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

NEWSLETTER

#wdsguk APRIL 2024

Hello and a very warm welcome to the 2024 newsletter, which sets out to give you some idea of what the Group has been up to over the past 12 months. One highlight for me was our first face to face gathering and AGM since before the Pandemic, which took place in Cambridge last Summer. It was lovely after all this time to be among so many familiar faces again.

It did, however, mark the end of Graeme Alexander's four year term as chairman of the committee. He has left to enjoy retirement to the full in his new home in Scotland. We thank him for all his support for us, and his hard work in raising the profile of Wilson's disease among the wider medical community. In his place we are delighted to welcome Dr Bill Griffiths, a consultant hepatologist at Addenbrooke's Hospital, Cambridge, and the previous lead of the BASL WD SIG. We look forward to being guided by him in the future.

Although not officially part of the Group, Drs Godfrey Gillett, James Dooley and Sam Shribman continue to offer us their unfailing support. We are so grateful to them for finding time to attend our various meetings, in person and on line, with a special acknowledgment to Dr Gillett for his willingness as ever to advise and help at the drop of a hat.

Getting material for newsletters is always a struggle, so I am grateful to those of you who have responded to my request. In particular, I would like to single out Jane Ridley and James Manning for sharing their patient stories (always a popular read) and Dr Rupert Purchase, who took up our concerns about Vitamin D levels and Wilson's disease and, after researching the subject thoroughly, has written an article based on his findings. Also, thank you to everybody who contributed to the news section at the back and to Abby Morell for kindly conjuring up another bumper page of puzzles for us to ponder over!

Our Facebook Group continues to thrive and now has 1,400 members. It is a private group (so only members can read posts) and is a great resource for keeping up to date with all that's going on. All kinds of topics are discussed with new patients from across the world reaching out for guidance. The current shortage of penicillamine has obviously featured strongly with patients being alerted to the problem as soon as it developed. A new member joined the Fb Group, recently, with the same concerns as the rest of us. But the meds were not for her; they were instead for her Border terrier, Bentley!

On hearing this, I was reminded of an article written by a patient thirteen years ago (at the age of eighty-two) in which he drew our attention to a condition called canine copper toxicosis often found in Bedlington terriers. I have tracked the article down and thought you might be interested in reading it. Not only is it educational, but highly entertaining!

We are often asked about travel insurance for Wilson's disease patients. It seems that premiums are highly inflated no matter what the course of the disease. If anybody has managed to find a good deal recently, perhaps you would care to share it. It would be good to have a list of recommended insurance companies on our website for patients to consult.

Sadly this year, we have lost two prominent members of our Group. Sam Fitzgerald from Northants, who was diagnosed with Wilson's disease in 2014, tragically lost his battle with the illness last August. He was only 23. Sam's smiling face stood out in a crowd and his humour was infectious. We send his mum and siblings our very warmest wishes as they struggle to cope with life without him.

And one of our earliest members, Helen Lothian Khan, also died last year, at the age of 77. She was diagnosed with Wilson's disease back in 1959 when the use of penicillamine was still in its infancy. She was a regular attendee at our meetings and a devotee of Dr Walshe. She will be greatly missed.

Finally, our 14th AGM and gathering has been arranged for Sunday, 7 July, from 1130—15.30. Details are on the enclosed booking form; we very much hope you will be able to join us.

Also enclosed is a subscription renewal form for those who don't pay by standing order. If you could possibly attend to these at your earliest convenience, that would be very helpful. Don't forget if you are paying by bank transfer to either send your completed forms back to me in the post or scan them and return them electronically.

Looking forward to seeing you in the Summer, *Deo Volente* …

Many thanks,

Valerie

IN THIS ISSUE …

Chairman's Report for 2023-24

It was a pleasure and privilege to take on the Chair of the WDSG last summer. I have to thank my predecessor Graeme Alexander for his worthy steering of the Group for several years: a well earned rest there, having served British liver disease admirably. He was my mentor and had great foresight. Importantly, in relation to Wilson's disease, whilst President of BASL (The British Association for the Study of the Liver) he got the Special Interest Groups (SIGs) going and invited me to lead one of the first ones that was set up (the BASL WD SIG). He also met and engaged with Mary Bythell, of what was then Public Health England, a relationship that has blossomed to the forefront of the national Wilson's disease arena.

My foray into Wilson's disease was via a different metal – iron. When I was starting life as a registrar I broke off early to do a PhD on iron transport in haemochromatosis with another esteemed mentor (emeritus) Professor Tim Cox. I started a clinic for patients with haemochromatosis in 2000 and later incorporated other inherited diseases such as alpha-1 antitrypsin deficiency and Wilson's disease. The clinic now includes an ever increasing number of rare inherited liver diseases and such a clinic is quite rare in itself. The Cambridge cohort of WD patients is relatively small and I cannot claim to have the experience of the better known figures in UK Wilson's disease circles but I for one have certainly learnt a lot through running the SIG and attending meetings such as the one in Aarhus in 2022. The Cambridge connection no doubt brought me to Valerie and having just handed over the reins of the BASL rare disease SIG, including WD, it was a natural point to take on this new role.

So now to business and at my first committee meeting in October, and at the subsequent meeting in January this year, we discussed a number of important topics. It had been a plan of WSDG for some time to set up an educational video to raise awareness—particularly in primary care. We now hope to start recording this very soon with the help of Orphalan (manufacturers of Cuprior—a trientine medication). The task is to convey simple messages in relation to early neurological signs, typical psychiatric manifestations and investigation of abnormal liver blood tests in both children and adults. A combination of these facets such as tremor and abnormal liver function is a powerful starting point for performing a caeruloplasmin check. For children and adolescents, behavioural disturbance is an important aspect. We hope that this will lead to earlier testing and referral so that a long delay in diagnosis can be avoided.

Psychiatric manifestations of WD have gained increasing prominence within the SIG meetings and, through the current lead Professor Aftab Ala, we are delighted to have Dr David Okai on board, a consultant neuropsychiatrist at the Maudsley Psychiatric Unit, London. Professor Ala's fellow James Liu Yin is undertaking a PhD on WD which includes work at the Maudsley to see whether Wilson's disease is being and has been routinely considered in patients. We are planning to write up a case series of WD patients with psychiatric presentation which will be helpful for the medical community.

Mary Bythell has sadly returned to the US but James will be working with Jeanette Aston and the team at Rare Disease Registration Service of NHS England to continue the projects that Michelle Camarata and Osob Mohammed had been instrumental in developing as hepatology trainees at the time. James is also helping to set up a national audit of Wilson's disease management with reference to our guidance published in *Lancet Gastroenterology and Hepatology* in 2022. We are grateful to James and his mentor Professor Ala for pursuing important clinical research in WD. I should also mention that Dr Sam Shribman continues his neuroimaging research in WD through The CROWD Study (as reported in previous issues of the newsletter). He is now a consultant at St George's hospital, London, alongside his work and WD clinic that he runs at the UCL National Institute for Neurology, London (Queen Square).

In terms of drugs for WD, there were some shortages of penicillamine at the start of the year which serves as a reminder to patients to try and maintain a 'stock' for such (fairly regular) supply chain issues. A couple of less familiar generic trientine dihydrochloride formulations are hitting the UK market (Tillomed and Waymade). The SIG is evaluating these and there is talk of an NHS England tendering process this year to decide which products are prescribable. Gene therapy trials for WD are underway which could ultimately mean patients will not need to take medication for the rest of their life. Professor Ala has a site set up for each of the Vivet and the Ultragenyx gene therapy trials (see *p 17*).

Between the aforementioned committee meetings was a successful and well attended face to face annual BASL WD SIG meeting in London on the 24th November (*p 16*). Other successful events over the past year include the WDSG-UK AGM on 23rd July (*p 6-7*) and a 'Zoom' coffee morning on 25th February (*p 10*).

On a very sad note, Professor Sir Roy Calne passed away on the 6th January aged 93. Sir Roy was a pioneer of liver transplantation having performed the first successful transplant in Europe in 1968. Many patients owe their

life to his inspirational, courageous and determined work to see liver transplantation the very successful procedure it is today. It was not just the surgical techniques that he pioneered but also immunosuppression where he showed that cyclosporin prevented rejection, a huge advance at the time without which grafts would fail. I was delighted to see (*p 20*) that Joan Smith and WDSG co-founder Caroline Simms are celebrating 20 years of life after liver transplantation this year.

WDSG patients continue to raise funds and awareness and to share their stories as you will see a plenty in this year's newsletter. Once again a *dingbat* challenge — I looked up *dingbat* and it either means a stupid person or feelings of unease - both apply when I try to do it!

I hope you all have a peaceful and happy rest of 2024!

Bill Griffiths 2024

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 again made to **Giuseppe Cardone** who, together with a colleague, raised **£660** through their workplace. We should also like to thank **John Winser's** family for nominating us to be the beneficiary of monies collected in his memory.

We only had one member fundraise this year being **Guy Clarke**. He bought, designed, printed and donated some bespoke WDSG-UK T-shirts for us to sell at our AGM in the summer. This raised £233 (see ***p5***). Should you wish to hold a sponsored fundraising event, then please remember that our online fundraising platform ***GoodHub*** (formerly known as *InvestMyCommunity)* is an easy and efficient way of collecting sponsorship. They are now accepting ApplePay through their website. Their fees are minimal and where appropriate they collect Gift Aid on our behalf.

GoodHub

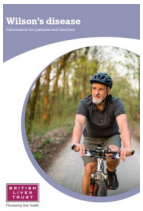
Of course, all donations made to WDSG-UK can be Gift Aided, if appropriate. Gift Aid forms can be downloaded from our website or obtained from Valerie direct.

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. 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.

The British Liver Trust (BLT)

The BLT continues to strive to raise the profile of liver disease in the UK under the direction of its Chief Executive, Pamela Healy, OBE. At the end of October, she, clinicians, patients and NHS England met with parliamentarians and policy makers at Westminster to highlight the current rise in liver cancer in the UK (liver cancer is now the fastest rising cause of cancer death in the UK). BLT is pressing for the Government to keep its promise to provide FibroScans (non-invasive machines for detecting liver disease) in 100 Community Diagnostic Centres in England. These should offer earlier diagnosis of liver disease with subsequent improved outcomes for patients. This also has the potential of picking up patients with Wilson's disease early.



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/uploadsWilsonsDiseaseweb DEC.21.pdf or alternatively a single hard copy can be ordered free by emailing info@britishlivertrust.org.uk.

