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

# NEWSLETTER

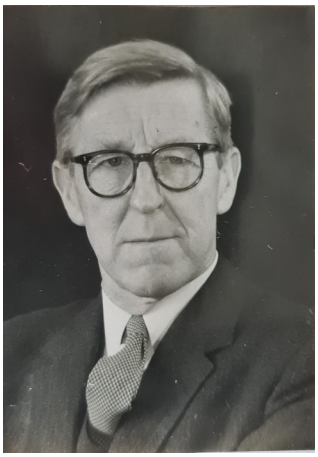
#wdsguk APRIL 2023

It is with great regret and sadness that we report the death of our President, the inimitable Dr John Walshe, who at the age of 102 died peacefully at his home in Hemingford Grey after a short illness.

Many of you will have known him personally, having been patients of his in the past. Others will have met him at one of our annual meetings in Nottingham/Cambridge over the past 20 years and/or read articles about him in our newsletters every year. But if you are a patient with a Wilson's disease diagnosis, wherever you are in the world, then you are almost certain to have benefited at some time or other from his treatment discoveries in the past.

Prior to 1955 there was no effective long-term therapy for treating Wilson's disease. Patients would have had little or no hope of recovery. Dr Walshe changed all that. In 1955, while taking up a Fulbright Scholarship in Boston, USA, he had a lightbulb moment! It was there that he joined up the dots! He had noted a few years earlier that one of the breakdown products found in patients taking penicillin was a molecule known as penicillamine and his lightbulb moment came when he realised that penicillamine had exactly the right chemical formula to combine with copper. Without further ado, he managed to obtain 2 grams of it from his American colleagues, together with a patient with a recent diagnosis of Wilson's disease on whom he could test it.

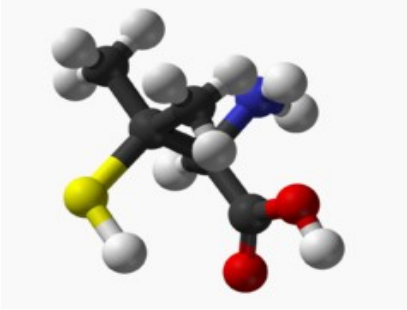
Fortunately for us, there were no such things as 'clinical trials' in those days or it might have been a different story. Not wishing to kill the patient off, he returned to his accommodation and took one gram of it himself (equivalent to 4 tablets today). He was delighted when he woke up the following morning feeling fit and well. When the patient also later survived his first dose, and more importantly excreted large quantities of copper in his urine, Dr Walshe was overjoyed!



**Dr John M Walshe**
**24.04.1920—14.10.2022**

He returned to the UK later that year with a further 10 grams and, through the contacts of his father Sir Francis Walshe – an eminent neurologist at the National Hospital for Neurology & Neurosurgery (which still has a WD clinic today) 3 patients were found for him to test it on. One was a 16 year old newly diagnosed patient called Shirley. Bedbound in a London hospital, shaking uncontrollably, unable to walk, talk, eat or dress herself, she slowly started to recover. Shirley went on to lead a normal life and is still alive today at the age of 83.

The following year, Dr Walshe took up a Cambridge University post in the Department of Investigative Medicine and an honorary consultant's position at Addenbrooke's Hospital (then located on Trumpington Street) and it is here that many of us had the good fortune of meeting him and becoming his patient.



**Penicillamine:**
*Courtesy of Wikimedia Commons*

During his time at Addenbrooke's he developed two further chelating therapies, the first in the late 1960s (in collaboration with Dr Hal Dixon) called trientine dihydrochloride, and the second in the 1980s called ammonium tetrathiomolybdate (a drug being trialled for commercial use today). By the time he retired from Addenbrooke's in 1987, Dr Walshe had a patient base of over 320 and was recognised as the world's leading authority on Wilson's disease.

However, not content with a life of leisure nor pursuing his interests in Gothic architecture and mediaeval stained glass, he took on a monthly clinic at the Middlesex Hospital where, until the year 2000 when he turned 80, he continued to treat many of his former patients and even some new ones. He then handed over to Dr Gillett who has run the clinic ever since.

At this same time Caroline Simms and Linda Hart were setting up WDSG-UK and Dr Walshe was a great enthusiast. He wrote to all his former patients inviting them to join. He headed the Group as President and worked closely with it right up to his death, making himself available to patients and their families whenever they wanted to speak to him.

Widowed in 2011 after a happy 55 year marriage to Ann, he continued to write medical papers and give lectures until well into his 90s. He remained in the family home until the very end, with his daughter Susan and her partner Phil caring for him well. He leaves another daughter Clare and three grandsons Simon, Oliver and Ben and a much cherished greatgrandchild, little Annabelle.

As an outstanding and compassionate doctor, an exemplar to all in his profession, he also leaves many grieving patients with whom he became friends. We thank him for his lifetime's devotion to us: we will miss him sorely and be indebted to him for ever.

*Valerie*

# Chairman's Report for 2022-23

For those of us fortunate enough to come through Covid without suffering personal loss it is already hard to recall just how restrictive life in 2020 to 2022 had been as we return to a more normal life. The NHS however, has not come through unscathed. All measurable parameters suggest a service that is in crisis, without a single sighting of the cavalry coming over the hill.

Covid has driven change. In particular, face to face appointments are no longer considered essential in routine follow up care, either in primary or in secondary care. This has distressed some patients in the Group, but the majority say they prefer telephone clinics, reflecting the fact that attendance at specialist clinics in Wilson's disease often necessitates them travelling long distances from home.

At our recent virtual Coffee Morning meeting to mark Rare Disease Day 2023, patients highlighted other concerns. Many patients have never had a liver scan, even some of those where the liver is and has always been, the focus of concern; in similar fashion, many patients have never had an MRI scan of the brain, including some of those with neurological disease. A very small number of those with Wilson's disease have talked about developing cancer in the liver, which is still believed to be a rare association. But the most concerning stories I have been aware of in the past year relate to patients presenting with psychiatric disease as the first manifestation of Wilson's disease. This particular group is not unusual in suffering a delayed diagnosis (for most patients it can take 2 years for the penny to drop) but it appears that ignorance of this mode of presentation is a further complicating factor.

National UK guidelines were published in 2022 that deal with each of these issues (and many more besides) and it is the responsibility of everyone of us to ensure that these are available to anyone who might encounter Wilson's disease, a very long list. The BASL SIG, in which doctors across all relevant specialities meet annually to discuss Wilson's disease, has proved an excellent forum for teaching - even the experts are learning from each other. In addition, through the SIG, Prof Aftab Ala has a current PhD student whose studies will include some of the psychiatric aspects of Wilson's disease and important work on public health continues with Dr Mary Bythell.

Activities of the Support Group are all described later in this issue, but I'd like to highlight the online meetings run so smoothly by Liz Wood. It is clear that there are many patients who are much more comfortable asking questions from their own home, while many will not be able to attend the face to face meetings because of ill health or the distance involved. Many thanks to all of those who have taken the time to join in and to raise important issues for discussion. I would also like to add my thanks to all of you who have generously contributed patient stories to the newsletter in the past which serve as a good educational tool and in particular this year to Flóra, Denise, Joan and Guy.

This year we lost both of our centenarians. Dr John Walshe's contribution to Wilson's disease is unparalleled both in the clinic and in the laboratory (I met him when he was 94 still writing academic papers). As the president of WDSG-UK he will be greatly missed. James Kinnier Wilson was the son of Samuel Kinnier Wilson who first described Wilson's disease back in 1912. Although James pursued a career outside medicine he was a loyal friend to this Group. Both are remembered with enormous affection and respect and it was fitting that representatives from the medical world and patients from this Group were able to attend their funerals on behalf of us all.

I'd like to pay tribute to another who lost his life suddenly this year: Dr Bryn Jackson worked at Orphalan and we had planned to develop a film with him as a teaching aid for patients and physicians. I relished his can-do attitude and positivity. I hope that someone will push on with his ideas.

It is time for me to step down as chair of WDSG-UK at the AGM in the summer. It has been an honour to be involved with the Group. I'd like to thank the committee Mary Fortune, Liz Wood, Debbie Buckles and Valerie Wheater for their contributions, support and especially tolerance. I hope they will forgive me if I pick out Val for particular thanks. We met more than 30 years ago; we have been verbal sparring partners for much of that time but always friends and she remains this group's greatest asset.

*IN THIS ISSUE ...*

I am not sure if I have a hard exterior (that's for others to decide), but I have always been a little soft on the inside and hope you also enjoy the romantic story at the bottom of page 9. I may be showing my age, but who needs Julia Roberts?

**Graeme Alexander**
**April 2023**

# Notices

### Donations and Fundraising

We rely exclusively on our members and their families and friends for our income. We should therefore like to thank all of you who made generous donations with your subscriptions this past year and those of you who have set up regular standing orders to us. A special mention is made again to **Giuseppe Cardone** who, together with a work colleague, raised £700 through Google non-profits. We thank **Sue Boysons** and **Dympna Sands** for nominating us to be beneficiaries of monies made in memory of their loved ones. We also thank Dr Walshe's daughters **Susan** and **Clare** for sanctioning a memorial page through *InvestMyCommunity* to collect monies for WDSG-UK in their late father's memory. Thanks to all who have contributed to it so far. The page will remain open until the end of April Dr John Michael Walshe 1920-2022 by Valerie Wheater (investmycommunity.com) for anybody still wishing to leave a tribute. Dr Walshe kindly left us a small legacy in his Will which we should like to formally acknowledge here with our thanks.

Our fundraisers this year are **Maria Selwyn, AFC Rushden & Diamonds Youth Section** and **Phil Angus** & **Kurt McGuinness** who brought in a combined income of **£3,300** through their fundraising pursuits (see ***pp6-7***). We thank them for this. Should you wish to hold your own fundraising event, then please remember that our online fundraising platform *InvestMyCommunity* is an easy and efficient way of collecting sponsorship. Their fees are minimal and where appropriate they collect Gift Aid on our behalf.

Of course, all donations made to WDSG-UK can be Gift Aided, if appropriate. Gift Aid forms can be downloaded from our website or obtained from Valerie direct.

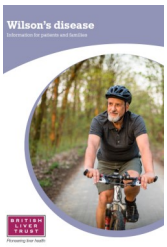
### HSBC Bank Charges for Charity Accounts

Now that our bank HSBC has introduced bank charges on our account for over the counter transactions, we thank those of you who responded to our request to make payments to us electronically in the future. If you are able to pay your annual membership by bank transfer or even better by setting up a standing order, that would be much appreciated.

