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

NEWSLETTER

#wdsguk APRIL 2021

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

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

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

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

AFFILIATED TO :

BRITISH LIVER TRUST

The Liver Patients' Transplant Consortium

GENETIC ALLIANCE UK

RARE DISEASE UK

Children's Liver Disease Foundation fighting childhood liver disease

METABOLIC SUPPORT UK Your rare condition. Our common fight.

Who would have thought this time last year that we would still be facing restrictions one year later! It's been a long old haul! Hopefully, with non-essential shops opening today and outside venues now able to serve food, things will gradually start to return to what we have been told will be our *new normal.* Actually, the situation has had its benefits—certainly for me—like foregoing the housework in the absence of visitors and hiding my wrinkles behind the mandatory face masks!

On the down side, I haven't seen **Dr Walshe** since his 100th birthday nearly a year ago and then it was from the other side of the street! However, I am assured that he is fit and well and will be welcoming visitors as soon as his second vaccine has taken effect. It is his 101st birthday shortly, for which we wish him a very Happy Birthday.

Thinking of new material for the newsletter every year is quite a challenge and I thank everybody who has contributed to it this year. In particular, my thanks go to **Debbie Buckles** who has shared her patient story with us and also **Eddie Watt** who has shared his. In the case of the latter I shall look forward to joining up all the dots.

Graeme Alexander has thrown himself into the role of chair with great aplomb and I thank him and **Mary Fortune** for producing an article about Gene Therapy which, if proved successful, may be the treatment of the future for WD patients. I should also like to thank **Dr Godfrey Gillett** who works tirelessly for the Group behind the scenes and who is ever willing to offer advice and give practical help to new patients when they come to our attention.

As far as fundraising is concerned, we are grateful to **Katie Hibbard, Chris Billinghurst** and **Lynn Martin,** who despite the pandemic still managed to raise funds for us. Like all charities, we have faced a significant reduction in our income this year and are always looking for innovative ideas for fundraising in the future. One plus, however, is that we have had more new members join the Group this year than ever before and we offer them a warm welcome and thank them for their support.

Do join us on Facebook. We now have over 1100 members and it is an excellent medium to advertise what the Group is doing and any problems we are facing and also offer new patients the benefit of our own experiences. Our webmaster, **Michael McConnell** has kindly revamped our website <www.wilsonsdisease.org.uk> which is well worth taking a look at. There are medical articles, patients' stories, pamphlets, information about treatment, diet sheets, archived newsletters and much more and if there's anything vital missing that you think should be included, then please let us know.

As a Group, our greatest sadness this year has been the passing of **Emma Collcott** last September at the age of 33. Any of you who have attended WDSG-UK annual meetings in the past will remember Emma for her strong presence at these meetings, never missing an opportunity to ask questions of the doctors and always ready to exchange smiles and encouragement to the rest of us. Her mum and dad, Lesley and Ian, have courageously shared her story with us and the short poem written by Lesley for Emma's woodland burial is included overleaf. We thank the family for nominating us as their chosen charity when collecting donations in Emma's memory and we send them our love.

Your subscriptions for 2021-22 are now due so please renew promptly. For those paying by Bacs, please don't forget to return your form to me—emailing it is fine! Details of this year's AGM via Zoom are also enclosed and we hope to have a good attendance again this year. See you then, and meanwhile I wish you a warm and safe Summer.

Valerie

IN THIS ISSUE ...

Chairman's Report for 2020-21

The past 12 months have been extraordinary, but with careful management the next 12 months offer real optimism. In the UK, Covid has had serious indirect effects on the health of patients with liver disease by curtailing or reducing hospital in-patient services, including liver transplants. Covid has had a more direct effect on patients with liver disease, many of whom acquired the infection whilst in hospital in the early days of the pandemic before adequate tests were available and hospitals were subject to the severe pressure of large numbers needing admission. In addition, we learned that those patients with *very advanced* liver disease that acquired Covid suffered increased complications and an increased risk of death. Fortunately, for the large majority of patients living with liver disease, including those with Wilson's disease, there was no increased risk of illness due to the virus or the combination of virus infection with liver injury.

Covid has caused havoc with every aspect of life including the WDSG-UK. The committee has learned to work effectively on Zoom. The 2020 AGM also took place on Zoom and to the surprise of all, this proved a real success with members joining us from all over the world and we 'met' members who have never been able to make the face-to-face annual meeting. There were many more contributions from the membership than we had anticipated, perhaps because people were more confident asking questions in their own homes. Because the AGM had worked so well, we ran a coffee morning on Zoom on Rare Disease Day in February (open to all comers) and the international contingent was even more noticeable, a lesson for all of us. Our warm thanks to Liz Wood who has been leading the chorus of members on Zoom, a real achievement.

