fluorescent markers; for instance, the first colour to be detected will correspond to the first base present, since the fragment with a single base is shortest.

Once we have the sequence for the whole of the ATP7B gene, then we can search through it for mutations known to cause Wilsons Disease. If we already know where in the gene to look (for example, if the mutations possessed by relatives are known) then this process can be made much more efficient, since we can focus on sequencing only the region of the gene required. This process is very expensive - the machines can cost hundreds of thousands of pounds, and even the reagents used in the reactions are costly. Hence, in the UK, it is common for labs to first run a small test looking at the regions where mutations are most commonly found, and to reanalyse using a broader test only if nothing is found in these.

Matthew's Story

by Matthew Tucker



Matthew

Hello everyone; my name is Matthew and I am 25 years old and come from Knowle in the West Midlands. This is the story of my experience of Wilson's disease. It started when I was 9 years old. I had just come back from my GP's with a prescription for an antibiotic for an infection that I had been suffering from. Once I started taking the antibiotic I broke out in a very itchy rash that began on my neck and over the course of the next few days spread across my whole body. My joints seized up and rendered me immobile and the rash went from itchy to incredibly painful. My parents took me to our local hospital in Solihull, and after a careful examination the doctors diagnosed glandular fever and said to keep taking the antibiotics.

I got no better so after a few days I returned to our local hospital where the diagnosis was changed to scarlet fever. Still I took the antibiotics. Finally, things started to improve to the point where the joint pain went away but the rash remained, though it was not painful anymore. My parents were still concerned and through my dad's work's private health cover they arranged for me to be seen at a private hospital. After several consultations with a paediatric doctor there, as well as a lot of tests, the diagnoses of scarlet fever and glandular fever were discounted. However, the tests did flag up some irregular findings and the private hospital referred me to a paediatric liver specialist at Birmingham Children's Hospital.

Not only did I have to have a lot of blood tests taken again, but I had to do urine samples and have ultrasound scans as well. When my parents explained the situation to my school one of my teachers, who was an ex-nurse, said she thought it might be Wilson's disease. Consequently, we suggested it to my liver specialist who said it was an extremely rare illness and was unlikely. However, she agreed to test me for it and after about a week we went to see her and she confirmed it was Wilson's disease. I was immediately booked into hospital and was asked to do a 24 hour urine collection, which confirmed the diagnosis of Wilson's disease, and I was started on penicillamine. Two weeks later I went into hospital for about a week for a liver biopsy, and a K-F test which proved negative.

About a month later my liver specialist said she wanted me to see a psychiatrist as I was experiencing high levels of anxiety and restlessness. He diagnosed Tourette's syndrome. At the same time he also diagnosed me with anxiety and started me on Prozac, while my liver specialist put me on Vitamin B6. If that wasn't enough, by the age of 10 my parents noticed that kids that I used to be taller than had overtaken me in height and weight quite considerably and I was not thriving at school or in social situations. So the next referral was to an endocrinologist who x-rayed my hands and confirmed the bones that should have grown had not. He then ordered an insulin stress test which still ranks as one of the worst tests I have ever had! He diagnosed me with Growth Hormone Insufficiency and put me on daily injections of growth hormone which I had to administer myself in my legs. As my muscles developed I had to start injecting it in other parts of the body such as my stomach. However, I was told that Wilson's disease did not account for the growth hormone insufficiency though no other explanation was even given and I remained on hormone injections until I was 18. Thereafter I was transferred to adult services. The new doctors weren't clear what the impact of copper had been on my health, (small pockets of copper had been detected in my brain on an MRI scan of my head), but my neuro-psychiatrist stated that the earlier diagnosis of Tourette's syndrome was wrong and he therefore quashed the diagnosis.

At around the age of 22 I noticed my knees started to click when I went up and down the stairs so my GP sent me for X-rays, which turned out to be inconclusive. Next I had an MRI scan which showed that the cartilage in my knees had changed and I was referred to an orthopaedic specialist. He said as a result of Wilson's the cartilage had hardened and the fluid in my knees had become very thin instead of being thick. Because of this it was causing the cartilage in my knee joints to rub and as a result my knees can be painful, restricting for how long I can drive and play fast contact sports. Currently I am also having problems in my hands and lower back, but my specialists have not been able to confirm whether they are related to the Wilson's.

Recently I have been to one of the first Wilson's disease clinics at the Queen Elizabeth Hospital, Birmingham *(see p19)*, where I saw on the same day my liver specialist, neurologist and a neuro-ophthalmologist. As a result of this meeting I have been referred to a rheumatologist, and to an ophthalmologist for advanced eye examinations.

That's everything: thank you for reading my story.

Members' News

Allie and **Rita Johnston** from Edinburgh would like to bring you up to date with their news. Allie says,

"We have just returned from a very pleasant short break in Venice, flying there from Edinburgh via Amsterdam and arriving on a speed boat with the waves crashing against the side. After checking into our hotel and before finding somewhere to lunch, we took a gondola trip along the ancient Venetian canals. A very kind Japanese lady captured the moment as we were being serenaded by the gondolier.

Our next holiday is scheduled for June, when we plan to sail up the Aegean coast on a Turkish gulet. More news of that next year. For those of you like mum and I who enjoy shopping for bargains, you might be interested to know that we recently met Tim Wonnacott of *Bargain Hunt* fame at the Antiques & Collectors Fair in Edinburgh.

Finally, I have found two local outlets through which to sell my handicrafts and greetings cards. I am delighted to say that I have now raised a further **£100** for Group funds."