### BASL WDSIG (British Association for the Study of the Liver Wilson's Disease Special Interest Group)

The 7th BASL WDSIG meeting hosted by lead Dr Bill Griffiths, took place via Zoom on 25 November 2022, a report on which can be found on ***p14.***

### The British Liver Trust




Don't forget that the British Liver Trust, which is an umbrella group for all liver disease patients in the UK, has recently produced an excellent booklet on Wilson's disease which can be downloaded from their website https://britishlivertrust.org.uk/wp-content/uploads/WilsonsDiseasewebDEC21.pdf or alternatively a single hard copy can be ordered free by emailing info@britishlivertrust.org.uk.

### NHS BT (Blood & Transplant) Organ Donation & Transplantation Directorate (ODT) and the Liver Patients' Transplant Consortium (LPTC)

VW attended the 13th NHSBT meeting via Zoom with the Liver Patients' Transplant Consortium on 31st October '22 where NHSBT reported that they were developing a new strategy (2021-2030) with the Minister of Health to increase organ utilisation. The aim is to ensure equity of utilisation across all liver transplant centres in the UK, allowing more collaboration between centres. NHSBT also wants to develop a plan around live liver transplants but this is a complex area. An overview of liver transplantation in each region of the UK for 2021-22 was presented in slide form and these infographics are available on the NHS website Organ specific reports—ODT Clinical—NHS Blood and Transplant.

### UK Liver Alliance (UKLA) & UK Liver Patients Alliance

In 2021 the UKLA was set up to bring together the leading players in liver care with the aim of influencing policy and improving liver services and care across the UK. It is made up of the Chief Medical Officers of the 4 devolved nations, together with representatives from BASL (the British Association of the Study of the Liver), BSG (British Society of Gastroenterology), the Royal College of General Practice, the British Liver Nursing Association, the Liver Advisory Group, the British Liver Trust, the Children's Liver Disease Foundation and the newly formed UK Liver Patients Alliance of which WDSG-UK and other Liver Patient Groups are members. As a direct result of UKLA lobbying the Department of Health & Social Care (DHSC) over including liver disease in departmental policy and into its *Major Conditions Strategy*, it has been agreed that FibroScan services will be introduced by NHS England into 100 Community Diagnostic Centres by March 2025 to aid early diagnosis of liver disease. This has the potential of picking up patients with Wilson's disease early.

# A Date for your Diary 2023-24

| Date | Time | Event |
|---|---|---|
| **Sunday, 23 July 2023** | **1130—1530** | **WDSG-UK 13th AGM – Cambridge RUFC, 52 Grantchester Rd, Cambridge CB3 9ED** |

### Cambridge Rare Disease Network (CRDN)

CRDN was set up in 2015 to address the challenges faced by people affected by rare diseases and to find ways of improving lives and bringing hope to those affected by rare conditions. To this end patient groups, scientists, researchers, clinicians, pharma and biotech companies locally and from around the world are brought together. WDSG-UK attended CRDN's *6th RAREfest Summit* on 26th November 2022. It was a lively event which included talks, short films, panel discussions and exhibitions.

### EASL (The European Association of the Study of the Liver)

EASL is joining forces with ERN (The European References Network—Rare Liver Diseases) to produce their own European guidance document on the "Management of Wilson's disease." As a patient representative, Valerie has been invited to give feedback on any proposed guidelines.

### ELPA (European Liver Patients Association)

Based in Belgium, ELPA is made up of liver patient representatives across Europe. Its aim is to improve awareness of all liver diseases and to ensure that treatment and care is of the highest standard. It works closely with EASL.

### Wilson's Disease Policy Network (WDPN)

Sponsored and initiated by the Pharma Company, Alexion, the WDPN was formally established in 2022 by a Health Policy Consultancy (HPP) working with a group of leading stakeholders within Europe including WD specialists. Its aim is to drive and support changes in policy that will benefit people living with Wilson's disease and their families. It has already produced a WD Policy brief which they, through Prof. Dr Karl Heinz Weiss WD specialist in Germany, launched in collaboration with ELPA to EU policy makers in Brussels on Rare Disease Day 2023.

### Aarhus WD Symposium—Denmark— 5-8 May 2022

Prof. Peter Ott and Thomas Sandahl hosted their 2nd WD Symposium in Aarhus, Denmark which was attended by WD specialists from around the world. Dr. Sam Shribman attended from the UK. Lisbett Ottesen, the chair of the Danish WD Support Group, hosted a virtual get-together for European patient representatives from France, Germany, Spain and Italy on the Sunday.

### WDSG-UK Management Committee Meetings

During 2022-23 the management committee met formally three times in May, October and January via Zoom and informally after our 12th Annual General Meeting in July.

### WDSG-UK 12th AGM—Sunday, 3 July 2022

See report opposite.

### WDSG-UK—Coffee Morning Rare Disease Day 2023

On Sunday, 26 February 2023 WDSG-UK held its third Zoom Coffee Morning to mark Rare Disease Day, the first Rare Disease Day having been introduced by the European Organization for Rare Diseases in 2008. Our event was run exclusively for members of the Group and we thank Dr Gillett, Dr Dooley, Dr Shribman and Miss Maggie Burrows for joining us too.

### WDSG-UK 13th Annual General Meeting*

The 13th AGM will be held face to face at the Cambridge RUFC on **Sunday, 23rd July 2023** at 1130 a.m, an invitation to which is included with this newsletter. The accounts for 2022-23 and the minutes of the 12th AGM will be made available to you then. Election of officers and members of the WDSG-UK Management Committee for 2023-24 will take place then. Current members of the committee**, Mary Fortune, Liz Wood, Debbie Buckles** and **Valerie Wheater** have submitted their names for re-election for the coming year and **Claire Stapleton** has asked for her name to be put forward. If you would like to join the committee, then please write to Valerie.

***This will be our first face to face meeting for 4 years, so we hope to see as many of you as possible. Happy Easter**!

### The Wilson's Disease Patient Register—UK

WDSG-UK supports research into all aspects of Wilson's disease. If you haven't yet signed up for the Register, please consider doing so. A pamphlet and information sheet are available to download off our website, together with the corresponding registration form.

### WDSG-UK Fb Site

Our WDSG-UK Facebook Group is a great resource for keeping up to date with what's going on at WDSG-UK headquarters. It is a private group so only members can read posts. If you aren't already a member, why not give it a go?

# Wilson's Disease Support Group Meeting & 12th AGM

## Sunday 3 July 2022 via zoom

We would like to thank everybody who attended our *virtual* AGM last July hosted by committee member Liz Wood. In particular we would like to thank representatives from the medical profession including Dr James Dooley, Dr Godfrey Gillett and Miss Maggie Burrows who gave up their morning for us. Turn-out amongst members was good and it was pleasing to welcome some new members and lapsed members, too.

As Chair of WDSG-UK Graeme Alexander swiftly conducted the business side of things. The minutes of the 2021 AGM were agreed and signed and apologies for absence at this year's AGM given. Graeme reported on the success of our informal Coffee Morning Zooms where members who lived too far for face to face meetings were able to join us from the comfort of their own homes.

He gave an update on the work of BASL (The British Association for the Study of the Liver) in relation to its Wilson's Disease Special Interest Group (WDSIG) that meets every year. National Guidelines on the Investigation, Treatment and Management of Wilson's disease have now been published and are available to download off the BASL website or off the home page of the WDSG-UK website. Graeme explained that the aim of the guidelines is to standardise the care offered to patients across the UK providing guidance to less experienced doctors in the management of their patients. Contact details of WD specialists are included in the guidelines such that doctors everywhere should have access to best practice.

The audited accounts for April 2021 to April 2022 were presented and members were thanked for their continued support. It had been an exceptional year for fundraising with Laura Nicolson, Maria Selwyn, Liz Wood and Val Wheater raising £4,500 between them. The formalities of the meeting were concluded at this point with committee members being elected for 2022-23.

Liz then arranged for us to be split into smaller *chat rooms* for the remaining 25 minutes during which time various other topics were discussed. These included current storage conditions for trientine, joint disease and connective tissue disorders in patients with Wilson's disease, psychiatric presentation in patients and what developments there had been in gene therapy over the past 12 months. Graeme offered to look into some of the questions raised and report back at our 2023 AGM.

The meeting concluded at 1205.


***Screenshot of some of the Attendees at the 12th AGM***

# Fundraising 2022-23

Valerie

## Maria's Half-Marathon — Mar '22

In its 40th year, the Brentwood half-marathon organised by the Rotary Club of Brentwood à Becket, took place on Sunday, 20th March 2022 with 4,000 competitors taking part. One such competitor running for the first time on this 13.1 mile course was Maria Selwyn, the mother of Oliver, one of the younger members of the Group. She says,


*Maria—No. 1697 on the right*

"Oliver was diagnosed when he was around 7. He had been having a lot of nose bleeds and the GP sent him for a full blood test, which showed elevated LFT levels. Initially, this was put down to his body fighting off infection, but fortunately the GP didn't just leave it there. She sent him for further tests which eventually confirmed a diagnosis of Wilson's disease. Luckily for Oliver the disease was caught early with no other symptoms and apart from taking penicillamine and avoiding certain foods his life has carried on much the same as before.

He is now a normal, healthy 15 year old teenager currently taking his GCSEs. He enjoys playing football, going to the gym and driving his mum mad!!"

We thank Maria for taking part in this fundraising event and for raising the very considerable sum of **£619** for the Group. We are very grateful to her and to everybody who sponsored her and we wish Oliver well in his forthcoming exams.

## AFC Rushden & Diamonds Youth—Ladies Football Match — 8 May '22

Tracy Stephen led the "Blues" to a 6-4 victory in the Annual Ladies' Charity Football match on Sunday, 8 May, after a two year hiatus because of the Pandemic. The Youth Section of the Club has been fundraising for us since 2016, after one of their promising juniors Sam Fitzgerald had to stop playing football following his Wilson's disease diagnosis the previous year. The Club wanted to show its support for Sam and other patients in the UK affected by the disease.