Children's Liver Disease Foundation (CLDF) and BLT

It was announced at the beginning of the year that on 31 March 2024 the BLT will be merging with the CLDF (which is based in Birmingham and has several children with Wilson's disease on its books). By merging, "the aim is to ensure that all liver disease patients receive the essential treatment, support and care that they need and deserve."

NHS BT (Blood & Transplant) Organ and Tissue Donation & Transplantation Directorate (ODT) and the Liver Patients Groups (LPG) and Liver Disease Charities' Transplant Consortium (LPTC)

On 4 July 2023 Valerie attended the 14th NHSBT ODT meeting with the LPTC via *Microsoft Teams*. An overview was given of service provision among the 7 Liver Transplant (Tx) centres in the UK. The ODT continues to strive to give patients the best care from referral to transplantation. Liver perfusion is going well and the liver Tx community has embraced it. The Living Donor programme is expected to be sanctioned by NHS England shortly. Statistical data were given on the liver Tx programme in the UK over the previous 12 months. Of particular note there were 10 liver Txs carried out on patients with Wilson's disease between 2020 and 2023 (inclusive) —2 more than there were in the preceding 4 year period 2016-2019. For more information, please visit *Organ specific reports—ODT Clinical—NHS Blood and Transplant.*

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

CRDN Cambridge Rare Disease Network

by rare conditions. To this end patient groups, scientists, researchers, clinicians, pharma and biotech companies locally and from around the world are brought together.

New committee member Claire Stapleton and Valerie attended the 7th RAREsummit on 12 October 2023 at the Wellcome Genome Campus outside Cambridge. It was a lively event which included talks, short films, panel discussions and exhibitions. For a more detailed overview of the day, please refer to the report on the CRDN website: RS23_ImpactReport_Compressed_Email_SocialMediaShare.pdf - Google Drive

The Wilson's Registry—NHS England—NCARDRS

NCARDRS (The National Congenital Anomaly and Rare Disease Registration Service) was set up in 2015 by Public Health England (PHE) with the aim of improving the management and care of patients living with congenital abnormalities and rare diseases in England. Out of 7,000 rare diseases, Wilson's disease was chosen to be used in their pilot study and Mary Bythell of PHE and Dr Osob Mohammed worked with the BASL WD SIG to procure the relevant data. It was an onerous task using sources such as hospital episode statistics (i.e. records of patients' hospital attendances at A&E, admissions and outpatient appointments), prescription records and death certificates where WD had been mentioned. The information acquired gives an idea of prevalence of WD in England (thought to be around 600), prescription choices, patient management and outcomes. One aspect of patient outcomes that is being monitored at the moment is the incidence of hepatocellular carcinoma in patients with liver involvement.

After 6 years of working on the project, Mary Bythell has moved back to the USA but Dr James Liu Yin has been contracted to work alongside the team on the Registry for the next 12 months.

EASL (The European Association of the Study of the Liver) — a forum for research hepatologists)

In 2022, following the publication of the BASL WD SIG's Wilson's Disease Guidance document *The Management and Treatment of Wilson's Disease,* EASL joined forces with ERN (The European References Network—Rare Liver Diseases) to produce a European guidance document of their own. This is still in the consultation stage and Valerie, by invitation, is involved in giving feedback on its content on behalf of patients.

The next EASL Congress will take place 5th–8th June in Milan and Bill Griffiths will be giving a poster presentation on Drug Adherence in Wilson's disease patients. Thank you to everybody who took the time to fill in the short questionnaire relating to this which was advertised on our Fb site and circulated to patients via email.

Wilson Aarhus 2024 Symposium— 2-5 May 2024

This event is held every two years and the 3rd Symposium will take place in Aarhus, Denmark, in May this year. Hosted by Danish WD specialists Prof. Peter Ott and Thomas Sandahl, the event attracts specialists from across the globe. Dr Sam Shribman and Prof. Oliver Bandmann from the UK will be presenting papers. Claire and Valerie will represent WDSG-UK and other patient representatives from France, Germany, Spain, Italy and the USA are expected to attend.

International Community of Wilson Disease Associations

This is a What's App Group that has been set up by the Argentinian WD Support Group and includes committee members from 11 Support Groups from South America, Latin America, the USA and Europe. Claire has joined recently on behalf of WDSG-UK. If there is enough interest among participants, it is proposed that a World Wilson's Disease Day be recognised by the World Health Organization (WHO) and other International Health Organizations. The date suggested is December 6th (the birthday of Samuel Kinnier Wilson who first described Wilson's disease in patients in 1912). What are your thoughts on this?

Hong Kong WD Association (HKWDA) and WDSG-UK

The committee of the HK Support Group has arranged a Zoom call between the two Groups for Saturday, 27 April 2024 between 0930 and 11.30 (London time). Please let Valerie know if you would be interested in attending.

WDSG-UK Management Committee Meetings

During 2023-24 the management committee met three times—in May, October and January, via Zoom.

WDSG-UK—Rare Disease Day Zoom Coffee Morning

On Sunday, 25 February 2024 WDSG-UK held its fourth Zoom Coffee Morning to mark Rare Disease Day. For a full report see *p10.*

WDSG-UK Annual Meeting and 13th AGM

For the first time since 2019, WDSG-UK's annual meeting and AGM took place in Cambridge last July. It was tinged with sadness as this was the first time in the history of the Group that the event was held without our President and closest supporter, Dr John Walshe, being present. As most of you will know, he died at the end of 2022 aged 102.

The meeting took place at our usual venue, Cambridge RUFC, on Sunday, 23rd July 2023. The accounts for 2022-23 and the minutes of the 12th AGM were circulated and agreed and the election of officers and members of the WDSG-UK Management Committee for 2023-24 took place. **Bill Griffiths, Mary Fortune, Liz Wood, Debbie Buckles, Claire Stapleton** and **Valerie Wheater** were duly elected on to the committee for 2023-24**.** For more details of the day please see the report on *pp 6-7.* If you would like to join the committee, then please write to me at val@wilsonsdisease.org.uk.

Fundraising 2023-24

This past year is the first year since the Group gained charitable status in 2010 that there haven't been any sponsored events to report. I have to say that I've always been amazed at the variety of challenges undertaken by members and sometimes by people previously unknown to the Group. The latter cohort often has friends who have been badly affected by Wilson's disease or close family members who have died of or with it.

Some of the sponsored events already undertaken include marathons, half-marathons and triathlons; walks and cycle rides; mountain ascents, skydives and swimming events; and golf competitions, football matches and boxing rounds. Should you have any ideas for future sponsored fundraising, please let me know. Our fundraising platform *GoodHub* is a useful resource.

In addition, over the years people have raised funds by holding coffee mornings, tea parties and bring and buy sales; car boot sales, quiz nights and movie nights; concerts; baking and selling cakes, jams, chutneys and Christmas puddings, making and selling handicrafts such as greetings cards, pottery pieces and painted pebbles; and not least running raffles, selling wristbands and flogging Dingbats! Below is the most recent innovative fundraising idea...

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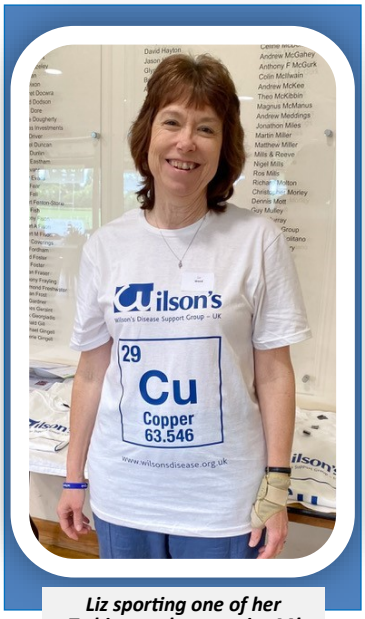
Guy's Own Design and Printing of WDSG—UK T-Shirts

Guy Clarke joined the Group in 2022 after being diagnosed with WD at the age of 39. His patient story appeared in the 2023 newsletter where he tells us that a fluke accident to his eye at Christmas '21 resulted in a diagnosis of Wilson's disease. Prior to that, he had been seeking medical help because of his symptoms for the previous 18 months but had got no satisfactory answers.

Throughout his WD journey Guy luckily has been able to continue working in his printing business in Slough (Vario Press). The company specialises in 'Creating, Printing and Digital Marketing', though not normally T-shirts!

When last year's fundraisers, Phil and Kurt (friends of Ben Ryan—see *p 19*) set out to tackle the 24 hr National 3 Peaks' Challenge, they wanted to wear WDSG-UK branded T-shirts for their venture. We did once commission our own T-shirts, but not any more. Guy very kindly offered to help out and bought a special printing machine to do the job. Rohan donated the T-shirts, but the design was his own and the finished items were impressive.



Liz sporting one of her T-shirt purchases—size M!

Although he couldn't make the AGM himself in July, Guy generously printed and donated a further 20 bespoke T-shirts to be sold there. They came in 3 sizes, were a different colour from the originals and Rohan didn't supply them this time! A minimum donation of £10 was asked for each. At the end of the meeting only one remained and **£233** was raised. Many thanks for this, Guy.

Should anybody wish to buy the remaining T-Shirt (size S), please let me know. It will be sold to the highest bidder!

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A few *shout-outs* here to people who have made special donations this year.

First of all, may I thank Dr Walshe's two daughters, **Susan** and **Clare,** who when clearing out the family home last year after their father died held a Yard Sale and generously donated half the proceeds to us.

Thank you to everybody who contributed raffle prizes to the AGM last summer—not only members but their friends, too. The raffle was a huge success and raised a total of **£148**.

Next, I'd like to congratulate **Sue Boysons** and her husband, **Chris**, for their quiz victory at the retirement complex near to where they live and where Sue's mum has recently relocated. They kindly donated their winnings to us.

And finally, I would like to thank again all those who made generous donations with their subscriptions, in particular **Joan Smith** who makes a donation every year which matches her age in £. She was 13 when she was diagnosed in 1963, so the amount is now considerable!

WDSG-UK Meeting & 13th AGM
Cambridge, Sunday 23 July 2023

After four long years of only meeting on line, our 13th AGM was finally held in person again at the Cambridge Rugby Union Football Club, just a stone's throw away from junction 12 of the M11 motorway with ample free parking and extensive facilities.

July hadn't been a great month weatherwise, but we were lucky that the rain managed to hold off for the day and the wind subsided allowing the meeting to go ahead in relative calm. From a slow uptake of places when invitations were first sent out to a sudden flurry of bookings towards the end, we were delighted to greet around 50 members, several of whom had never attended the meeting before.