Allie and her mum, Rita, in Venice

* * * * *

Ashok Pandit, a Wilson's patient from Nepal, who featured in our newsletter in **2012** and is a regular contributor to our fb page, has just returned from a recent adventure with his cousins. He says,

"Recently I went to my maternal uncle's home which is 30 kms from where I live in Kathmandu to stay with him and my two cousins, Asmita aged twenty-four and her brother Amir, aged twenty-two. When I arrived, Asmita asked me if I would like to go to Sundarimai temple the next day and I smiled and said her ok without thinking. Early next morning she came to wake Amir and me up, but Amir didn't want to go. I insisted him so much and then convinced him to go with us. We were all ready to set off to temple, then Asmita told me to wait for uncle. I didn't know he was going with us too.

So four of us started our journey. Well, temple is in hilly terrain in middle of Sundarijal forest. We need to go uphill first and then downhill through the trees and eventually arrived at temple, where we offered our worship. There was a monkey, so we gave it grapes. While we were returning, uncle said, "Let's go to Mahendrashor Mahadev's temple too." Asmita and I were excited because we have never heard of it. We set off but unfortunately my uncle forgot the way to it. We were lost in middle of forest at peak of mountain and we don't know what to do. I then said let us now go back down the hill but my uncle didn't want to return the way we have went in, because we still wanted to reach the temple of Mahadev.

We went back down a short way holding on to every tree, because there were all leaves that has made the way slippery. It was really fun catching every tree. Then, at middle of mountain, suddenly there was a path and uncle remembered the way to temple. We found it and offered our worship and then returned home. Well guys it was an unplanned adventure that I enjoyed a lot."

Ashok

Sundarijal Forest

Sundarimai Temple

Our committee member **Anne-Marie Le Cheminant** and her husband Steve would like to share their Spanish holiday adventures from last year with you. Anne-Marie says:

"When planning a fly drive holiday to Spain last year I persuaded my husband, Steve, to consider visiting a little known region to the south-west of Madrid which hugs the border with Portugal called Extremadura. It was an area I had wanted to discover for years. I knew that there were important Roman remains there and it was a place from which many of the *conquistadores* of the 15th and 16th centuries had come, so had lots of history. Trying to do some research we found little information in guide books except for Gisela Radant Wood's book *Walking in Extremadura.* Its subtitle, *Discover Spain's Secret Paradise* was most enticing and the book proved invaluable.

She describes the sheer space of the region as 'exciting, exhilarating and exquisite.' We were not disappointed. At more than 41,600 square kilometres Extremadura is bigger than Switzerland and is sparsely populated. It has impressive forested mountain ranges such as the Sierra de Gredos which rises to over 2,500 metres and the Sierra de Gata. There is an important wine growing area which produces some excellent wines although unfortunately few are exported. There are also many delightful villages to discover.

On our travels we enjoyed stress-free driving on the open road. Extremadura's network of roads is impressive and a dream, as there is so little traffic**.** Another highlight was the food. The *jamón ibérico,* ham of Extremadura is one of the best in the world and has won all the top awards. It is known as Pata Negra in Spain. The black pigs roam freely and feed on acorns and herbs which give the ham the most wonderful flavour. Steve commented that he had the best pork scratchings ever there!

Apart from the very beautiful and varied countryside we loved some of the cities we visited, Cáceres and Trujillo were a revelation. They both have stunning historic centres which display, in the shape of the most marvellous fortified houses and palaces, the enormous wealth enjoyed in the Middle Ages by the Church and noblemen benefiting from the riches brought back from the Americas by the conquistadors. There were also the vestiges of a previous time when the Christians fought for control of the country over the Moors.

Mérida reveals Extremadura's Roman history when it was capital of Lusitania province. The city with its Roman Bridge over the River Guadiana was built to last. Among the amazing remains are the Roman Theatre, the Amphitheatre, the Forum and the towering Aqueduct. The recently opened Roman Museum, too, was spectacular, the building itself and the displays inside.

Finally I must also mention the Montfragüe National Park. It is world famous for birds and is home to more birds of prey than anywhere else in Europe. Hundreds of birdwatchers visit the park in spring. We were there in September when it was very quiet, but we did see large numbers of the different kinds of vulture enjoying the thermals. A fantastic sight against the vivid blue sky.

We used *Booking.com* to organise most of our accommodation and stayed in a variety of places including a family run guest house, a converted clothing factory, a converted monastery and a former medieval palace. The weather didn't fail us either. The sun shone the whole time we were there. All in all a holiday to remember!"

Steve and Anne-Marie in the Sierra de Gredos

Red roofs of a typical Extremaduran village

Black Iberian pigs

Trujillo: statue of Pizarro who conquered Peru

Roman Theatre in Mérida

Inside the Parador in Plasencia: a 15th century monastery, now a 4 star hotel

WDSG-UK 2014-15 EVENTS

Date	Time	Event
May 17	**0900 - 1830**	Morbus Wilson e.V. Annual Symposium - University of **Heidelberg,** Germany
July 13	**1100 - 1530**	**WDSG-UK Meeting and 4th AGM** – Cambridge Rugby Union Football Club Grantchester Road **Cambridge** CB3 9ED.
August 23	**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 of the death of one of our younger patients and members of the Group, Victoria Weaver from Devon, who died on 25 April 2013 aged 25 after fighting a long and hard battle with Wilson's disease. Our deepest sympathy and warmest thoughts go out to her family. We were pleased to hear that Sylvia Penny and her granddaughter Emma were able to attend the funeral.

* * * * *

We send our condolences to Emma Collcott and her family for the sad loss of her grandmother, Elizabeth Galloway, who passed away on 23 December 2013 less than a year after her husband, Charles, died. This must be a particularly difficult time for the family and we would like to thank them for holding a collection at the funeral for WDSG-UK, which raised the very welcome sum of £490.

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 Co-Founder

WDSG-UK

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