*Photograph Courtesy Prestige Photography*

*Mark presenting us with a Cheque for £500 after the Match*

Fundraising continues throughout the season and culminates in the ladies' football match where, since 2019, "The Wilson's Cup" has been fiercely contested. A lot of work goes into match day preparations and the talent of the ladies is formidable. The match was a great appetizer for the 2022 UEFA European Women's Football Championship which took place only two months later.

We thank Mark Swindells, Club Lead and Treasurer, and Tracy Stephen for their fundraising efforts and for presenting WDSG-UK with a much appreciated cheque for **£500**. We also thank all those who supported the fundraising throughout the season and in particular the ladies for providing such a fitting end to it.

**ANSWERS: 1.Dingbats 1** Triangle, **2** Piano (P an' O), **3** Recorder (LP is a record + a), **4** Clarinet (clar-in-et), **5** Cymbel (÷is a symbol), **6** Cello (chair—low), **7** Gong (g on g), **8** Tuba (Tube a), **9** Double Bass; **2.Wordsearch** You're left with Nul Points!; **3.Anagrams 1** Fly on the Wings of Love (2000), **2** Love Shine a Light (1997), **3** Making your Mind up (1981), **4** Waterloo (1974), **5** Hard rock hallelujah (2006), **6** Puppet on a String (1967), **7** Save your kisses for me (1976); **5.Riddles 1** a xylophone, **2** a rubber band, **3** The letter *P*, **4** Your voice; **6.Flags a** Italy, **b** Netherlands, **c** Belgium **d** Poland, **e** France, **f** Germany, **g** Iceland, **h** Romania, **i** Denmark, **j** Austria.

# Phil & Kurt's 24hr National 3 Peaks' Challenge – 22/23 July '22

Phil Angus and best friend, Kurt McGuinness, former Regimental Gunners in the British Army, set themselves challenges each year. This year they decided to compete in the National 3 Peaks' Challenge which involves climbing the highest mountains in Scotland, England and Wales, (Ben Nevis, Scafell Pike and Snowdon) in under 24 hours. The time starts from the first step on Ben Nevis, includes travel times between peaks and finishes when they return to the foot of Snowdon (nowadays referred to as Yr Wyddfa) after completing its 1,085m (3,560 ft) ascent. To make the challenge tougher, they decided to carry 35lb (15kg) Bergens on their backs, Bergens being the standard military back-packs carried by soldiers.

***Phil—at the top of Snowdon in 72 mph winds and torrential rain! At least it didn't snow!***

And to make their venture worthwhile, Phil and Kurt decided to raise funds for two causes close to their heart. We had the good fortune of being one of those causes because of our association with Ben Ryan, a patient from Lancashire for whom Wilson's disease has left its scars. Phil and Kurt were good friends with Ben at school back in the early 2000s and wanted to do something to highlight the disease. By using a crowdfunding platform, Phil advertised their challenge as follows:

***Ben***

"Another year another challenge, which myself and Kurt are doing for a couple of reasons and to raise money towards 2 extremely valuable causes listed below.

...In September 2016 a good friend of ours from High School Ben Ryan was unfortunately diagnosed with Wilsons disease. This disease has left Ben unable to walk and communicate properly and It goes without saying that this has not just had a life changing effect on Ben but also on his family and friends. This is a disease that most of us have probably never heard about and after seeing the impact this has had on Ben, it feels only right that we do something to not only raise money for the Wilsons Disease Support Group—UK, who've played a massive part in not only helping Ben with his recovery but also helped support his family throughout the challenges they've had to face, but to also raise awareness so people have an understanding of the signs and symptoms to enable them to recognise this potential disease at the earliest opportunity and seek medical advice."

Originally the challenge was to have taken place at the beginning of April, but 24 hours before they were due to set off there was a heavy snowfall on Ben Nevis which led to reduced visibility on the paths at the higher altitude. Disappointed after all their training, but with no control over the weather, Phil and Kurt took the tough decision to reschedule the climb for later in the year when the weather was guaranteed to be warmer.


***Kurt and Phil at the bottom of Snowdon with 15 minutes to spare!***

Meanwhile, Phil asked if WDSG-UK had any branded T shirts that they could wear for the challenge, which alas we don't any longer. However, we approached new member, Guy (his story appears on *p16)* who runs his own printing firm *Vario* in Bucks and he kindly offered to help. With his generosity and the generosity of clothing manufacturer *Rohan* who donated the basic T-shirts, Guy designed, printed and supplied two T-shirts for the occasion. Donning these T-shirts, Phil and Kurt completed the challenge in 23hr 45 mins 36 secs with literally 15 minutes to spare!

In all they raised a staggering **£2,175.68** for us. We congratulate them on completing the challenge, thank them for the hard work that went into it and wish them well in all future challenges. We also thank all who sponsored them and supported them to make the challenge happen.

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# Flóra's Story

by Flóra Kovács

My name is Flóra and I'm 25 years old. I was born and raised in Hungary. I was always a good student and took a degree in Commerce & Marketing at the University of Budapest. While I was at University, I met my fiancé Dániel to whom I got engaged in May 2019.[1] He was working and living in Oxford at that time and as soon as I graduated I moved to the UK to be with him.

Once in the UK, I started working as an Administrator at MINI Plant Oxford. It was October 2019 and all was well. However, less than ten months later in August 2020 I started to develop a tremor in my left hand followed a couple of weeks later with uncontrollable shaking in my right leg and later my left leg. I called my GP who wanted to see me immediately. After two appointments, I was diagnosed with stress and prescribed Propranolol. It was the first year of the Pandemic and lockdown had affected the lives of everybody. I was struggling with motor control in my daily life finding brushing my hair, handling cash or cards and chopping vegetables extremely difficult. Dániel helped me with everything. I was very frustrated, I had mood changes and I was depressed.

In December that year, I went to visit my family in Hungary for the Christmas holidays. When my family saw me shaking and not being able to do anything for myself, they were naturally worried. I attended hospital and they admitted me. None of the doctors had a clue what was going on. Because of the Pandemic, visitors weren't allowed, and I had difficulty using my phone because of my severe tremor. I barely spoke to my mom and my fiancé during that time. It was a nightmare. I was sent home on the 23rd December, the day after the lead neurologist finally started to investigate me for Wilson's disease.

I quitted my job in Oxford and remained in Hungary after the holidays. I had so many appointments and undertook lots of tests. I was tested for serum caeruloplasmin (0.05 g/L) and had high urinary copper output in a 24-hour collection. I had an MRI scan and ophthalmological examination, but a formal diagnosis required having a genetic test. In February 2021, we received the test results: a heterozygous mutation in ATP7B (H1069Q) was identified.

Initially I was treated with zinc acetate pending approval to start penicillamine. I kept to a strict low-copper diet. I had a disabling tremor in the upper and lower limbs at this point and had developed dysarthria (difficulty speaking) and drooling with angry outbursts. I was unable to write, use cutlery, wash myself, drink, dress or carry out other daily activities. I was hopeless.

My treatment was switched to penicillamine in March 2021 and the dose was gradually increased to 1350 mg per day over six weeks.

My mother looked after me during this time and by summer the tremor was much improved, and the dysarthria and drooling had stopped. I was able to write a few words and even eat by myself slowly. Finally, I felt something good was happening.

But around August 2021, my condition deteriorated again. The tremor in both the upper and lower limbs became worse. I had difficulties walking up stairs and developed an unusual gait disorder where I was pulled backwards after walking more than a few metres forward. Of the 10 million people living in Hungary only around 300 have Wilson's disease. Doctors don't have a lot of experience of managing patients and so they didn't know what to suggest.

***"...even my neurologist and gastroenterologist told me to come back!"***

In the meantime, from Hungary I joined the WDSG-UK Facebook Group. I wanted to come back to the UK, but as I was unable to work I was concerned about the future. I chatted a couple of times with Valerie and she got in touch with Dr Gillett who agreed to take me on as his patient once I returned to the UK and he even offered to speak to my doctors in Hungary if I wanted him to. That changed everything.

My family saw that Hungarian doctors couldn't help any more, so with bleeding hearts but in the hope that experienced doctors in the UK could intervene, they let me return to Oxford and even my neurologist and gastroenterologist told me to come back!

*In Eger Hospital, Hungary, 2021*

I finally returned to the UK in November 2021. I was still in a bad shape and needed help with everything. My fiancé was cooking for us, doing the laundry, helping me dress, feeding me, brushing my hair, and brushing my teeth. While he was working, my friends stepped in to help out. After a couple of weeks, I received a referral to the Queen's Square Wilson's Clinic at the National Hospital for Neurology & Neurosurgery in London.

The appointment was to have been a face to face appointment with Drs Godfrey Gillett and Samuel Shribman in January 2022. I was very excited to meet them at last. Unfortunately, due to the new Omicron variant of Covid that had arrived in the UK, the appointment was switched to a virtual one. However, we discussed everything and I was truly impressed. They knew exactly what I was struggling with and they already had solutions to my problems. They insisted on seeing me in person the following month as my tremor was so bad.

***"...another setback happened. Storm Eunice arrived..."***

Again I was super excited to be meeting them at last, but then another setback happened. *Storm Eunice* arrived and all forms of transport were affected making it impossible for me to travel. But it was third time lucky. In March, finally, I made it to Queen Square. My penicillamine dose was decreased to 1000 mg per day, and I was prescribed 40 mg Propranolol three times a day for the tremor. My fiancé came to the appointment with me, and we both left feeling like we had known them for years! They were so helpful and friendly and treated us with great kindness.

Two to three weeks after the visit, I began to feel that something was changing. After seeing these two brilliant doctors my condition started to improve. Slowly, I was able to wash my hair and brush my teeth. It wasn't the same as before I was ill, I was still clumsy, but we saw improvements from one day to the next. In May 2022 I was able to do everything for myself and I even went back to work.

Since then, I have got my life back fully. By the end of September 2022, I changed my job and started working for Oxford University Press as a Product Data Coordinator and I'm very proud of it. Now, with my fiancé, we are planning our wedding. I couldn't be happier. He has stood by me throughout. All my life I had been independent, so accepting help from others was very hard for me. I didn't like asking favours or asking for help, but I had no other choice. This whole journey has made me appreciate the small things in life like using my hands and feeding myself, things that before I had just taken for granted. It changed my view of everything. I had always been a cheerful and smiling girl, but now I'm even more joyful. Positivity is the key, but at times it was difficult.