Arriving from as far afield as Northern Ireland, attendees were welcomed with refreshments and encouraged to mingle before settling down for the day. Old friendships were renewed and new ones made. Our chairman, Graeme Alexander, who now lives in the far reaches of Scotland (see *p18*), was unfortunately unable to join us and so vice-chair and Zoom host, Liz Wood, ably conducted the day's proceedings in his place.

She started by formally welcoming everybody, paying particular thanks to Drs James Dooley, Godfrey Gillett, Sam Shribman and Miss Maggie Burrows for giving up their valuable time to join us. Then followed the 13th AGM in which Liz read a report prepared by Graeme detailing the achievements of the committee over the past twelve months. She spoke about the success of the Zoom coffee mornings that we have been holding since the beginning of the Pandemic and gave updates on the matters raised at the last AGM. Accounts were circulated to members in advance and were available to be inspected at the meeting. With no questions about them forthcoming, matters quickly moved on. Finally, the meeting concluded with committee members for 2023-24 being proposed, seconded and duly elected (listed on *p4*).

This was the first face to face AGM that new committee members Debbie Buckles and Claire Stapleton had attended and they, together with their mums, threw themselves into the spirit of the occasion by running the raffle and selling the WDSG-UK T-shirts which had been donated, designed and printed by Guy Clarke. In all a total of £381 was raised. As a florist with 45 years' experience in the industry, Debbie had also created the magnificent floral centre pieces for each of the tables, which were a great talking point and highly appreciated.

Our speaker, Peter Bull, who was part of the team which identified the Wilson's disease ATP7B gene in Toronto in Canada in the early 1980s, succumbed to an annoying cough at the end of a bad cold and asked if he could postpone speaking until our meeting next year. This therefore left a gap in the pro-

Making their introductions

The Doctors answering our questions

Discussing their concerns with Dr Gillett

Jess, Peter, Chris and Sue

gramme which we decided to fill by holding an extended and more formal Q & A session with the doctors than usual. What, remissly, we forgot to do was tell the doctors in advance, but luckily they agreed to take part and did so with great resilience and good humour!

The questions that were put to them were generated by patients and the topics covered included the importance of WD patients keeping their Vitamin D levels up, the reason why zinc is not used as a monotherapy as much in the UK as in other countries and why it is that many Wilson's disease patients are unable to smell noxious gases such as bad eggs and flatulence!

We thank Dr Rupert Purchase, chemist and former chairman of WDSG-UK, for also joining the panel and giving us his take on the storage of Cufence. He has since researched the subject of Vitamin D and Wilson's disease and has written an article about it which can be found on *p11*.

A splendid buffet lunch was provided by the Club's in-house caterers over which members were able to continue to chat, pour over the commemorative book presented to Dr Walshe in 2020 on his 100th birthday, and buy raffle tickets and T-shirts if they felt so inclined. They were also able to speak informally to the doctors about any personal health issues which might be of concern.

Our usual photographer, member Anusha Joseph, was unable to make the meeting, so we were grateful to Steve, husband of former committee member Anne-Marie Le Cheminant, for taking over Anusha's role and producing so many wonderful photographs of the day.

The meeting finished promptly at 1530, after which there was a final group photo for any patients who had stayed the course! We look forward to hosting another AGM in person this year and have booked the same venue for Sunday, 7 July 2024 (details and booking form enclosed).

* * *

Anne-Marie and Gloria

Preparing the Raffle Draw

Claire and her mum, Helen

Our customary Group Photo

Jane's Story

by Jane Ridley

Hello, I am Jane and my Wilson's disease journey started back in 1978 when I was 20 years old.

I was living in South Lincolnshire where I was brought up, in a small village outside Holbeach. The parish of Holbeach is one of the largest and least populated parishes in England and nowadays is a popular destination in springtime for tourists visiting the tulip fields.

I was an only child (the reason being my mother suffered morning sickness all the time she was carrying me and vowed never to get pregnant again!) I had a happy childhood and my school life was unremarkable. I left school at the age of 16 to work in a department store nearby. I was a sales assistant there and often worked on the tills. I passed my driving test when I was 17 and was married by the age of 20. My husband and I had our own house and life seemed to be going well.

I had many friends from school and an active social life. One day in 1978 we travelled to London to attend a recording of *Top of the Pops*! I had eaten a packet of peanuts and got a piece stuck in my tooth. When I tried to dislodge it with my tongue, my jaw locked on one side and I couldn't talk or move my mouth. Things didn't improve so my friends took me to A&E where three hours later doctors managed to reposition it. Thinking it was an unfortunate accident, we continued to the show spending the night at a hotel in London with no further problems!

"...They decided it was all in my mind and I should pull myself together."

However, in the following days and weeks, I started to notice I was dribbling and slurring my speech which was embarrassing particularly at work. Customers used to stare and snigger. I went to see my GP and he gave me some tablets to control it. At first they seemed to work but not for long. Eventually, I was admitted to the *Pilgrim Hospital* in Boston, a community hospital where they ran tests. After 10 days as an inpatient, I was discharged as they couldn't find anything the matter with me. They decided it was all in my mind and said I should "pull myself together!"

My mother wasn't happy about this and asked for a second opinion, so my GP arranged for me to see a psychiatrist in Spalding—9 miles away. This time I was given injections in my bottom to dry up the saliva and although they worked for a short time, by the time I saw him a month later I would be dribbling badly again. After six months the psychiatrist finally declared "You baffle me; I haven't got a clue what's wrong with you."

Luckily for me he didn't leave it there and referred me to Dr Yealland, a neurologist at Addenbrooke's Hospital in Cambridge. I had to wait two months to be seen by him. It was the 14th April 1979. At my appointment, Dr Yealland (who was a colleague and contemporary of Dr Walshe's) immediately recognised my symptoms and did a blood test on me. He told my parents to bring me back later that day for the results. When we returned, he told us that he knew exactly what the matter was and that I had a very rare condition called Wilson's disease. Of course, we'd never heard of that before. He then arranged for me to be admitted to Addenbrooke's under the care of Dr Walshe, the world authority on the disease.

"... on hearing this Dr Walshe, or his assistant Kay, made a point of coming to the ward at mealtimes ...

It was a turbulent time. My husband had walked out on me and my friends deserted me, so I only had my parents for support. I was in and out of hospital for the next six months. To begin with, everybody thought I was going to die. I was so weak I could hardly walk and had to be pushed around in a wheelchair. I had lost a lot of weight and was barely five stone. I was drooling more than ever and my speech was slurred. Nobody could understand what I was saying. Dr Walshe lent me a machine to type on so I could write down what I wanted to say. My days were filled having blood tests and collecting urine samples, with trips in between to various departments including radiology for brain and liver scans and ophthalmology, where the ophthalmologist got very excited about the "lovely" Kayser-Fleischer rings he found in my eyes!

During this time, my left hand had slowly begun to contract with my fingers and knuckles bending over towards the palm of my hand. My wrist had twisted round and I could only use my thumb. It was very painful and made holding cutlery and cutting up my food very difficult. The matron on the Ward insisted I do it myself or I'd "never get better." On hearing this Dr Walshe, or his assistant Kay, made a point of coming to the ward at mealtimes to cut up my food or take the clingfilm off my sandwiches for me!

After about six weeks under Dr Walshe's care, I started to improve slowly and was transferred to the Rehab Unit on the Addenbrooke's site. I was told I would have to stay there until I was well enough to look after myself and go home. I was given speech therapy. I also had physiotherapy on my legs and hand. I was soon allowed to go home for weekends but it took several more weeks before I was discharged altogether. By then my speech was improving, I had put on weight and I was able to slowly walk again.

It took a further year for me to get back to a new kind of 'normal', but sadly my left hand made no improvement at all. My parents had taken a lot of time off work while I was ill and needed to go back full-time. As I couldn't wholly look after myself in the early days, I relied on Home Help volunteers to pop in to help and keep an eye on me. Dr Walshe continued to see me at regular intervals and I would stay in hospital for tests for a week at a time. When he retired from Addenbrooke's in 1987, I followed him to his new clinic at the Middlesex Hospital in London.

That same year at the age of 30, I married again. My husband lived and worked in London which is where we set up home welcoming the birth of our daughter, Tanya, the following year. We remained living in London for the next ten years before buying our first property together in Peterborough, nearer to my family.

A couple of years after diagnosis I had an operation on my hand whereby they immobilised my wrist to stop it from getting any worse. Although I have adapted to this disability in all other ways, I am sad not to have been able to drive again.

When I was first diagnosed with Wilson's disease I was prescribed trientine. However, it caused me to be anaemic and after two years and a blood transfusion, Dr Walshe switched me over to penicillamine. I have been on that pretty much ever since.

Like a lot of patients on long term chelation therapy, I have had trouble with my skin. It has caused *cutis laxa (*which is where the skin is loose in folds) and is particularly noticeable around my neck. The other problem is a skin condition called Elastosis Perforans Serpiginosa (EPS) which affects 1 in a 100 patients on long term penicillamine use. It is thought to have a genetic base. It appeared on the back of my neck in 2015 and presented as small nodules under the skin which then crusted over and erupted. I had them removed at the hospital in Peterborough which caused extensive scarring, but after later treatment at Addenbrooke's (where the skin was heated and then frozen), they cleared up altogether. Since presenting with EPS, I was advised to go on a slightly reduced dose of penicillamine.

A detailed article about EPS written by the late Dr Alan Stevens appeared in the 2017 newsletter and shows a photograph of my neck when EPS first presented. The article can be downloaded from the WDSG-UK website and is certainly worth a read:

82280c41-69a8-4c6f-ae63-2ef3fbcf9490_WDSG_UK_website___AS_Elastosis_Perforans_Serpiginosa___2017.pdf (wilsonsdisease.org.uk)

I am 66 now and was widowed two years ago. Although I live on my own I am fortunate enough to have Tanya and my two grandchildren (aged 7 and 4) living round the corner. I joined WDSG-UK when it was set up in 2000 and have attended nearly all their meetings since. Through them, I have made friends with other patients along the way. I am grateful to Dr Walshe for all that he did for me and to my current doctors who continue to look after me today.

* * * * *

In 1984 an article appeared in *The Guardian* newspaper featuring a 28 year old man from nearby Spalding, who had recently been diagnosed with Wilson's disease. He had severe neurological symptoms and recovery was proving slow. On hearing about him, Jane contacted their local paper as she wanted to reach out to this man and let him know that recovery was still possible.

A reporter was sent round to interview her and an article was later published. Here is a photo of the original article headlined, "Jane's Brave Offer of Hope."

Meanwhile, Dr Walshe was excited to have a "cluster" of patients from the same area in Lincs. (there were now three of them). He and Kay spent time researching their family histories in the hope of finding they had a common ancestor. Despite getting back as far as Nelson's time, they didn't find any connection at all. Annoyingly, in the end they had to abandon looking!


JANE'S BRAVE OFFER OF HOPE

A STORY OF COURAGE

WDSG-UK Rare Disease Day *Coffee Morning*
Sunday, 25 February 2024

Some of the Attendees at The Coffee Morning marking Rare Disease Day 2024

This is the fourth such consecutive Zoom meeting WDSG-UK has held to mark a Rare Disease Day. The first Rare Disease Day was introduced by the European Organization for Rare Diseases and took place globally in 2008 — on 29th February — a fitting day to celebrate something rare. Although this year is indeed a Leap Year, generally speaking it isn't, and so Rare Disease Day is then celebrated on 28th February instead.

Wilson's disease is only one of around 7,000 different rare diseases. The general objective of marking the occasion is to raise awareness amongst the general public and policy-makers of the impact that living with a rare disease has on patients and their families. Events such as social gatherings, fundraisings, art exhibitions, workshops and press conferences are held across the UK, and parliamentary lobbying is encouraged. Amongst our diverse community, mutual support is of primary importance and therefore coming together over a morning cup of coffee on the last Sunday in February seems fitting for us.

Hosting a virtual event like this is challenging, but Liz Wood is well practised in the art**.** During lockdown and Covid restrictions, she ran various choirs and music events over the internet from home. She took charge of the day's programme with great aplomb and welcomed 28 people to it. Attendance was by invitation only and was restricted to paid-up members of the Group.

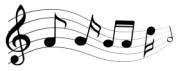
Wilson's disease specialists Drs Godfrey Gillett, James Dooley and Sam Shribman also attended and we would formally like to thank them here for so generously allocating time at the weekend to join us.

Once everybody had *signed in* and said "Hello", we were divided into two separate *break out rooms*, each with a doctor and committee member present. This allowed individuals a platform to ask questions and share their concerns within a smaller group. After 20 minutes we reconvened. We were asked if we would like to be randomly allocated to a different *chat room* for the remainder of the session or return to our original rooms. The general consensus was to remain with the familiar—so back we went!

Topics that were discussed included the recent difficulty patients experienced obtaining their penicillamine medication. It was explained that this has happened before at this time of year and is probably due to funding procedures for rare disease meds within the NHS. The advice to patients was to make sure that they always held plenty of stock by ordering their tablets well before supplies ran out, particularly in the run up to Christmas and the New Year. Patients were also advised to contact their hospital specialist if they ran into difficulties rather than to chase round other community pharmacies looking for it. Hospital pharmacies are better placed to find out where the problem lies and to procure supplies from a variety of different wholesalers.

Other topics discussed included patients being seen by WD specialists at specialist centres, liver transplantation among WD patients and how gene therapy trials were progressing in the UK. Finally, a suggestion was made that singing therapy could be advantageous for patients whose voices were affected by Wilson's disease. It was noted that patients with Parkinson's disease already undertake this practice. Wilson's patients were advised that exercise in general was greatly beneficial and singing in the shower wouldn't do anybody any harm!

On that note, we sang our *farewells* and the meeting was duly drawn to a close.



Vitamin D and the Wilson's disease Patient

Introduction: Vitamin D

We need vitamin D for healthy bones, teeth and muscles. Vitamin D is a fat-soluble vitamin which we can absorb from a select number of foods (egg yolk, oily fish – cod liver oil – for example). Vitamin D is also synthesised naturally under our skin by the action of sunlight on a derivative of cholesterol, which is present in our bodies. In the summer, there should be enough sunlight to maintain the required level of vitamin D, but in the autumn and winter in the UK we are advised to take a vitamin D supplement in the form of a tablet in order to secure the correct amount of vitamin D for our health. Currently for the general population, the UK Department of Health and Social Care recommends taking a daily supplement of 10 micrograms of vitamin D throughout the year, particularly for those who are not often outdoors. However, for particular ethnic sub-groups in the UK, higher daily doses of vitamin D are advised. Some medical practitioners suggest extending these higher daily doses to the general population.

Vitamin D – too little and too much!

Too little vitamin D can lead to bone deformities such as rickets in children, and bone pain caused by a condition called osteomalacia in adults. Joan Smith told us about her joint problems in last year's (April 2023) WDSG-UK newsletter (*p 17*), which may have been caused in part by being indoors during the Covid pandemic. Joan is now on a course of vitamin D tablets.

As with any food supplement, too much vitamin D may be just as harmful as too little. Consuming too many vitamin D supplements in the long-term or intermittently can cause elevated amounts of calcium in the body (hypercalcaemia). This can weaken the bones and damage the kidneys and the heart. Opinion about an upper safe limit for vitamin D supplementation has shifted in the last few years, and a dose not exceeding 20 – 25 micrograms per day is now considered safe.

Vitamin D – What does it do and how does it work?

Among many other attributed health benefits, vitamin D maintains bone strength. It does this by regulating calcium and phosphate levels in the body. The correct levels of calcium and phosphate are necessary for bone and cartilage formation. The actual form of vitamin D responsible for calcium and phosphate regulation is produced in two stages. The first stage takes place in the liver (to give the metabolite 25-hydroxyvitamin D) and the second stage in the kidneys to give the 'active metabolite' of vitamin D, 1,25-dihydroxyvitamin D. Impairment of this two-step metabolism of vitamin D may have consequences for calcium and phosphate regulation and therefore for bone development.

Wilson's disease patients and joint problems

Although skeletal changes were recognised in Wilson's disease patients by the early 1970s, a clear explanation of why these occur has perhaps not been forthcoming. Too often penicillamine or the consequences of an excess of copper as a result of Wilson's disease have been identified as the culprits. Relative to the other symptoms of Wilson's disease, joint issues are somewhat overlooked. However, concerns about joints are often mentioned at our WDSG-UK meetings by patients and their families, and it is clear that further work in this area would benefit the Wilson's disease community. Application of current vitamin D research offers a way forward for understanding joint problems in WD.

Wilson's disease and vitamin D metabolism: Further research

Enzymes in the liver and kidneys are responsible for the metabolism of vitamin D. Wilson's disease can cause both liver and kidney injury and this might decrease the activity of the drug-metabolising enzymes. Any impairment of vitamin D metabolism (i.e. reduced endogenous synthesis of the two metabolites, 25-hydroxyvitamin D and 1,25-dihydroxyvitamin D) as a consequence of liver and/or kidney injury in Wilson's disease patients risks the occurrence of bone disorders. In order to check the status of vitamin D metabolism in patients with Wilson's disease, it might be worth considering the following checks on a small group of patients:

1. Monitoring their vitamin D levels.
2. Measuring the levels of the two vitamin D metabolites, 25-hydroxy- vitamin D and 1,25-dihydroxyvitamin D.
3. There is evidence that the vitamin D metabolite 25-hydroxyvitamin D is better absorbed than vitamin D and potentially more potent. Trials could be conducted to compare the relative benefits of 25-hydroxy-vitamin D and vitamin D in Wilson's disease patients.

In the longer term, it would be helpful to reexamine drug metabolising cytochrome enzymes in Wilson's disease patients.

Conclusion

Vitamin D is an important part of daily nutrition. This note has highlighted some aspects of vitamin D metabolism and Wilson's disease. A more detailed version with literature references appears on the WDSG-UK website.

Helpful weblinks on vitamin D

- United Kingdom: Vitamin D - NHS (www.nhs.uk)
- USA: Vitamin D - Health Professional Fact Sheet (nih.gov)

Rupert Purchase, February 2024

29
Cu
Copper
63,546

Spare Us Copper

By Aubrey Morris

COPPER: — "Who needs it?" you may well ask. Well, actually all of us do, in minute quantities. Our problem, of course, is that we have too much of the stuff. But, if we had none at all, we'd be in even deeper trouble. The reverse of Wilson's disease (almost) is called Menkes disease. I say "almost" because, while sufferers have insufficient copper in their blood plasma, liver and brain, they have overload in their kidney, spleen and skeletal muscle, so it is a very perverse illness indeed. As with Wilson's, it is inherited, but, unlike Wilson's, its symptoms appear very quickly - when one is just a baby of two or three months - and they are dire: muscles become weak and floppy, baby has seizures, can't hold a rattle, his cheeks sag and his scalp hair becomes coarse and twisted. Because of this last symptom, Menkes is familiarly known as "kinky hair disease." Kinky hair or rings in our eyes - we all have distinctive symptoms arising from our copper imbalance. So what else do we know about this metal which looms so large in our lives?

Courtesy of Wikimedia Commons

Copper is a moderately hard reddish-brown metallic element which occurs both as the free metal and in various ores, the most notable of which is chalcopirite, otherwise known as copper pyrites. Its chemical symbol is Cu, short for the Latin *cuprum*, which is derived from the Latin name for Cyprus, the principal source of the metal in Roman times. Nowadays, it is mined in Chile, New Guinea, Australia, Zaire and Zambia. It is Zambia's biggest export, and the region of Central Africa along the Zambia/Zaire border is known as the Copper Belt. Copper can be hammered into shape without breaking, but its chief uses are as an electrical and thermal conductor. It melts at 1083°C and boils at 2595°C. Thus much is common knowledge. Here are a few random facts that are more esoteric.

- Copper is deadly to dangerous bugs! In a trial study at Selly Oak Hospital, Birmingham, a set of taps, a lavatory seat and a push plate on an entrance door were replaced with copper versions. The copper items had up to 95 per cent fewer bugs on their surface than traditional fittings. Lab tests showed that copper kills MRSA and C difficile superbugs as well as the flu virus and the E coli food poisoning bug. Scientists conjecture that the metal prevents germs from breathing and possibly from feeding.

- Copper in the soil is an essential nutrient needed for the normal growth and development of cereal crops. Chlorophyll production, protein synthesis and respiration are important plant functions that require it. About 70% of the copper in plants is found in chlorophyll. Copper deficiency can result in early ageing or lower levels of chlorophyll, which leads to yield reduction. Peat soils are prone to copper deficiency.