I would like to take this opportunity to say thank you to my amazing family and friends, especially my mom and my fiancé, who were always beside me, Valerie for her support and for putting me in touch with Dr Gillett and Dr Shribman and Dr Gillett and Dr Shribman for their brilliant work, care, expertise and kindness!

* * * * *

[1]For anybody who likes romantic stories, here's one for the books. Dániel and Flóra had been at High School together but he was 2 years older and they had never actually spoken. This he bitterly regretted. But he never forgot her and when he was living 1500 kms away in the UK he decided to look her up. He found her on Facebook and they started to chat. That was at the end of 2016 just as Flora was about to go to University. He flew back to Hungary to see her as soon as he could and it was love at first sight for them both!

Fast forward 3 years. One day Flóra and her best friend went to the cinema. As the two of them were sitting there waiting for the trailers to begin, to her utter bewilderment and disbelief a video appeared instead showing photographs of her and Dániel which had been taken during their 3 years of courtship. There were even clips of Dániel asking his and Flóra's parents for permission to marry her! Dániel had spent 6 months making the video and as it finished he emerged from the wings and got down on one knee and proposed. To rapturous applause, Flóra accepted! "It was the most romantic experience ever," she says. Anybody wishing to capture the moment, here is the link: https://www.facebook.com/daniel.sebestyen.9/videos/2426115090771970/

***An Unexpected Proposal***

We wish them every happiness in their future life together.

# Research Update—The CROWD Study

## The CROWD Study, UCL Queen Square Institute of Neurology, London — Update

The CROWD (Cohort Research On Wilson's Disease) Study was launched in December 2018. In Part 1, the aim is to identify genetic factors that determine whether someone with Wilson's disease will develop neurological problems or not. People across the UK were invited to participate by completing an online questionnaire and sending in saliva samples. We have now finished collecting samples, extracting the DNA from the samples and are proceeding with the genetic analyses.

In Part 2 of the Study, the aim is to understand how to measure and monitor the effects of Wilson's disease on the brain. Forty patients attended Queen Square[1] for clinical assessments, blood tests and MRI brain scans in 2019 and this part of the study has been completed. Some findings have been discussed in last year's newsletter. In this article, **Dr Samuel Shribman** and **Miss Maggie Burrows** discuss some of their work looking at cognition in Wilson's disease. Please do not hesitate to contact them at s.shribman@ucl.ac.uk, if you have any questions about their research.

*Dr Sam Shribman*

## Cognition, MRI and Wilson's disease

Last year we discussed how magnetic resonance imaging (MRI) scans can help us understand how Wilson's disease can affect the brain. This years's article describes research findings exploring the link between cognition and changes on MRI brain scans in Wilson's disease.

***What is cognition?***

Cognition is the mental action or process of acquiring knowledge and understanding through thought, experience, and the senses. It can be divided into different *cognitive domains* including abstract reasoning, memory, language, calculation, social cognition and executive function. The latter is a term used to describe the mental processes that allow us to plan, focus attention and multitask.

As individuals, we all have strengths and weaknesses in different cognitive areas – some of us may be very competent at map reading and reverse parking our cars – others not so. Some of us may be proficient at having several things on the go at the same time AND hold a conversation on the phone – others would find this very difficult. Some diseases, however, affect cognition in very specific areas. This may not bother the person too much unless it starts to affect their daily life – at this point we would say the person has dementia.

***How is cognition measured?***

A range of tests can be used to measure different aspects of cognition. This usually involves working through a task with a clinician. There are at least 10 different tests for assessing memory alone, including tests for different types of verbal and visual memory and short- and long-term memory. Tests scores are compared to average scores from the general population to help determine how someone is performing. Where someone performs below the expected level for a given test this is referred to as a *cognitive deficit*.

***What was already known about cognition in Wilson's disease?***

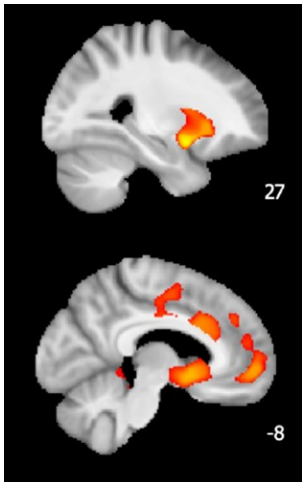
A number of studies have looked at cognition in patients with movement disorders and/or psychiatric symptoms due to Wilson's disease. They found that several cognitive domains can be affected and that cognitive deficits were often subtle. A small minority of patients only were considered to have cognitive impairment that limited daily life. Patients with Wilson's disease with liver disease only had not been rigorously tested for this. It was also unclear how cognitive problems relate to the changes that can be seen on MRI brain scans.

***What did we do in the CROWD study?***

We performed a range of cognitive tests in 40 people living with Wilson's disease who visited the National Hospital for Neurology and Neurosurgery as part of the CROWD study. We also performed MRI brain scans to test whether there was an association between test scores and subtle changes in brain structure.

***What did we find?***

Firstly, we found that patients with movement disorders and/or psychiatric symptoms had lower scores for tests of processing speed and executive function than patients with liver disease only. These scores also correlated with the severity of the movement disorder meaning that patients with more disabling movement disorders were also more likely to have lower test scores.
Secondly, we found that both patients with movements disorders and/or psychiatric symptoms and patients with liver disease only, had subtle problems with memory, specifically related to recognising faces.




***How does this relate to MRI brain scans?***

We compared the scores for each cognitive test with the volume of different brain regions. For tests of processing speed and executive function, scores were closely associated with the volume of the basal ganglia. These are the deep structures in the brain that control movements. This area is highlighted in red/yellow in the upper image on the right showing a side view of the brain.

Scores for test of memory for faces were associated with more subtle but widespread changes in the brain. These areas are highlighted in red/yellow in the lower image on the right showing a side view of the brain.

***What does this mean?***

These findings suggest that the disease process leading to problems with movements and executive function may differ from the disease process responsible for more subtle problems with memory. It is possible that copper affects different parts of the brain in different ways or other consequences of the faulty Wilson's gene might explain our findings.

***Why is this important?***

A small minority of people living with Wilson's disease have ongoing problems with cognition that limit them at school, college, work or in other aspects of their daily life. It is important that we understand the type of problems with cognition that can occur, so we know how to identify these early and offer support. Developing a clearer understanding of the associated changes in brain structure also provides insights into how Wilson's disease can affect the brain.

**We would like to thank all the people who took part in the study and the WDSG-UK for their support.**

*Reference*
Neuroimaging correlates of cognitive deficits in Wilson's disease: a multimodal, whole-brain MRI study. Shribman S, Burrows M, Convery R, et al. Mov Disord 2022; doi: 10.1002/mds.29123

***[1]Queen Square, London***
***National Hospital for Neurology & Neurosurgery, University College London***

# Denise's Story

by Denise Jarvis

Hello, I am Denise and my Wilson's disease journey started a long time ago in 1968. I was nearly 8. I was living in a small village in Warwickshire with my parents and younger brother when I started to experience symptoms. All I wanted to do was sleep. I was thin, weak and lethargic and my tummy was very swollen. I was having terrible nose bleeds which were difficult to stop.

My mother took me to see Dr Boyd, our local GP. He had no idea what the matter was, but thought it might be glandular fever because I was so tired. Luckily, he didn't leave it there and he had me admitted to the Horton Hospital in Banbury for further investigation. I was there for a week. During this time a paediatrician from Oxford came to see me and recognised immediately how ill that I was. He arranged for me to be transferred to the Radcliffe Infirmary in Oxford where he was based, so that he could carry out further tests.

I had actually been in hospital four years earlier but I don't think that was connected. I had been given a vaccination against diphtheria and suffered a severe allergic reaction which caused me to pass out. Unable to breathe, I was rushed to my local hospital in Leamington Spa and given a tracheotomy. I remained in hospital for two months during which time I was nursed in an oxygen tent and only fed soups. I made a full recovery, though I still have the scar on my neck to remind me!

## *"There was no time to lose…"*

Once at the Radcliffe, Dr Bower carried out a further barrage of tests including a bone marrow biopsy. I was there for two weeks and eventually my parents were told that I probably had a rare, inherited metabolic condition called Wilson's disease. An ophthalmologist had found the characteristic Kayser-Fleischer rings in my eyes which supported this diagnosis. There was no time to lose and Dr Bower arranged for me to go to Addenbrooke's Hospital in Cambridge to be looked after by the leading authority on Wilson's disease, Dr John Walshe. An ambulance took me the following day with my parents in hot pursuit.

We arrived at Addenbrooke's (which in those days was on Trumpington Street in Cambridge), and Dr Walshe and his assistant Kay Gibbs were there to receive us. This was the third hospital I had been admitted to in under as many weeks and I remember being tucked into a bed on the children's ward by a staff nurse wearing a tall, white, starched cap balanced on her head! Dr Walshe satisfied my parents that I was in the best possible hands and so with great courage they settled me in before returning home.

There was another girl on the ward with Wilson's disease called Lynn. She was a few years older than me but they put our beds side by side and we soon became friends. She was very ill, too. Kay would pop down to see us every day when she had time and every evening after work my father would come to visit. It was a 160 mile round trip, but he was determined to read me a bedtime story before I went to sleep. My mother seldom visited during the week as she had my brother to look after so I used to look forward to the weekends when I saw them altogether. Lynn wasn't so lucky, however, as her father lived in Switzerland and he rarely came to visit.

## *"...it was 50/50 whether or not I would survive."*

Every day Dr Walshe would come to see me to do various tests. I remember having to draw elephants for him and in return he once drew an elephant on my tummy around my liver and spleen! He took regular bloods which I hated, distracting me by telling me to think of my Christmas dinner! I also had to do 24 hour urine collections which aren't at all easy when you are only 8. Nurse Hazel, who was so kind to me and became a surrogate mother, would take me into the sluice and turn on the tap in the hope that the sound of running water would help "get me going!"

My condition remained critical. Dr Walshe told my parents that they had "a very sick daughter on their hands" and it was "50/50 whether or not I would survive." Perhaps today I would have been offered a liver transplant, but it was the end of the 1960s and liver transplantation was in its infancy and had a poor outcome. Instead I was treated with penicillamine, which I still take today 55 years later. I am the model patient as I have never missed a tablet in my life!

I remained in hospital for several months until I was considered stable enough to be allowed back home. Dr Walshe told my parents to ring at any time if they were at all worried and I did in fact get readmitted on a couple of occasions. After that I would be seen regularly as an inpatient every 3 months, lengthening to every six months and later every year. I missed a lot of school and

although my parents sent me at one stage to a small private school, I never really caught up. Once I started work my employers recognised my potential and sponsored me to go to College where I gained an HNC.

I have had an enriched life and have stayed in good health. I will always be indebted to Dr Walshe and Kay for the part that they played in my life. They made a wonderful team. Dr Walshe continued to look after me until 1999, when my husband and I moved to the States with our 3 year old son.

Since returning recently to the UK to be near my elderly parents, I have been attending the clinic at Queen Square run by Dr Gillett and Dr Shribman. I feel I am in safe hands and thank them for supporting me as I continue along the path of my Wilson's disease journey.

# Kay Gibbs

by Valerie

Denise speaks of Kay in her patient story, but not all of you will know who Kay is. For those who don't, allow me to introduce her and the role that she played in the lives of Wilson's patients at Addenbrooke's and beyond.

*Kay at a WDSG-UK meeting in Cambridge in 2012*

Kay qualified as a biochemist from the London Institute of Medical Sciences during the 1950s. She later took up a post in Cambridge at the Institute of Animal Physiology (now known as the Babraham Institute) working on pregnancy toxemia in sheep! She felt well qualified to do this job as she had worked on call at several maternity hospitals in London whilst she was doing her training!

Unfortunately, the job at Babraham didn't work out and Kay looked around for something else. By chance at this time Dr Walshe was advertising for a chemist to help with his work, so although it wasn't what she was really looking for she thought she'd give it a go until she found something better! As she got on well with him and the job grew more and more interesting, she decided to look no further and there she stayed for another 27 years until they both retired.

The original brief was for her to work on penicillamine, but as Dr Walshe's patient base increased so did the amount of work that was required to take care of them. Her main responsibility in the lab was to carry out copper analyses (of urine and tissues). She would collect the bottles of urine from the wards, getting to know the patients as she did so. Sometimes patients would be asked to carry their own, unbagged bottles of urine over to her to save her time. It was a chance conversation she had with an ex-patient after she retired that she found out that patients had found this highly embarrassing. She apologises for that. **She** was so used to handling it herself every day that it never occurred to her that they would be embarrassed.

For vulnerable children like Denise, whom she remembers very well, she would go out of her way to spend time with them and make their stay more comfortable. If they were fit enough to leave the hospital she would even take them on short outings down to the River Cam to feed the ducks and watch the punts go by.

When Dr Walshe started to use nuclear medicine to map copper deposits in patients, it was Kay's job to drive to Amersham to pick up radio copper. It had a half life of 12 hours, so she would rush it back to Cambridge before standardising it, making it into a solution and sterilising it. Only then was it safe to be injected into patients.

By the end of the 60s there were several patients intolerant of penicillamine. Dr Walshe and Dr Hal Dixon, a Cambridge chemist, had worked on an alternative chelator, a trientine compound known as TETA. Kay would have to make this up for patients in the lab by purifying it, drying it, testing it and loading it into capsules. She would then pack and post the capsules to patients across the globe. When there were 10 patients relying upon it she said she had reached her limit!

During her Wilson's career, she co-wrote several medical articles with Dr Walshe and because authors were listed alphabetically her name always appeared first on the paper. Believing her to be the principal author, she was once invited by professors in the United States to give a talk on TETA. She found this very amusing as Dr Walshe wasn't (initially) invited, too!

Asked what she misses most about her time at Addenbrooke's, Kay says, "Firstly, it's the stimulation. At 95, there's not much of that about! I miss the contact with patients and am always happy to hear from them when any of them contact me.

I was so fortunate to have had such an interesting and enjoyable career. It was a privilege to care for so many lovely patients and to see the sick children grow up into healthy adults. I think of them as my Wilson's family. I thank them for being so tolerant of all the trials we put them through and send them all my love and best wishes."

# Reports, Clinical Trials & Updates

## The BASL Wilson's Disease Special Interest Group (WDSIG)

*Dr Bill Griffiths*
*Lead BASL WDSIG*

Through the British Association of the Study of the Liver (**BASL**) a number of Special Interest Groups (**SIGs**) has been established in certain areas of liver disease in order to bring relevant expertise together from around the UK. The **WD SIG** was one of the first to get going and continues to attract more clinicians to its membership. It had its first meeting in December 2017. Wilson's disease lends itself well to this new initiative, being a rare disease that has several types of clinician involved in its care. Sitting in the Group are hepatologists, neurologists, clinical chemists, paediatricians and psychiatrists as well as research scientists and representatives from WDSG-UK and NHS Digital, formerly Public Health England (**PHE**). Over 100 individuals from across the UK are registered on the WDSIG database and the SIG has now approved 15 specialist WD adult centres across England. For further information on the remit of the WDSIG and minutes of its meetings, please visit the BASL website www.basl.org.uk.

### 7th Meeting—25.11.22 via Zoom—Attended by over 40 members

The WD SIG remains a close-knit and productive group. We held our annual meeting on 25.11.22, again via zoom as this allows as many people to attend as possible. We also score highly there for 'sustainability'. 'Hybrid' meetings work well but need the technology to go with them - something to think about for the future meetings. There is something about 'face to face' though which makes meetings more enjoyable, cements the interactions and fosters collaboration more easily. Having said that further research collaboration was born from this meeting, I was delighted to find out later.

So what did we discuss? We initially talked about the fact that Cufence (trientine dihydrochlorine) no longer requires refrigeration whether opened or unopened (this is the advice pharmacies are giving to patients in line with approval from the European Medicines Agency in May, 2021) though we feel some vigilance is worthwhile. Rupert Purchase explained that the chemistry of the dihydrochloride form (Cufence) means it is more 'reactive' than the tetrahydrochloride form (Cuprior) and that storage temperature still needs to be borne in mind.

A fundamental and highly successful output from the WDSIG is guidance on managing patients with WD, published in April 2022 via one of the Lancet journals. We have disseminated this through relevant professional bodies and hope it will be a useful resource for clinicians across the UK and beyond. Indeed, I have received positive feedback from various corners of the Earth. A key message is that genetic testing should move further up the diagnostic algorithm though in reality labs are struggling to catch up after the pandemic.

We received an update from colleagues at NHS Digital – a key step forward was the acquisition of 'Blueteq' data allowing mapping of trientine prescriptions across England. Cross-referencing the WD database they hold with stored genetic laboratory data will be interesting. Helen Patrick and Miranda Durkie are studying the Sheffield genetic data as to how different forms of WD might be explained.

We were fortunate to host some excellent research talks. The first was from the University of Sussex where George Kostakis explained how they are developing new copper chelators. They are utilising predictive models to assess properties of chelators including whether they will cross into the brain. We heard about exciting work from the 'PET' imaging department at Kings where George Firth described state of art machinery that can follow copper around the body, potentially useful for both diagnosis of WD and seeing how treatments work. Sam Shribman showed us insightful neuroimaging work on the poorly recognized cognitive deficits which are not uncommon in WD.

In the clinical section, Chris Cussen reminded us of some of the manifestations of WD that can occur outside the liver and brain. Jan Coebergh described cases where neuropsychiatric symptoms in WD can add an extra layer of complexity when evaluating patients with WD.

We heard from James Liu Yin about the latest studies on WD, presented at the American Liver Association (AASLD), and from Aftab Ala about ongoing and upcoming clinical trials. Of note, through Kings and the Royal Surrey, Aftab is embarking on novel gene therapy studies in WD. He is also setting up a couple of pilot studies to look at the diagnostic potential of ATP7B peptide testing and a hand-held method for spotting 'KF' rings, the archetypal eye sign in WD.

Finally, the SIG formally acknowledged the sad passing of Dr John Walshe.

**Bill Griffiths, Consultant Hepatologist and WDSIG Lead, Cambridge—13.03.23**

## VIVET THERAPEUTICS – The GATEWAY Clinical Gene Therapy Study

Vivet Therapeutics is a biotechnology company based in France which is developing gene therapy treatments for rare liver disorders including Wilson's disease. Their new gene therapy treatment for Wilson's disease is known as VTX-801 and it is currently under investigation in the GATEWAY Study, a clinical trial which will explore safety, efficacy and durability of the treatment. Centres have been opened in the United States, the United Kingdom, Denmark and Germany to undertake the trial. Vivet is collaborating with Pfizer on the clinical supply of VTX-801.

In the UK GATEWAY is now enrolling patients for the trial, which will be conducted in the UK by Prof. Aftab Ala at The Royal Surrey County Hospital, Guildford. Patients need to have a hepatic Wilson's disease diagnosis, be between 18-65 years old and be stable on WD treatments. The Study involves patients receiving a single dose of VTX-801, which will occur in a hospital setting. No patients will receive a placebo treatment. Patients will then return to The Royal Surrey at planned times over the following year so that the study team can assess how they are doing. These visits will occur more often in the early part of the trial (first year) and the number of visits will decrease thereafter. The study will continue for 4 years after the initial 1-year trial period, which will be a critical part of understanding how well VTX-801 works over time.

**For more information please visit the dedicated website www.gatewaytrialwilsondisease.com or https://clinicaltrials.gov/ct2/show/NCT04537377 or contact Valerie.**

## Drug Trial — UNITED Study—UNIVAR Solutions B.V.

### A Post Authorization Efficacy Study (PAES) to gain a better understanding about the effects that Cufence® (trientine dihydrochloride) has on the body in adults and children with Wilson's disease

Univar is conducting a post authorization efficacy study to characterize the pharmacokinetics and pharmacodynamics of Cufence (trientine dihydrochloride) and to investigate the efficacy and safety in Wilson's disease patients. This study is open for children, adolescents and adults, from the age of 5 years on. Patients included in the study should previously have been treated with D-penicillamine (≥18 years) or D-penicillamine or zinc (<18 years).

During the past year, Univar has opened one additional study site in Germany that has resulted in a total of 11 study sites across different countries in Europe. These countries include the UK - with two study sites at King's College Hospital in London - France, Germany, Denmark and Poland. Currently, Univar has enrolled more than half of the patients they are aiming for. Enrolment is ongoing until the end of August 2023 and the first patient who was included in the study will finish the study in August this year.

On March 10th 2023, investigators and personnel from the study sites met for a one-day Investigator meeting in Amsterdam. This meeting was a great opportunity to update the investigators on the study progress and to discuss lessons learned related to the study.