- Average broadband speeds in the UK are only the 37th fastest in the world - slower than Latvia and Romania - because our infrastructure relies on ageing copper wiring, whereas other countries have skipped straight to fibre-optic connections.*

A Bedlington terrier

- Bedlington terriers can be born with Wilson's disease! Well, not exactly - but an article in the Journal of Experimental Animal Science in April 2002 stated that "Canine copper toxicosis is an important inherited disease in Bedlington terriers because of the high prevalence rate and similarity to human copper storage disease." However, it continued: "The responsible gene for copper toxicosis in Bedlington terriers has recently been identified and was found not to be related to the human Wilson's disease gene." Although copper toxicosis is commonest in Bedlington terriers, dogs in general have copper storage problems more rarely and many breeds are affected. Labrador retrievers, for instance, can get copper-associated hepatitis. In Bedlington terriers tetramine is a safe and rapid chelating agent. No research appears to have been done on using this substance on humans.

- Copper was possibly the first metal to be used by Man, and the Bronze Age was from about 4000 to 1000 BC. It came between the Stone Age and the Iron Age.

- The price of copper fell back a little from a record high of $8,966 per tonne reached on 11 November 2010, but then took off again when a mystery buyer, believed to be a global hedge fund, started buying steadily. By early December "the fund" had acquired more than 280,000 tonnes, amounting to 80% of the copper market.

- Brass is an alloy of copper and zinc. The proportions are variable, but using more than 36% zinc results in a stronger alloy. Bronze is an alloy of copper and tin, and again the proportions are variable - tin may account for between 4% and 11%. "Copper" coins are not copper but bronze consisting of 95% copper, 4% tin and 1% zinc.

• Someone with a good tan is described as "bronzed" but the makers of a popular suntan oil chose to name their product *Coppertone*.

• The Daily Mail recently advised its female readers on how to create a copper eyeshadow look. "Take a medium soft brush and, using a bronze shade, start from the inner eyelid and sweep across right up to under the eyebrow arch …"

• In 1940, George Formby (No. 1 at the British cinema box office in 1939) starred in a film called *Spare A Copper*. This title was a pun, because George took the role of a policeman. George never played a ukulele, as is commonly supposed. Anyone who has seen the Ukulele Orchestra Of Great Britain (highly recommended!) will know that a ukulele is shaped like a guitar. George's instrument was shaped like a banjo and was in fact a hybrid called a banjolele. George encouraged the misconception with songs like *With My Little Ukulele In My Hand … My heart it leapt with joy, I could tell it was a boy, cos he'd his little ukulele in his hand*.

• A *copper* in the sense of a policeman has nothing to do with the metal but is simply one who cops (catches). This pair of words — "cop" and "catch" — is unusual in that they are synonyms in two senses. Both also mean "to receive punishment" ("You won't half cop it when your father gets home").

• Newspaper theatre critics are fond of the phrase "a copper-bottomed hit" when they wish to forecast guaranteed success for a show. The expression is of maritime origin and refers to the period when the leaky timbers of the hulls of sea-going vessels gave way to riveted copper plates. The ships were copper-bottomed — guaranteed safe. Copperplate handwriting derives from the perfect script used when it was engraved upon a copper plate and a print was taken therefrom. An old tongue-twister runs: "Are you allumining 'em ma'am?" "No, I'm copper-bottoming 'em my man."

• In December 2010, an 1827 book *Birds Of America* by wildlife artist John James Audubon became the most expensive printed book in the world when it was sold by auctioneers *Sotheby's* for £7,321,250.00. Measuring 40 inches in height and containing life-size illustrations of nearly 500 breeds, it was printed from copper plates, a process which would cost £1,250,000 in today's money.**

• Copper is just one of several metallic elements that are essential to human wellbeing. Most notable of these is iron — so much so that the makers of most breakfast cereals boast that their product is iron-fortified. Iron is essential to the production of haemoglobin (red blood cells) which transports oxygen in the blood from the lungs to the body's tissues. Magnesium keeps heart rhythm steady, supports the immune system and helps strengthen bones. Zinc also boosts the immune system, promotes wound healing and is vital to reproductive health. Chromium works with insulin in the metabolism of sugar and stabilises blood sugar levels. And copper? Well, copper works with vitamin C to form elastin, a component of muscle fibre throughout the body. It helps to maintain healthy bones and assists the thyroid gland in balancing and secreting hormones. And — guess what? — the much-vaunted iron wouldn't be much use without copper because it actually combines with copper and the protein globin in the creation of haemoglobin.

• So copper is *A Good Thing*. But as we know to our cost, you can have too much of a good thing. You can have too much iron as well. The disease is called haemochromatosis and, like Wilson's, it is inherited. It is also like Wilson's in that the excess builds up gradually, so that the symptoms don't show until it has already damaged the organs where it has accumulated. Thus, though we need copper, iron, magnesium, zinc, chromium and others, we need them in strictly regulated amounts. The recommended daily allowance (RDA) for iron is 8mg, for magnesium 400mg, for zinc 11mg and for copper 0.9mg.

• In a normal healthy human-being, the liver stores what we need and converts the toxic excess into neutral urea for removal from the body through the kidneys, but for us with defective livers, we have to rely on penicillamine or trientine to remove excess copper. Some of us do, that is — in others the damage before diagnosis is already so great that nothing short of a liver transplant will do.

• And did you know that 'living donors' are occasionally used in liver transplantation? This is possible because the liver of a healthy person is much larger than needed and, if part of it is removed, it will regrow within weeks to almost its normal size. What an extraordinary organ our liver is! An old play upon words runs as follows. Question: "Is life worth living?" Answer: "That depends on the liver!" What I find interesting about that is, if you translate the question into French, you get an equally valid pun. "C'est une question de foie" (that depends on the liver) which sounds exactly the same as "C'est une question de foi" (That's a matter of faith).

Aubrey Morris, 2011

* *Facts no longer correct*
** *This record still stands*.

Aubrey Morris 12.05.28—19.12.13

This article was written in 2011 when Aubrey was 82 years old. He was a journalist by profession and didn't show symptoms of Wilson's disease until he was 40. Earlier in his life, after University, he embarked on his 2 years' National Service during which time he was selected for officer cadet training in the Royal Air Force. This necessitated a rigorous medical which he passed with flying colours!

In 2008, he submitted his patient story for the newsletter. It was so long it took up six A4 sides and had to be published in two successive issues (2008 & 09). But it was a fascinating read. It begins by telling us that in the space of a year he had a WD diagnosis, kidney infection, foreign object in his bladder and 2 episodes of mania. Fortunately for him, Dr Walshe came to the rescue...

James' Story

Whilst my younger sister, Katie, wants to believe my story starts with her liver transplant on her 18th birthday in December 2014, my story starts fourteen years prior with a misdiagnosis. After my mum noticed something wrong with the way I was using my cutlery, she took me to the GP who subsequently sent me to a rheumatologist where I was diagnosed with distal arthrogryposis. I was eight years old.

At that stage, the only symptom of Wilson's I was displaying was muscular dystonia, something common in arthrogryposis patients. Over the next ten years I would go through two or three weekly physiotherapy appointments, wear wrist splints designed to keep my wrists in a straight position, see various doctors from a variety of specialities and spend endless hours crying to my mother in pain and frustration. Despite the work I put in with my physios and occupational therapists, I showed no improvements, any new symptoms being attributed to the arthrogryposis. My clumsiness, bradykinesia (impaired and slow movement of limbs) and difficulty with repetitive movements were all dismissed by my rheumatologist, despite them not always fitting with arthrogryposis. I was repeatedly told, "When you stop growing, your symptoms will improve, you won't be in pain all the time and the cramping will gradually end." It was only my paediatric physiotherapist who disagreed with my diagnosis and encouraged me and my mum to continue to explore an alternative diagnosis. I would later end up working with her and she was chuffed when I told her I finally had an answer.

Mum and I on crutches at the same time after I'd had yet another fall.

Entering my late teens, I started displaying psychiatric symptoms of Wilson's. My grades at school deteriorated, I started to become withdrawn and anxious, I developed speech problems and I was less able to focus in class and do homework. My physical symptoms were also escalating with my muscle cramping becoming more intense and regular and I started falling and tripping consistently. I was beginning to dribble regularly, have difficulties swallowing and I had a noticeable and constant tremor in my legs and fingers. Unfortunately for me, all my symptoms were not unique to Wilson's: the anxiety was put down to teenage angst, my drop in school performance blamed on new friendships taking me away from my studies, choking whilst eating was thought to be because I wasn't chewing my food properly and the shaking in my legs was passed off because I was simply cold and shivering. Because of these alternative explanations, my Wilson's evaded diagnosis.

Despite my grades slipping, I got a place at university via clearing and in September 2011 I started at Hertfordshire University with optimism and gusto studying Sports & Exercise Science. My optimism was short lived however and I decided to take leave for the rest of the academic year to focus on my mental health. I returned to university in September 2012 and developed a hugely supportive friendship group. I also had more support from my tutors and the on-site therapy team. Despite my mental health never truly recovering and not being able to focus for longer than thirty minutes at a time, my tutors gave me every tool they could to succeed, and boy did I. Over the next two years I achieved what I wanted to both in and out of the classroom, achieving first class grades and coaching rugby in my spare time. However, in summer 2014 my mental health took a nosedive.

I started to experience auditory and visual hallucinations and became convinced I was receiving messages through the radio and TV. I was referred to the local mental health team where I was diagnosed with psychosis. My symptoms worsened throughout that summer break and by the time I was preparing to return to university, my GP advised me against it. I had a summer job filing medical records at my local hospital and I have continued working in the NHS in various roles since.

December 2014 was the beginning of my getting a Wilson's diagnosis. In the late summer of 2014 my sister began experiencing sickness and fatigue along with rapid weight gain. She was 17 years old. Whilst I was convinced she was pregnant, my parents took a more pragmatic approach and sought medical advice. Katie was admitted into our local hospital and subsequently transferred to King's College Hospital, London, in early December; she would leave just over a month later with a new liver.

It was during the days prior to Katie's transplant Wilson's was first mentioned as a possible reason for her liver failure, although they would not confirm this until the histopathology came back post-transplant. Once my sister had her diagnosis, *google* was consulted to find out everything about a condition I had previously only heard of on Hugh Laurie's TV medical drama *House*. As I started researching Wilson's and looked through the neurological symptoms, it was like looking at a *This is Your Life* script. I had every single symptom on the list. From the more obvious symptoms, tremors, dribbling, speech issues to symptoms I did not realise I even had; from having small scruffy handwriting, decreased school performance, forgetfulness to being very easily irritated. After going through genetic testing, urinary analysis, and a head MRI scan I was finally diagnosed in September 2015. At last I had my answers for the constant pain, for why I had not been able to finish my university degree and why despite all the work I did with physios, speech therapists and occupational therapists as a child, I had made no improvements.

I was prescribed penicillamine and pyridoxine and remained under the care of King's College Hospital for the next three years, under both the hepatology and neurology depart-

ments. At first, I was extremely optimistic about starting treatment and about the care I was receiving. I was looking forward to getting better. Unfortunately, my optimism was short lived. Whilst King's is a wonderful hospital and I will forever be grateful to them for saving Katie's life, I feel I personally did not receive a good standard of care from them, often leaving appointments with questions unanswered and feeling incredibly frustrated. For my hepatic appointments, I would see a registrar on Professor O'Grady's team every 6 months with the appointments going the same way every visit. With Wilson's being a rare condition and the registrars unlikely to have seen a patient before they would, understandably, not want to change any part of my care, despite me clearly deteriorating. I would return six months later after the registrars had rotated and have an incredible sense of *deja vu*. I was getting nowhere. My neurology appointments fared no better after being lost to follow up for 18 months. I did not feel cared for and as such, did not care for myself with my tablet adherence being less than rigorous.

By this point I was unable to live independently. I was living with my dad and sister at the time and required my dad's help with all aspects of my life. I needed help getting in and out of the shower, cutting my food up and getting dressed. I would regularly end up with black eyes from holding the phone against my face while working as a medical secretary. I needed my mum or dad to take and pick me up from work daily. My tremors were relentless and out of control. I was unable to be mobile without a walking aid and I could not climb stairs. Work became increasingly difficult as my colleagues and patients that I interacted with could not understand me as my speech was so mumbled and stuttered. I would also frequently shake myself off my chair on to the floor due to my tremors; requiring three or four nurses to assist me back up. It was after one of these falls my life would take a turn for the better.

After being helped back into my chair for the fourth time the same day, my manager and senior nurses insisted I go down to A&E to get help. I was admitted on to the neuro ward where I stayed for two nights. On my third day on the ward, I was visited by a neurologist, Dr Jan Coebergh, who had a special interest in Wilson's disease and he told me about a specialist multi-disciplinary clinic he was helping to set up at the Royal Surrey Hospital in Guildford. He wanted me to be one of the first patients to attend and just like that, I had a glimmer of hope back. I had my optimism back.

In September 2018 I attended the clinic and was seen by a gastroenterologist, pharmacist, dietician, ophthalmologist, neuropsychiatrist and the research team. I would be seen by the same doctors, be monitored with regular blood tests, regular 24-hour urinary collections, regular scanning and most importantly, my medication was adjusted to what was needed dependent on my results. I felt cared for and made remarkable improvements under Prof Ala, Dr Coebergh and the rest of the team. I felt extremely lucky I met Dr Coebergh when I did and was able to attend this clinic. I was not going to waste an opportunity to improve and followed their advice to the letter.

By summer 2019, I was living more independently. I still required some help but I had made incredible improvements. So dramatic were they, I was asked to share my story at various hospital events. The first was at the Royal Surrey Hospital's AGM, the next was to neurologists at St George's Hospital, Tooting, and the third slightly *awkward* talk was to consultants at King's College Hospital with both my former liver and neuro consultants present in the audience.

I have since been part of medical trials for potential new treatments, including an 8 week inpatient trial for a new drug therapy and a 48 hour sleep study. I have also been involved in focus groups discussing proposed new research into Wilson's disease and improving diagnosis rates. All the while I have continued to make improvements though admittedly smaller ones now. I still have my issues, my muscles still cramp, I still fall over a lot and I still forget what I had for breakfast by lunchtime, but I feel like a new person!

Christmas shopping with my biggest champion, Georgina.

The support I have received from my family, partner and work colleagues has kept me going and kept me positive throughout my entire journey. Now, age 31, I have managed departments in the NHS and am looking at transferring my university credits into an Open University course to get my BSc.

I can now drive safely too and in summer 2023, made the 200 mile move north to Liverpool where I now live with my amazingly supportive partner, Georgina. I have continued advocating for people with chronic health problems and hope to do so within WDSG-UK. Lying in St Peter's Hospital Stroke Ward in Chertsey, Surrey, five years ago unable to stop my tremors or string a cohesive sentence together, I would never have thought these things possible.

James (then 22) and Katie (18) on Christmas Day 2014 at the Ruskin Park Bandstand round the corner from King's College Hospital, London. Katie had received her liver transplant 8 days earlier and James hadn't yet been diagnosed.

After James told his little sister, Katie, that he was writing an article for the WDSG-UK newsletter based on his Wilson's disease journey, Katie who has had a liver transplant due to Wilson's disease wanted us to know that it was entirely due to her suffering that his life had been saved!

She says that she had come within 36 hours of death due to Wilson's disease. Her subsequent transplant and all that it entailed including her selfless surrender of alcohol, sushi and visits to Seville to enjoy the local oranges, was the sole reason why James had survived to tell the tale!

The BASL Wilson's Disease Special Interest Group (WD SIG)

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 WD SIG 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. For enquiries to Prof. Aftab Ala, new BASL WD SIG Lead, please contact him at <aftab.ala1@nhs.net>

8th Meeting—London—24.11.23 1400—1700 hrs

At the end of November 2023, the BASL WD SIG Group held its first face-to-face meeting since the Covid pandemic. The event took place at St Ethelburga's Centre for Reconciliation and Peace Venue – an historic church in the City of London -- – and was organised by **Prof. Aftab Ala** of King's and Royal Surrey, BASL's new Chair for Rare Diseases and the Wilson's disease sub-SIG lead. It was kindly supported by the Institute of Liver Studies at King's College Hospital, London.

There were 29 attendees including representatives from neurology, psychiatry, adult and paediatric hepatology, genetics, pathology, metabolic disease, nuclear medicine, the National Institute for Health Research (NIHR) and WDSG-UK. Prof. Ala set the scene and discussed the unmet needs in Wilson's disease. He outlined priorities in diagnosis and management and emphasised the importance of collaboration between the specialist centres in the UK. A summary of the talks is as follows:

Cuproptosis: Dr Rupert Purchase outlined the mechanism of cuproptosis – a regulated cell death induced by copper(I), and the implications of this recently formulated (2022) biochemical sequence for the pathogenesis of Wilson's disease.

BASL Wilson's Disease Guidelines and National Audit: Prof Oliver Bandmann (Sheffield) and Dr James Liu Yin (King's College, London), spoke about the recently published BASL Guidelines in the Management of Wilson's disease and the need to find out how widely it is being referenced by clinicians in the UK. James Liu Yin, Bill Griffiths, Aftab Ala and Oliver Bandmann will develop an electronic questionnaire on the subject to be circulated initially among WD SIG members. Thereafter, the aim is to disseminate it more widely among centres not affiliated to the BASL WD SIG.

Genetics and Wilson's Disease: Dr Bill Griffiths described a sibling trio with positive genetics for Wilson's disease where it was difficult to ascertain whether the trio actually had the condition and whether to observe, perform liver biopsy or treat, and if treat whether zinc would be appropriate. There was no consensus from the room which shows how tricky this situation can be. One can of course revert to the Leipzig scoring system but it is not perfect. The important message is to involve Wilson's disease expertise which the SIG is able to provide.

Skin Associations in Wilson's disease: Dr James Liu Yin explained that there is increasing recognition of skin related side effects due to long term chelator treatment. The possible implications of overtreatment and approach to management were also discussed. The potential importance of monitoring was emphasised and a paper on the subject has since been published—Yin JL, Salisbury J, Ala A. Skin changes in long-term Wilson's disease—*Lancet Gastroenterology and Hepatology* 2024 Jan;9(1):92.

Psychiatric Presentation in WD: Dr David Okai (Maudsley, SLaM) explained that psychiatric presentation in WD is poorly understood and yet remains a common association. He described the key links within neuronal pathways. As a WDSG-UK initiative, James Liu Yin is planning to do a retrospective and prospective study of patients presenting with associated psychiatric symptoms at King's/Maudsley to see how many have been/are being routinely investigated for WD.

Trientine Formulations: Dr Rupert Purchase explained the trientine free base equivalents of the four trientine formulations currently authorised by the UK's Medicines and Healthcare products Regulatory Agency (MHRA) and discussed some current issues associated with the pharmacology, pharmacy and cost of this established treatment for WD.

Setting Up and Developing Patient and Public Involvement and Engagement (PPIE) in Wilson's Disease and guiding clinical trials: James Ratcliff described the importance of listening to peoples' lived experiences in rare diseases such as Wilson's disease when making decisions on the direction of clinical research. He also explained how patients can help set the priorities for physicians and researchers.

Wilson's Disease and PET: Prof. Phil Blower (King's College, London) described a technique he has developed for visualising copper *in vivo*. [Positron emission tomography (PET) and electronic autoradiography using the positron (β^+) emitter copper-64 in the form of a copper-64(II) bis (thiosemicarbazonato) complex]. In Denmark, copper-64/PET has been used in human volunteers to better understand the mechanism of drugs used to treat WD.

WD Highlights from the American Association for the Study of Liver Disease (AASLD) Annual Conference 2023: Dr James Liu Yin summarised the various video presentations given including updates on dried blood spot testing of newborn babies in Washington state, clinical data on the use of Methanobactin for WD, comparison of non-caeruloplasmin copper by protein speciation and 24hr urinary copper excretion results from the CHELATE trial and work investigating other biochemical pathways in the development of liver disease in ATP7B deficient animal models.

Clinical Trials/Updates

Clinical Trials—Current:

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.

GATEWAY is enrolling patients for the trial, which **is** 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 in a hospital setting. No patients receive a placebo treatment. Patients will then return to The Royal Surrey at planned times over the following years so that the study team can assess how they are doing. These visits occur more often in the early part of the trial (first year) and the number of visits decreases thereafter. The study continues for 4 years after the initial 1-year trial period, which is 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

ULTRAGENYX - The Cyprus2+ Gene Therapy Study

Similarly, the American biopharmaceutical company, Ultragenyx, has developed a gene therapy treatment for Wilson's disease patients known as UX701. This is being trialled on patients in a 3 part study called Cyprus2+. Prof Aftab Ala is recruiting patients for the Cyprus2+ Study at KCH, London. For more details please visit https://ir.ultragenyx.com/news-releases/news-release-details/ultragenyx-announces-completion-dosing-across-stage-1-cohorts or contact Aftab at aftab.ala1@nhs.net

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

The United Study is a Post Authorization Efficacy Study (PAES) which aimed to gain a better understanding about the effects that Cufence® (trientine dihydrochloride) has on the body in adults and children with Wilson's disease. This trial had 11 study sites across Europe including 2 at King's College Hospital, London (Adult & Paediatric). Enrolment has now closed and we are waiting for Univar to share its results.