Univar is looking forward to continuing this trial and sharing the first results sometime in the future.

**If you are interested in taking part in this study, please contact WDSG-UK**

## Public Health England (PHE)—Wilson's Disease Pilot Study—Update

NCARDRS (The National Congenital Anomaly and Rare Disease Register) landed in NHS England, along with the rest of NHS Digital at the beginning of February 2023. This is the third organisation that NCARDRS has been in since September 2021.

NCARDRS worked with NHS England Highly Specialised Services Commissioning to access the trientine prescription data and will continue to collaborate with the BASL WD SIG and Dr Osob Mohamed on making the best use of this valuable data.

NCARDRS also has done some work with Miranda Durkie and Helen Patrick at the Sheffield laboratory. This was the first time that NCARDRS data was used to support a rare disease genetics project. This project highlighted issues about data access on the NCARDRS side. Now that NCARDRS is settling into its new home, it is hoped that progress will be made on the national Wilson's Disease registration and output.

The registry currently contains over 600 patients from England. These have been reported from clinicians at 28 hospitals or identified using data linkage techniques developed by Dr Osob Mohamed and the NCARDRS team.

**Mary Bythell—March 2022**

## Supply of WD Meds in the UK (March 2023) - Update

**D-Penicillamine**

There was a hiccup in December with the availability of penicillamine but no problems reported since. Wholesalers including Alliance, AAH and Phoenix have ample supplies. Please let us know if you have any problems obtaining it.

**Cufence® previously known as Trientine Dihydrochloride**

There are no production or distribution problems anticipated with this product.

**Cuprior® (Trientine Tetrahydrochloride) made by Orphalan Ltd**

Trientine tetrahydrochloride (Cuprior®) was licensed in Europe in 2017 and first commissioned in the UK in 2019. It does not need to be refrigerated at all and is supplied as scored splitable tablets in blisters, each pack containing 72 tablets. There are no production or distribution problems with this product. Cuprior also comes with the option of home drug delivery for as long as required, which may include phlebotomy and/or urinalysis.

# A Lucky Shot

by Guy Clarke

The first time I noticed symptoms was in March 2020 after a friend's stag do. I was eating cereal and I couldn't stop my hand shaking. Initially I put it down to overconsumption of alcohol the night before, but over the next few days and weeks it didn't get any better. I decided it was time to go to the G.P.

By then the dreaded Covid had struck so I was only able to have a Zoom call with my doctor. He thought the symptoms would pass but when they didn't he asked me to go into the surgery to have some blood tests done. These all came back negative and he therefore concluded I was suffering from Covid anxiety, which I thought quite unlikely as I am generally a laid back type of guy. I was used to running marathons and triathlons and was always on the go, with a normal resting heart rate of around 45 bpm.

My tremors got worse, though I didn't have any other symptoms, or so I thought. The doctor still diagnosed it as Covid stress. However, at Christmas '21 things took a dramatic turn.

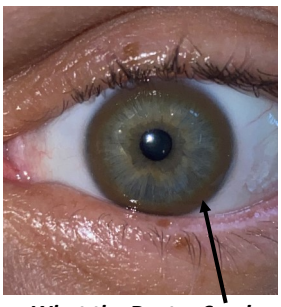
I run a printing business and every year we have a Christmas party at which we play silly games. This year we decided to take Nerf guns and have a battle after dinner (and a couple of drinks!) For those of you who don't know what Nerf guns are, they fire foam bullets with a hard plastic end. All was good fun until I got hit in my left eye with one of the bullets and partially lost my vision.

The following day was Christmas Eve. I woke up and thought it was much better but by the time I was in Santa's Grotto with the children later in the day I knew there was a problem! I went straight to A&E where I received treatment. They told me to book an appointment with Eye Casualty, which I did for Boxing Day.

***"It won't be Wilson's; it's never Wilson's"***

However, at 8.00 o'clock, Eye Casualty rang me to ask me to attend on Christmas Day instead. Concerned that something worrying had been found, Christmas lunch was put on hold! My dad gave me a lift to the hospital and waited with me. It was 4 hours before I got out by which time he had finished the book I had just bought him for Christmas!

At Eye Casualty the process seemed slow and now I understand why. Something worrying **had** been found! I was reassured that my eye sight would return to normal, but the ophthalmologist, Dr Rajammal, asked if I had any hereditary illnesses. When I said, "No," he asked me about my tremor which he said he couldn't help noticing. He also explained that he had seen copper deposits called Kayser-Fleischer rings around the corneas of my eyes which would suggest that I had a illness called Wilson's disease. I had naturally never heard of Wilson's disease and rang a friend who is a doctor as soon as I got home. My friend said, "It won't be Wilson's; it's never Wilson's!" With that kind of response, it is no wonder patients have difficulty getting an early diagnosis.



***What the Doctor Saw!***

A month later I was given an official diagnosis. That same day my wife joined WDSG-UK's Fb Group and explained what her interest was in Wilson's disease. Valerie messaged her privately. Once it was established I had a neurological presentation and wasn't being seen at a specialist hospital, with my permission Valerie contacted Dr Gillett and his team. Dr Gillet kindly agreed to take me on at his Queen Square clinic but continues to liaise with the consultants at my local hospital so they can all work together. I have received excellent care from everybody involved which no doubt has been instrumental in my making a good and speedy recovery.

When I started chelation treatment, my tremors got worse for 9 months and my speech became affected, but now 14 months later my symptoms are very mild. Unfortunately, my liver has been badly damaged but I hope that this will improve with time. I will be 41 this year.

Reflecting on events, it was the Nerf bullet in my eye which led to my diagnosis. I am so grateful that someone took aim at me and succeeded in hitting me, as without this the Wilson's disease diagnosis could have been further delayed and the outcome could have been a lot worse. I feel very lucky.

# Vitamin D, Covid & My Liver Transplant

by Joan Smith

Joan Smith, whose story appears in the 2004 newsletter (vol 5 issue 1), was diagnosed with Wilson's disease in 1963 at the age of 13. She presented at school with neurological symptoms which led to a decline in her school work. By the time she was diagnosed many months later she had tremors, was unable to walk or feed herself and was highly emotional.

An article highlighting the sunflower cataracts seen in her eyes was later published in a medical journal and was picked up by *The Sunday Times.* It was then reproduced around the world. Fame literally arrived on her doorstep when a letter was delivered to her addressed simply to:-


Sixty years later and after a successful liver transplant, Joan now issues an alert,

"I think I ought to make the Group aware of what has befallen me over the past couple of years, i.e. during Covid, in case it happens to anybody else.

I was diagnosed with Wilson's disease as a child and stayed mostly healthy until my liver failed in 2004 when I needed a transplant. I had remained on penicillamine since diagnosis but it had caused damage to my skin and as a result my dosage was reduced. Unfortunately, it must have been reduced too much as my liver started to deteriorate. My transplant was a great success and I gradually returned to normal life. I kept myself healthy by staying active and following all the advice given by the medical team.

I don't know if transplant patients are aware that the immuno-suppressant drugs you take to stop your transplant rejecting make you more prone to getting cancer, mainly skin cancer. I was given sunscreen on prescription and always applied it to exposed skin whenever I went outside. I was also prescribed a small dose of Vitamin D. After a few years I wasn't able to be as active as I would like as I was having problems with my legs. I put it down to ageing and my previous history of joint pain with Wilson's. However, my mobility problems got worse.

Then Covid happened and as a transplantée I was advised to shield. I barely went outdoors for two years. I exercised regularly indoors on my exercise bike until I could hardly pedal it any longer or even walk. I consulted my GP who sent me to see a physio. The physio thought it may be caused by the historic depletion of collagen in my body and I was given a course of exercises to do.

My pain got worse. I went back to the GP. At the same time my consultant at Addenbrooke's contacted me to say that my six monthly blood tests had indicated that I had a raised CRP level (a marker of inflammation or infection) which he suggested I ask my GP to investigate. For the next few months I was subjected to a variety of blood tests and investigations. I was now suffering chronic pain, my legs could barely support me, I was suffering anxiety, forgetfulness, hair loss, (I have lost half of my hair), and I was finding life miserable. One of the most recent blood tests has shown that I have a very low level of Vitamin D. For the past 5 weeks I have been prescribed a course of Vitamin D tablets - one tablet per week for 6 weeks. I haven't experienced much improvement yet but reading Dr.Google, any improvement may take a long time to show.

I had no idea that a lack of Vitamin D could make me feel so ill and I believe it could have been building up for many years. I would suggest that if anyone else has had a transplant and is suffering similar symptoms, please get it checked out immediately."

Now here's a coincidence. In the late 1960s there were few patients in the world who had been diagnosed with Wilson's disease and certainly only a handful in the UK. It was therefore somewhat of a surprise when Denise's grandmother (Denise's story appears on *p12*) got chatting to the lady who lived at the bottom of her garden only to find that she had a granddaughter with Wilson's disease, too! And that granddaughter was Joan Smith!

# Members' News 2022-23

It was only 18 months ago that **Laura** spent the summer cycling the length and breadth of Shetland raising funds for us. This was after a harrowing time getting a diagnosis of Wilson's disease and an even rougher time starting on her meds. Her story appeared in our 2020 newsletter. She now writes:

A little update for you all: my partner Neil and I welcomed a beautiful healthy baby boy on 26th January 2023. We named him Jack David Wishart. We are utterly delighted to become first time parents. Thinking back to how ill I was pre-diagnosis and under treatment at Aberdeen Royal Infirmary only 4 years ago, I never believed I would be able to look after myself far less a baby. I'm writing this to tell you that miracles do happen and to not give up hope. After we named Jack, I researched what the name Jack means and it means *God is Gracious*...and He certainly is."

* * * * *

**Linda Asher**, whose story appears in the 2009 newsletter, is a regular attendee at our annual meetings and a regular contributor to the newsletter. You might remember reading recently about the magnificent dolls house that she and her brother built, decorated and furnished between them. She now writes,

***Amy***

Just a short summary about me. I was diagnosed with Wilson's disease in 1957 at the age of 13 after my hands started shaking and my speech became slurred. My mother took me to see a neurologist at King George's Hospital, Hyde Park Corner (now *The Lanesborough Hotel*) soon after my symptoms began. The Doctor diagnosed my condition almost immediately by looking into my eyes with a bright light and seeing Kayser-Fleischer rings. He put me on medication straight away — penicillamine, a chelating agent—and I have remained on it ever since.