Future:

1. **Arbor Therapeutics.** Phase 1 – In set up Efficacy and Safety of *Methanobactin* in *normal* human and Wilson's disease patients. 2024-25 Set up. Afta Ala

2. **Orphalan.** A Phase I, Single Centre, Randomised, Interventional, Open-Label, Cross-Over Study to Evaluate the Pharmacokinetics (PK) and the Safety and Tolerability of a Total Daily Dose of 900mg of TETA 4HCL 2024-2025.

3. Developing the use of optical coherence tomography to assess Kayser Fleischer rings in Wilson's disease. Proof of concept work – single centre. James Liu Yin, Samira Anwar, Simon Taylor, Aftab Ala, NIHR CRN portfolio adopted 2024.

Previous: ALXN1840

1. Copper Concentration & Histopathologic Changes in Liver Biopsy in Participants With Wilson Disease Treated With ALXN1840. Phase 2 - results have been submitted to the Journal of Hepatology—2024.

2. Copper and Molybdenum Balance in Participants with Wilson Disease Treated with ALXN1840—Phases 1 & 2. Also submitted to the Journal of Hepatology—2024

Updates Mar 24

Supply of Penicillamine in the UK—Mar 24

There have been continual problems since December 2023 with the supply of 250mg strength penicillamine tablets from both UK manufacturers— Viatris and Kent Pharma. 125 mg tablets seem to be available, but your prescription will need to be altered accordingly to request them. The advice is to let your WD specialist know if you are experiencing problems as hospital pharmacies are better placed to source supplies than community pharmacies. In response to emails, Kent Pharma claims that "the limiting factor is demand" and Viatris says that their "normal supply will resume in mid-May."

Do contact **Valerie** by email or WDSG-UK Fb, if all else fails.

WD Clinical Trials, Research and The Wilson's Disease Patient Register—UK

WDSG-UK supports all aspects of research into Wilson's disease, but research and clinical trials cannot go ahead in the UK without pharmaceutical companies first engaging with patients. They may do this by asking the patient to complete a questionnaire anonymously or more often by a medical research company speaking to a patient directly. In the case of the latter, patients are normally offered a fee for their participation. If you would like to take part in future interviews, please make sure you have completed a WD Patient Register form and/or registered your interest with Valerie.

A pamphlet and information sheet about the Register are available to download off our website www.wilsonsdisease.org.uk, together with the corresponding registration form.

Members' News 2023-24

Our past chairman and former consultant hepatologist at Addenbrooke's Hospital, Graeme Alexander, retired from medicine in 2018 and together with his wife, Sandra, swapped their busy and demanding lives in Cambridge for the tranquility of life in north-western Scotland —home of his ancestors. Having built their own house on the shores of Loch Sunart, they are now able to indulge their passion for sailing—in their very own yacht!

Breakfast on 'Culla' at Tobermory (Mull)

Graeme made his mark on the Wilson's community while President of the British Association for the Study of the Liver (BASL) by setting up the Wilson's Disease Special Interest Group (WDSIG). He very kindly took over the chairmanship of WDSG-UK in 2019 and steered us through the muddied waters of the Pandemic which followed him into office. Many of you will have known him as patients or *met* him virtually through the Zoom meetings we held. We thank him for all his support and wish him and his wife well in the next phase of their lives.

May the Force be with you Graeme and all who sail with you!

* * * * *

Graeme isn't the only one who has been on the move. Vice-Chairman and Zoom host, Liz Wood, has also relocated recently. She writes:

Liz with Snape Maltings behind

My husband John and I first started visiting Woodbridge in Suffolk in 2002 and instantly fell in love with the area. We started off by going just once a year but this gradually increased to several times each year, so that by 2014 we had decided that we would like to retire to Suffolk when the time came. We expected that time to be sometime in the mid-2020s but when COVID hit, we rethought our priorities and decided that we would make the move as soon as we could. We put our house on the market in July 2021 and moved to Rendlesham in December of that year. Rendlesham is a village about halfway between Woodbridge and Snape Maltings, not far from the east coast of Suffolk.

We are surrounded by wonderful countryside, forests, heathland, rivers and some beautiful villages and towns. We really are very lucky. The biggest draw for me, as a classical musician and singing teacher, is being on the doorstep of the world-famous Snape Maltings Concert Hall. I spend a great deal of my time there either working with a children's opera group, volunteering as an usher at concerts, turning pages for pianists in concerts, singing in the resident chamber choir, listening to fabulous concerts, eating in the cafes (particularly cake!!) or walking along the various beautiful footpaths on the banks of the River Alde (as depicted in the photos).

Bridge over the River Alde

* * * * *

And after 15 years of living with and caring for Dr Walshe, daughter Susan and her partner Phil have sold their own family home in Cambridgeshire and moved to a village outside Bath, where their son Simon and his wife Natalie live with their young family. They have immersed themselves in community life and Phil continues his lifelong hobby of cycling—albeit a far more hilly and taxing pastime nowadays than he's used to!

In their new home with the family

Much as they miss their father's company, they are thoroughly enjoying their new life in the south-west, looking after the next generation— grandchildren Annabelle and Rose—as well as familiarising themselves with a part of the country previously unknown to them.

Ben was working as a project engineer at Heysham Nuclear Power Station until his diagnosis of Wilson's disease on his 28th birthday in 2016. He loved his job, but he hasn't been able to work since. Here is a resumé of what has been going on in his life since.

After his diagnosis and treatment began, Ben's condition deteriorated rapidly. He had pronounced vigorous tremors across the whole of his body, muscle contraction in his arms and legs (dystonia) and difficulty speaking (dysarthria). He was admitted to hospital a short while later. After four months as an inpatient, he was discharged to a care home where, with other care homes in between, he remained living until moving in with his mum in September 2020 along with 24/7 carers. The following year he took tenure of his own house where he is living today. He is supported by friends and family and a super 24 hour, dedicated care team.

The team all have different skill sets including case management, physiotherapy and speech therapy, and all play an important role in helping him with his rehabilitation. Since moving into his own home Ben feels that his mental and physical state have improved. Ben continues to engage with his psychologist.

When Ben was first diagnosed, his son Jacob, was only twelve months old. Jacob is now 9 and recently his school invited Ben, along with his two carers, to give a slide presentation to the rest of Jacob's class explaining all about Wilson's disease and the effect it has had on him. In amongst the facts about his personal journey living with the disease, Ben slipped in fun facts for the children and posed questions to keep them on their toes! It culminated in a quiz with a box of *Celebrations* for the children to share!

At the beginning of the year, Ben made several resolutions and set out to achieve various goals. Jacob is the centre of his life and Ben has resolved to help him more with his schoolwork and personal development and to set aside more time to have days out and holidays with him. A trip was planned to the Science and Technology Museum in Manchester recently and a short holiday to Turkey is on the cards in the Easter holidays. To enable Ben to achieve these goals the whole care team needs to be onboard.

On a personal level, Ben is striving to improve his fitness. With guidance from his Wilson's specialist and dietician he is following a better diet, losing weight and having earlier nights. To improve his language skills he is wanting to spend less time communicating through his I-pad and more time practising words — using exercises given to him by his SALT (Speech and Language therapist).

As a keen sportsman and self confessed adrenalin junkie before he was diagnosed, Ben hopes that his dreams can one day become a reality again. Nearer to home, he is looking forward to *hanging out* at the Lytham Festival in July (a live music event held on the seafront in Lytham—at which Ben will have a dedicated area) and watching his beloved Liverpool playing at home.

Finally, at the end of 2024 Ben is hoping to have a home gym and relaxation/therapy room, another positive step working on his rehabilitation with his Physio and Occupational Therapy team.

He might not be able to join his two friends Phil Angus and Kurt McGuiness as they set themselves challenges to scale mountains across the world (see last year's newsletter), but everything he does achieve is like climbing a mountain to him.

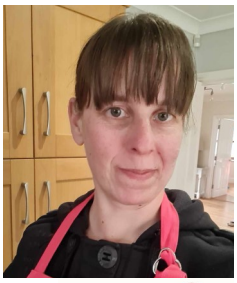
We wish him and all his family well for the future.

Ben with sister Sam and son Jacob just a few months before his diagnosis.

Doing his presentation at Jacob's school

At Anfield Mar '24

The New Therapy Room under construction



Katie Hibbard is a wife, and mother of two young children and lives in a village in Lincolnshire. She first introduced herself to us through her patient story in the 2016 edition of our newsletter. At the age of 25 she had started to experience neurological symptoms of Wilson's disease and was lucky enough to be diagnosed four months later after a brain scan and subsequent visit to an ophthalmologist. She only ever missed two months of work because of her illness and was recently presented with a 20 year Long Services Award from her employers, Lincolnshire County Council.

One such pebble commission

During lockdown, she discovered a talent she didn't know she had! With people being encouraged to take daily exercise and younger members of the community needing entertaining, an idea was put forward that the children could paint large pebbles and hide them around the village for others to find. Once found, they could then be rehidden. Katie thought this a great idea and set about buying herself some pebbles and paints immediately. So popular were her designs, that people were finding them, pocketing them and taking them home—never to be seen again! Word spread and she started getting commissions to paint personalised ones. Seizing an opportunity to raise some funds for WDSG-UK, Katie went into production and we came out of it £225 better off!

Tru-ly Scrumptious!

Fast forward three years. In Summer 2023, Katie decided to turn her hand to something else. She is passionate about baking and, instead of adding to her waistline, thought it would be nice to do it on a small commercial basis. She completed a Food Hygiene and Food Safety course and was awarded a certificate with a 5* rating. Trading under the name of *Katie's Cakes & Bakes,* she set to work in her kitchen and now has a steady stream of customers ordering cakes and cookies. In the near future she is planning to start a mail order service. This is an exciting prospect for us as we hope that some will find their way to our AGM in the Summer as a raffle prize! As soon as she launches, we'll let you know.

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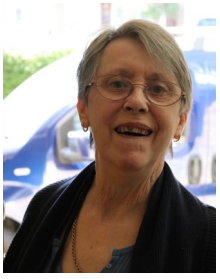


Joan Smith, who was diagnosed with Wilson's disease in 1963, has been a member of the Group since it was founded in 2000. Her patient story was included in the 2004 newsletter (*vol 5 issue 1*). Last year she wrote an article which, among other things, warned Wilson's patients of the perils of Vitamin D deficiency, a subject which Rupert Purchase has tried to address this year on *p11*. She now writes:

A Liverversary Card

I have a bit of news for the newsletter. It was the 20 year anniversary of my liver transplant on 10th January this year. We did mark the event. My son, an accomplished cook, came over and cooked us a fabulous meal. My liver is still going strong. We drank a toast to my donor and remembered their family.