I went to Grammar School but missed a lot of lessons and couldn't write because of my shaky hands but I always loved Art and managed somehow to paint and draw. My headmistress put one of my paintings on the wall in her office.

I am 79 now and attend a clay modelling class at my local community centre which I have been going to for quite a few years. I love every minute of it. We call ourselves *Crackpots*. There is nothing nicer than feeling a lump of clay in your hands and turning it into a sculpting. My hands are still a bit shaky but I manage. Determination is the key. My fellow classmates think my work is a bit quirky. I talk to my sculpting as I'm doing it and so far none of them has answered me back! I love sculpting figures and heads.

***Oil painting of my grandson's dog, Gypsy***

Clay has been used for thousands of years so it has a wonderful history of its own. After the sculpting is finished it has to dry and then be put in a kiln at 1000°C for 15 hrs. It can then be glazed or decorated before it's put back in the kiln as before. It's quite a long process as you can imagine.

I also enjoy painting, particularly en plain air (out of doors), and using my iPad to paint imaginary scenes. Here are a few of my works that you might enjoy seeing.

***Chinese Warrior***

In 2019, **Sam Ho** the co-founder of the Hong Kong WD Support Group came over to the UK to do a Masters Degree in Biochemistry at Manchester University and while here made contact with WDSG-UK. He was hoping to attend the AGM in Cambridge the following year when the Group had plans to hold a 100th birthday celebration for Dr Walshe. However, because of the pandemic Sam was forced to return home early and, of course, anyway the meeting had to be cancelled. Once home he was able to collect messages from many of the Hong Kong patients in time to send them to Dr Walshe to mark his special day. The messages thanked Dr Walshe for all that he had done for patients across the world in discovering penicillamine and trientine and for devoting his life to their care and wellbeing. They were included in the 60 page book that the Group presented to Dr Walshe on his birthday.

When Sam read the news of Dr Walshe's death on Fb in October 2022, he felt compelled to get the HK Group together again to write messages of condolence to the family. As Christmas was approaching he waited for their annual Christmas party when patient member **Daisy** arrived with two small individual oil paintings that she had painted herself. The patients then wrote their messages on the back of each before posting them to the UK for delivery to Dr Walshe's two daughters, Susan and Clare.

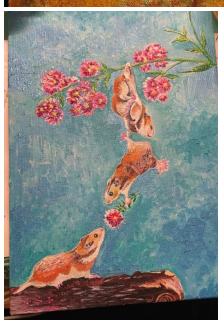
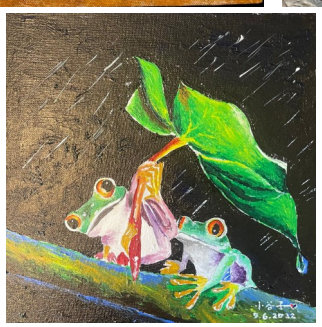
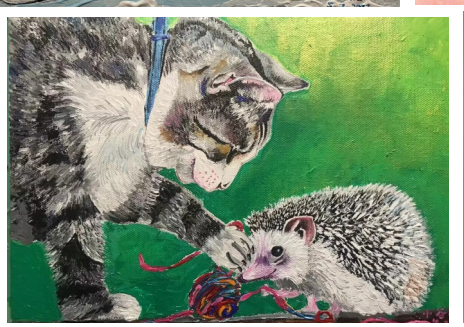

***Daisy's condolence cards to Susan and Clare***

Daisy's Wilson's disease journey started in 2001 when she was 20. She presented with neurological features including slurred speech and uncontrollable tremors. She wasn't able to do anything for herself. Music and Art had always been her passion, but she could no longer play the piano or paint. She remembers it as a dark period of her life, and in her depression she cut herself off from friends and family for fear of being ridiculed. At the same time she underwent a three year treatment plan at a hospital in Hefei, Anhui Province, and at last her health began to improve. She found gradually that she could manage to play the piano again and paint. Since then, she has never looked back and tries to concentrate on the positive. She would like to share some more of her artwork, which currently appears in an art exhibition in Hong Kong, with you now. Her message is, "I hope the purest love message from my artworks can bring positivity and hope to everyone, especially to my fellow Wilson's disease warriors. It is important for you to know, that along this difficult path, you are never alone."








**Danny Hiles**, whose patient story appeared in last year's newsletter, has fulfilled his recent ambition of taking part in the Transplant Games. Before his diagnosis of Wilson's disease at the age of 15, he had been very sporty and athletic playing football in a Sunday league and representing the County in running. However, he never fully regained his fitness after diagnosis knowing that at some time or other he was going to need a liver transplant. That transplant happened in 2019 when he was 33 years old. Since then and with Covid permitting, he has engaged the help of a personal trainer to the point at which he competed last summer in Leeds in the British Transplant Games. He represented the King's College Hospital Transplant Team and picked up a bronze medal in the 100m freestyle swimming, a silver medal in the shot put and came 4th in the 100m sprint. He was also a pivotal runner in the 400m relay race.

***Danny with his medals***

Meanwhile, in his professional life he bought an ambulance last year (having spent a good chunk of his life being rushed to hospital in one) and has spent the winter converting it into a mobile waffle business, called Dr Waffle (Instagram *@dr.waffleuk*). Having passed the necessary food hygiene exams, he launched the business earlier this month and so far it promises to be a great success. He specialises in Belgian waffles, chocolate fudge brownies and ice cream made from Jersey cows' milk—with vegan and gluten free options available! He has bookings every weekend for the foreseeable future and will unfortunately miss the Transplant Games in Coventry this year due to taking a pitch at the Secret Island Music Festival on Mersea Island in Essex that weekend.

***"Bel-gian Waffles: What's Your Emergency?"***

And if all that isn't enough, he has also started a clothing/ accessories brand for transplant patients called *Transplant Strong,* selling hoodies, T-shirts, tote bags and water bottles (Instagram *@transplant_strong*). We wish him every success for the future and look forward to watching many more of his inspirational YouTube-*Danny Chat* interviews with Wilson's patients across the world.

* * * * *

**Debbie**, whose patient story appeared in the 2021 newsletter, joined the WDSG-UK committee in 2021. She was diagnosed with Wilson's disease in 1987 and just two months later underwent a liver transplant. She tells us here what she's been up to this past year.

2022 was an *interesting* year for my family and me, encompassing a roller-coaster ride of highs and lows! This was how it went!

**January** saw me recovering from Covid **and** food poisoning.
**February** was spent planning work projects and eagerly anticipating the possibility of travel after two years spent shielding.
**May**, my beloved dog Bernard (a 5 year old Leonberger), who had been my constant companion and home office buddy throughout the pandemic, was diagnosed with lymphoma.
**June**, our business premises were destroyed by fire, along with the beautiful bespoke recording studio for our online floristry learning platform, that had become my second home. And Bernard, passed away.
**August** brought happier times with new business premises up and running, thanks to the determination, hard work and constant support from directors, customers, friends and family.
**September** was the high point of the year, when I celebrated my 35th transplant anniversary. 35 years is represented by coral. However, rather than coral reefs, my husband, mum and I ended the year on a happy note by celebrating for 10 days in Cyprus over Christmas.

***Fire at the 'Triangle Nursery' Premises***


***Debbie, with husband Simon***

Every day I am grateful to the pioneering and dedicated medical staff, my donor family and my own family and friends. 35 years ago, I never expected I would be living life to the full and facing all that is thrown my way with determination and enthusiasm. I am looking forward to the next adventure.

Our regular contributor and intrepid explorer, former committee member **Anne-Marie**, visited the Canary Islands at the beginning of the year. She writes,

## Discovering the two most eastern Canary Islands Fuerteventura and Lanzarote

Back in January my husband Steve and I went on a winter break to the Canaries. The plan was to visit two islands that we hadn't stayed on before and enjoy some warm winter sunshine.

The Canary Islands lie in the Atlantic Ocean to the west of Morocco in Saharan Africa. Although just 100 kilometres from Africa they belong to Spain, 1,100 km away. The hot winds from the Sahara ensure that the islands enjoy a warm climate all year round with temperatures averaging 18°C. Lanzarote and Fuerteventura have the lowest rainfall of all the islands. The islands are volcanic in origin and some of them are still volcanically active.

*Vines grown in the volcanic soil of Lanzarote*

Flying to Fuerteventura, we picked up a hire car and took the short ferry crossing to Lanzarote. We prefer to avoid the crowds and had booked a country house hotel in Yaiza, close to the wine growing area. The interesting building dated back to the 19th century and had been restored in keeping with traditions. Lanzarote offers an amazing lunar landscape. Most of it is covered with solidified lava in tones of black, pink and purple. Vineyards flourish in the fertile volcanic soil and interestingly, semicircles of stones are built to protect the vines from the prevailing winds. We were pleased to discover that, as a result of strict planning controls, the island is almost totally free of high-rise buildings and the trend is to build in the traditional style. This is largely due to the efforts of the late renowned artist and architect César Manrique who was born on Lanzarote and who campaigned to protect the island's natural environment from uncontrolled development. He built an amazing house above and below ground amid the lava flows. We spent the week enjoying dramatic drives through wild terrain, up narrow roads to cliff tops with spectacular views, as well as discovering picturesque fishing villages and quiet coves. This part of our plan lived up to expectations but the weather rather let us down. The temperatures turned out to be below average and it was not only cool but rainy into the bargain and we just wished we'd packed an extra jumper or two!

*Fishermen prepare their boats on a stormy day on Lanzarote*

*The stark, arid countryside of Fuerteventura*

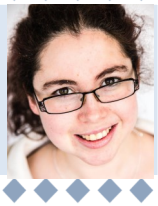
Fuerteventura, despite its proximity to Lanzarote has a very different feel. One of the least populated of the main Canary Islands, it is still developing its tourist industry and we came across several untarmacked roads leading to more remote coves. It is particularly famous for its stretches of white beaches. The name of the island literally means strong wind. It was particularly windy while we were there although thankfully slightly warmer and sunnier than before, and the surf was impressive. We watched plenty of windsurfers, who come in droves to the island throughout the year, taking advantage of the conditions. With accommodation outside the two main tourist resorts limited, we chose to stay at an elegant golf hotel, which was a novelty as neither of us plays golf! The green of the golf course certainly contrasted with the arid natural terrain in the surrounding area.