My liver may be fine but I have now been diagnosed with Lymphoedema. Nothing to do with the transplant, but caused by the removal of Lymph Nodes during Breast cancer treatment 8 years ago. This is a very unpleasant illness with no cure and the treatment is time consuming.

* * * * *

And Caroline Simms, the co-founder of WDSG-UK, also celebrates her 20 year *Liverversary* this year. In the 2017 newsletter she gave us an update on her Wilson's journey which began in 1987 when she was 14 years old. Now aged 50, she is embarking on a new venture setting up a Liver Transplant Support Group for patients from Nottinghamshire & Lincolnshire (LTNL), an area not previously covered by existing region specific groups. She writes:

Although Queen's Medical Centre in Nottingham is not a liver transplant centre itself, it runs a liver transplant clinic for the many pre- and post-op liver transplant patients living in its catchment. I am one such patient that attends the clinic. My new Group (LTNL) is just beginning. We have a Facebook page and are currently building a website www:ltnl.org.uk, which will provide information relevant to liver transplantation, current research and more. It will be run in a similar format to WDSG-UK producing an annual newsletter with articles on liver related issues, up-to-date research, patient experiences etc and host meetings and events to support patients, their families, friends and carers.

Please contact me if you know anybody in the area who might benefit from the Group.

In 2023 Anne-Marie turned 70, so she and her husband, Steve, decided to plan an extra special journey to somewhere new for them both. They opted for an expedition cruise to Greenland with Hurtigruten. As the ship departed from Reykjavik, capital of Iceland, they decided to hire a car and spend a week in the small coastal village of Grenivik in the north of the country before embarking on the cruise. **Anne-Marie** writes:

Adventures in Iceland and Greenland: two contrasting islands of the North Atlantic

We arrived in time for the summer solstice in June and were able to experience the midnight sun in northern Iceland: continual daylight and at midnight the sky glowed a wonderful red. We also loved the profusion of colourful wild flowers, an unexpected delight.

The sky glows in the midnight sun

We enjoyed the most amazing view across the Eyjafjardará fjord from our house and used to spend time watching the sea birds and looking out for signs of whales and dolphins.

Iceland is famous for its waterfalls and one morning we beat the crowds to walk up to the mighty Godafoss: "waterfall of the gods".

We spotted herds of the small, sturdy breed of Icelandic horse roaming free in the countryside. They can be trained for riding and we often came across holidaymakers enjoying a horse trek.

Horse trekking through the summer flowers

The distances are deceptive. It took us six hours to drive from the airport to Grenivik along the ring road which encircles the island but through wonderful dramatic scenery and some dramatic weather!

As for volcanoes, while we were away there was an eruption on the peninsular where the airport is located and we could see the smoke rising in the distance on our taxi ride to catch our plane home. Fortunately, there was no ash cloud to disturb air traffic.

Embarking on our cruise, we discovered that our beautiful brand new ship MS Fridtjof Nansen was aptly named after the Norwegian explorer who was the first man to lead a team across the interior of Greenland, the world's largest island, traversing it on cross-country skis in 1888.

Our home in Greenland—MS Fridtjof Nansen

Magical moments include our first sighting of an iceberg and subsequently learning about their lifecycle from an enthusiastic young lecturer. Different sizes have different names: small icebergs are called growlers and bergy bits! We soon got accustomed to sailing through the many shapes and sizes of icebergs that had calved off the Greenland glaciers and were making their long journey south to eventually melt away in the Atlantic ocean.

We embarked on several trips from the ship in zodiacs (rubber dinghies) to get up close to a glacier or to land on shore in uninhabited spots. These visits were carefully controlled and we had to wear special boots to avoid contaminating the pristine Arctic environment. The worst thing we had to contend with on shore were the midges, bigger than the Scottish variety, and a constant nuisance. Mosquito nets were essential headgear!

Up close to icebergs

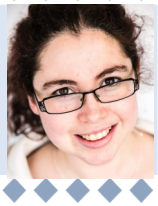
Warding off the midges

We also visited some of the settlements along the west coast including the capital Nuuk and were able to learn something of the Inuit culture and traditions. Greenland now belongs to Denmark but enjoys a certain amount of political autonomy.

The weather was typically very grey and rainy for much of the time but we had two spectacular sunny days. Sailing through the Nunaap Isua area at the tip of South Greenland through spectacular fjords with their stunning backdrops, was a truly emotional experience.

We returned from our wonderful Arctic adventure with certificates awarded for crossing the Arctic Circle to remind us of our very special holiday.

Abby's Puzzle Page



PARIS OLYMPICS 2024

Welcome to Abby's 3rd puzzle page which should have something to appeal to puzzlers of all abilities. It is themed on the Summer Olympic/Paralympic Games which will be taking place in France later this year.

At the Games 32 sports will be contested with hundreds of medals awarded. Here there are 7 categories with no prizes at all! Remember, it's the taking part that counts! Answers can be found at the bottom of *p 23* — opposite.

1. Spot the Difference—Easy

There are 10 differences in the pictures below—can you find them?

2. Sudoku - Easy

4	8			7				1
5				3			2	
		9	8					5
3				4		9	8	
		8		1		2		
	9	4		5				3
6					5	3		
	5			9				6
9				6			4	8

Sudoku puzzle provided by www.sudokuof theday.com

3. Emojis: Medium

Work out the Names of the countries from the following Emojis:

1. 2️⃣ + Ⓡ + 🔑
2. 🎛️ + 🅰️ =
3. 🧊 + 🏜️ =
4. ⭕ + 👦 =
5. 🐆 + Ⓡ =
6. 🎷 + 🍳 =

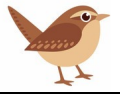
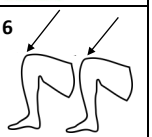
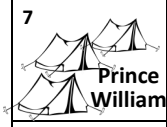
4. Wordsearch - Hard

Find the following Olympic events in the puzzle above and what sport are you left with?

ARCHERY CYCLING DECATHLON DIVING EQUESTRIAN FENCING FOOTBALL GOLF GYMNASTICS JAVELIN RUGBY SWIMMING TRIATHLON VOLLEYBALL WEIGHTLIFTING

5. Valerie's Dingbats -

Très difficile. Below are the names of 8 French cities & 2 historic French towns.

1 	2 
3 	4 
5 	6 
7  Prince William	8  O
9 O C	10 Planet A

6. Anagrams - Hard. Countries of the World (flags might help!)

1. Innate rag
2. Buxom gruel
3. Iodise nan
4. Fuchsia Rota (2 words)
5. Tetanus Diets (2 words)
6. Sooty Vicar (2 words)

7 Flags of the World:

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 the clinic manager Richard Bayley Richard.Bayley@uhb.nhs.uk or to Dr Holt's PA, Ms. Maria Del Vecchio Maria.DelVecchio@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 at Catherine.Stewart@uhb.nhs.uk and will help you make an appointment in clinic.

The Cambridge WD Clinic

Dr Bill Griffiths (Consultant Hepatologist) and **Dr Paul Worth** (Consultant Neurologist) run a joint clinic at Addenbrooke's Hospital every 6 months on a Wednesday morning for patients with neurological manifestations of Wilson's disease. Dr Griffiths sees a number of patients with hepatic-only disease separately on Thursday mornings in his adult genetic liver clinic. 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 35 years. It was set up in 1987 by Dr John Walshe after his retirement from Addenbrooke's Hospital, Cambridge. **Dr Godfrey Gillett** attended from the mid-1990s and ran the clinic after Dr Walshe stepped down in 2000 at the age of 80. Later the clinic was moved to the National Hospital for Neurology and Neurosurgery, Queen Square, continuing the long tradition of care for patients with Wilson's disease which began in 1913 with Dr Samuel Kinnier Wilson. Today the clinic has ~70 patients and is held on the third Friday of the month. **Dr Sam Shribman** joined the team in 2017 and now runs the clinic with the support of Dr Godfrey Gillett and **Miss Maggie Burrows**, Specialist Nurse. The clinic also benefits from access to the Neuropsychiatry, Neuro-Ophthalmology, Speech and Language Therapy, Physiotherapy and Vocational Rehabilitation services at NHNN. Referrals from across the UK are welcome, addressed to Dr Sam Shribman, Charles Dent Metabolic Unit, Internal Mailbox 92, NHNN, Queen Square, London WC1N 3BG, uclh.metabolic.enquiries@nhs.net.

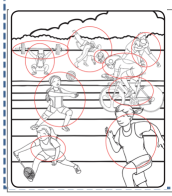
The Sheffield WD Clinic

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 or to **Dr Tammy Hedderly** Tammy.Hedderly@gstt.nhs.uk.



ANSWERS: 3.Emojis—Countries: 1 Turkey, 2 China, 3 Iceland, 4 Oman, 5 Qatar, 6 Japan; **4.Wordsearch** You're left with Rugby; **5. Dingbats—French Cities/Towns:** 1 Rennes (a wren), 2 Lyon ("**I**" sound swapping to "E" sound), 3 Toulouse (2 loos), 4 Versailles (firs +"I"), 5 Cannes (can), 6 Nice (knees), 7 Quimper (camp + heir), 8 Bordeaux ((black)Board + O), 9 Nancy (0 = None + C), 10 Marseille (Red Planet = Mars + A); **6 Anagrams** 1 Argentina, 2 Luxembourg, 3 Indonesia, 4 South Africa, 5 United States, 6 Ivory Coast; **7. Flags of the World** a) Ivory Coast, b) Australia, c) Luxembourg, d) Germany e) South Africa f) Indonesia, g) Jamaica h) Argentina i) United States j) Iceland.

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5	7	6	4	3	1	8	2	9
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3	1	5	6	4	2	9	8	7
7	6	8	9	1	3	2	5	4
2	9	4	7	5	8	1	6	3
6	4	7	1	8	5	3	9	2
8	5	3	2	9	4	7	1	6
9	2	1	3	6	7	5	4	8

A Date for your Diary 2024-25

Date	Time	Event
Sunday, 7 July 2024	**1130—1530**	**WDSG-UK 14th 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 Disease Support Group – UK
www.wilsonsdisease.org.uk

CONTACTS:

Dr Bill Griffiths:	Chairman email: **billgriffiths@wilsonsdisease.org.uk**
Elizabeth Wood:	Vice Chairman email: **lizwood@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**
Debbie Buckles:	Committee Member Email: **debbie@wilsonsdisease.org.uk**
Claire Stapleton:	Committee Member **Email: clairestapleton@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.