In spite of the disappointing temperatures and wishing we'd taken more suitable clothing, we were still able to enjoy some sunny days and were very happy to experience the fascinating landscape of these two very special islands.

*The Atlantic coast of Fuerteventura is popular with surfers*

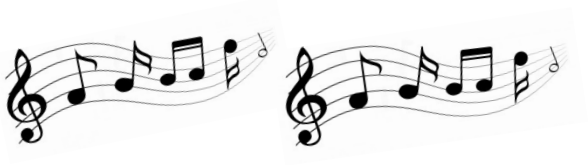
**Anne-Marie Le Cheminant,** March 2023

# Abby's Puzzle Page

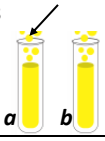


# Euro♥ision 2023

Welcome to Abby's 2nd Puzzle Page. I hope that you will find something here that you enjoy puzzling over. The flags represent those countries appearing in this year's Eurovision Song Contest which will take place in Liverpool between the 9th-13th May. See how many countries you can recognise from the flags and if you can identify those across the bottom of the page (6a-j) . *Answers to the puzzles can be found at the bottom of pages 6-7.*



## 1. Dingbats - Musical Instruments

| | | |
|---|---|---|
| 1  | 2 P&O | 3 LP a |
| 4 eclart | 5 ÷ | 6 *Chair* |
| 7 g g | 8   | 9 basebase |

## 2. Wordsearch - Medium

P U B L B Y O Q R A T F A A G H A B B A
F O N U N X V E G D B G F I I R R E S P
I B S I A S N X N H Q P E F F E Y C R O
A D V K T N W I H C S F N K P L J B R S
A S Z R I E H O O N Q Y I J K L D E W Y
R C C W T J D H V T N C A G A U E E W S
K S O N K O E K L S Q N R U L M C K Y S
M G N W N W U H I Q H A K D U E Y I Q Q
C M T N R T R Z F N H R U A S A M L Y U
X O E J P H O I R A G F M P H M B D F O
F V S L L T P D M U L D I P O N C F L F
G J T J F Z E N U F U S O F R A J L E E
L A V G P O O K R D U C F M C V D I V U
T Z N T T R J L L E D A A K H Z Q V B L
Z O Y N T F P R B Y D P X M E Q Z E U V
S O Q O W A Z R E P F Y J R S X R R X U
N X N G P J N T V U V L R Z T B O P O S
V B U Q Z H X E P Y X N K M R M B O Y S
K Y E K J W D Y G T H I W R A G T O C O
F G L K W U H C O H C G L V O S V L T Y

**Find the following words in the puzzle above and what word/words are left over?**

ABBA SONG EUROPE WINNER CONTEST
UKRAINE LIVERPOOL SAMRYDER MAEMULLER
NULPOINTS GRAHAMNORTON
UNITEDKINGDOM KALUSHORCHESTRA

## 3. Sudoku - Medium

| | | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | | 8 | | 5 | | | 4 | |
| | | | 9 | | | 2 | | |
| | 7 | | | 1 | 4 | | 8 | 5 |
| 2 | | | | | | | 6 | |
| 8 | | 6 | | | | 1 | | 4 |
| | 4 | | | | | | | 8 |
| 7 | 9 | | 5 | 4 | | | 1 | |
| | | 2 | | | 1 | | | |
| | 1 | | | 2 | | 9 | | |

Sudoku puzzle provided by www.sudokuof theday.com

## 4. Anagrams - Hard

**Solve these anagrams of Winning Euro-vision Songs (all English titles).**

1. Evelyn soothing Wolff - 6 words
2. elevating Shiloh - 4 words
3. amid rumping Yukon - 4 words
4. Aloe wort - 1 word
5. call horde hulk rajah - 3 words
6. stepping upon art - 4 words
7. so every famous skiers - 5 words

## 5. Riddles - What are the Answers?

1. What kind of phone makes music?
2. What type of band doesn't play music?
3. I am at the beginning of a phrase, I am at the end of a harp. I appear once in a xylophone, but twice in a set of bagpipes. What am I?
4. What musical instrument can you hear, but not see or touch?

6.a b c d e f g h i j

# WILSON'S DISEASE MULTI-DISCIPLINARY CLINICS

## Adult Clinics

### The Birmingham WD Clinic

**Dr Andrew Holt** (Consultant Hepatologist) and **Dr David Nicholl** (Consultant Neurologist) hold regular one-stop Wilson's disease clinics in the Centre for Rare Diseases at University Hospital Birmingham. 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 should be addressed to Dr Holt's PA, Ms. Maria Del Vecchio (Maria.DelVecchio@uhb.nhs.uk), or Richard Bayley the clinic manager (Richard.Bayley@uhb.nhs.uk).

For older teens under the care of Birmingham Children's Hospital, Ms. Catherine Stewart the Young Adult Nurse Specialist is happy to be contacted and will help you make an appointment in clinic (Catherine.Stewart@uhb.nhs.uk).

### The Cambridge WD Clinic

**Dr Bill Griffiths** (Consultant Hepatologist) and **Dr Paul Worth** (Consultant Neurologist) run a joint clinic every 6 months for patients with neurological manifestations of Wilson's disease at Addenbrooke's Hospital in Cambridge. Dr Griffiths sees a number of patients with hepatic-only disease separately in his adult genetic liver clinic. Both clinics run on Wednesday mornings. Referrals from clinicians can be made to Dr Griffiths at the Liver Unit, Box 210, Cambridge University Hospitals, Hills Road, Cambridge CB2 0QQ.

### The Leeds WD Clinic

**Dr Joanna Moore** (Consultant Hepatologist) and **Dr Jeremy Cosgrove** (Consultant Neurologist) run a joint clinic for patients with Wilson's disease at St James's University Hospital, Leeds. This currently runs on a Thursday morning. Referrals from clinicians can be made to Dr Moore at the Leeds Liver Unit, Merville Building, Beckett Street, Leeds LS9 7TF or Dr Cosgrove at Department of Neurology, F Floor, Martin Wing, Leeds General Infirmary, LS1 3EX.

### The National Hospital Wilson's Clinic, Queen Square, London

A dedicated Wilson's disease clinic has been held at University College Hospitals for the past 30 years. It was set up in 1987 by Dr John Walshe after his retirement from Addenbrooke's Hospital, Cambridge and at the invitation of Dr Gerald Stern. **Dr Godfrey Gillett** started to attend in the mid-1990s and has continued the clinic under the kind auspices of Dr Robin Lachmann at the National Hospital for Neurology and Neurosurgery, Queen Square. The clinic has 70 patients and is held on the third Friday of the month with the close involvement of **Dr Sam Shribman** and **Miss Maggie Burrows**. Referrals from across the UK are welcome, addressed to Dr GT Gillett, Laboratory Medicine, Northern General Hospital, Sheffield Teaching Hospitals NHSFT, Herries Rd, Sheffield, S5 7AU or to Dr GT Gillett, Charles Dent Metabolic Unit, Internal Mailbox 92, NHNN, Queen Square, London WC1N 3BG.

### The Salford WD Clinic

The longstanding neurology consultant led WD Clinic at Salford Royal Hospital has recently been progressed into a joint neurology-hepatology consultant led clinic (**Dr Georgeta Taylor**, Consultant Neurologist and **Dr Jumi Isibor**, Consultant Hepatologist). **Dr Karolina M Stepien**, Consultant in Adult Inherited Metabolic Disorders and her MDT (a dietician and a physiotherapist) review WD patients with hepatic-only manifestations, with the long-term plan to provide a comprehensive support for our patients in a joint neurology/hepatic/metabolic clinic. Referrals from clinicians can be addressed to any of the above mentioned consultants at Salford Royal NHS Foundation Trust, Stott Lane, Salford, M6 8HD.

### The Sheffield WD Clinic

The Sheffield clinic is jointly run by **Prof Oliver Bandmann** (Consultant Neurologist), **Dr Barbara Hoeroldt** (Consultant Hepatologist) and **Dr Godfrey Gillett** (Consultant in Clinical Biochemistry and Inherited Metabolic Disease). Clinics take place every six months at the Royal Hallamshire Hospital, Sheffield on a Tuesday afternoon. Patients may be seen in interim clinics by arrangement. Referrals should be addressed to Prof Oliver Bandmann, Department of Neurology, Royal Hallamshire Hospital, Glossop Road, Sheffield, S10 2JF. Either GPs or hospital specialists may refer to this clinic.

## Children's Clinics

### WD Clinic for Children and Young Adults at King's College Hospital, London

There is a multidisciplinary team Wilson's disease clinic for children and young people at King's College Hospital which is run by **Prof Anil Dhawan** (Paediatric Liver), **Dr Tammy Hedderly** (Paediatric Neurologist), **Dr Jemma Day** (clinical psychologist) and **Ms Bethany Tucker** advanced nurse practitioner (ANP) in children's liver disease. The clinic is directed at patients who are complex with both liver and neurological involvement. Referrals should be made via the Paediatric Liver Centre at King's or enquiries sent to **Lucia DeBiase (lucia.debiase@nhs.net)**, PA to Prof Dhawan.

# A Date for your Diary 2023-24

| Date | Time | Event |
|---|---|---|
| **Sunday, 23 July 2023** | **1130—1530** | **WDSG-UK 13th AGM – Cambridge RUFC, 52 Grantchester Rd, Cambridge CB3 9ED** |

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 :

Wilson's

Wilson's Disease Support Group – UK

www.wilsonsdisease.org.uk

**CONTACTS:**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email: **graemealexander@wilsonsdisease.org.uk**

**Valerie Wheater:** Secretary, Treasurer, Patient Register Coordinator & newsletter editor
email: **val@wilsonsdisease.org.uk**

**Mary Fortune:** Committee Member and Liver Transplant Patient Adviser
email: **maryfortune@wilsonsdisease.org.uk**

**Elizabeth Wood:** Committee Member
email: **lizwood@wilsonsdisease.org.uk**

**Debbie Buckles:** Committee Member
Email: **debbie@wilsonsdisease.org.uk**

**Dr Godfrey Gillett** Group Adviser, Honorary Member
**Dr Caroline Simms** Group Co-Founder
**Linda Hart:** Group Co-Founder
**Rupert Purchase, DPhil** Group Adviser on trientine
**Webmaster:** **Black Cat Websites**


*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 an early diagnosis.

**If more copies of this newsletter are required, please contact Valerie.**