You will read elsewhere in the newsletter *(p17)* that the BASL special interest group on Wilson's disease has continued to be active (on Zoom). The committee and I were delighted that Bill Griffiths was elected unopposed to chair the new BASL SIG on metabolic disease that will include Wilson's disease; his contributions to the BASL SIG have ensured that this is a truly effective group that will guarantee change. The majority of WD patients (adults and children) have or will soon have access to regional centres with expertise across all the relevant specialities. All of us in the group have been learning from experts in the field outside our own speciality. In many cases the regional centres will provide advice in a supervisory role for those patients living at distance from the centres (Covid has also taught us that clinics can run very well on Zoom). Bringing all centres up to the standard of the strongest units is an aim we can achieve soon.

No doubt there are patients with Wilson's disease in the community that are off the hospital radar, perhaps because they are well and do not need expert help. But the fear persists that some patients may be under-treated and the National Register (run by Mary Bythell at PHE) is making real progress in identifying all the patients in England who have had contact with the NHS. When it is fully functional it will help identify areas or patients in the UK with sub-optimal care and in other disorders this approach has educated even experienced physicians or experts.

The various Zoom meetings have allowed members to ask important questions. It appears that for many patients understanding the genetics of Wilson's disease is an area we should address and one aim of the UK guidelines on managing Wilson's disease (which Bill mentions in his report and which should be completed before the 2021 AGM) is to ensure that *all* patients have access to a genetic analysis.

We hope to see you, one way or another, at the AGM in July. Finally, I would like to thank the members of the committee for their contributions to WDSG-UK and in particular Valerie for her total commitment to the charity (and for keeping the rest of us in order).

Graeme Alexander
April 2021

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 last year and our two new members Sarah Morton and Eddie Watt who have set up regular monthly standing orders to us. In addition, we should like to thank **Giuseppe Cardone** who, together with a group of fellow Google employees, raised a further **£620** for us via Google.org support for non-profits. We also very much appreciate the **£70.00 Joan Smith** donated to mark her 70th birthday, the **£165** forwarded by **Dema Metreweli** from research monies she had received, the **£200** given by **Lisa Lawson** in memory of her mum Christine White and the **£600** collected by the **Collcott/Galloway** families in memory of dear Emma.

We thank our fundraisers **Katie Hibbard, Chris Billinghurst** and **Lynn Martin** who despite Covid restrictions have brought in additional combined income of **£1,150.** Their enterprising pursuits are detailed on *pp6-7*. When sponsorship is collected online through ***Virgin Money Giving***, donors can choose to gift aid their monies, which this year have generated an extra **£100.** Any donations to us, of course, can be gift aided. Forms are available from **Valerie** or directly off our website <www.wilsonsdisease.org.uk>

NHSBT Organ Donation & Transplantation Directorate (ODT), the Liver Patient Group (LPG) and the Liver Patients' Transplant Consortium (LPTC)

The 11th meeting of NHSBT and the LPG took place via MS Teams Video Conferencing on 8th July and Valerie attended on our behalf. The impact of Covid-19 on liver patients was discussed and the three main vulnerable patient groups

were said to be those with immune-suppression, those with decompensated cirrhosis and patients actively awaiting a liver transplant. All liver transplant centres remained open during the pandemic, but to begin with transplants on adults were dramatically reduced although all patients on the clinically urgent list received transplants. Liver perfusion before transplantation is still in its infancy, but is now available in 6 out of the 7 transplant centres. Full data can be seen on the NHSBT website https://www.odt.nhs.uk. The new deemed consent/opt-out system for organ donation in England seems to be working well.

The LPTC also held a separate video call on 11 December to discuss the views of Liver Patient Groups on the proposal of creating 2 new transplant centres in the UK at Plymouth and Liverpool to increase transplant capacity. The LPTC chose to back this.

The British Liver Trust

The British Liver Trust is producing a new pamphlet on Wilson's disease which WDSG-UK has had input into and we responded to their appeal for funds during the pandemic by making a £500 donation to them.

The CONCORD Project (Co-ordinated Care of Rare Disease) run by UCL's Dept. of Applied Health Research

Our past chairman, Jerry Tucker, was very much involved with this project and continues to keep us up to date. He attended a webinar on 16 March and reports, "Funded by the NIHR (National Institute for Health Research), the CONCORD Project has just completed its main research phase into co-ordinated care for rare disease patients. WDSG-UK was part of the initial focus group and patients, carers, healthcare professionals and other rare disease groups have since been canvassed. Key components of care were identified as being access to treatment, support for patients and their families, care planning and care co-ordination, much of which is lacking in patients with rare conditions. One of the problems is the difficulty in diagnosing rare conditions due to the small number of people with each condition. It is expected this study will now aim to support and test different ways of delivering coordinated care for people with rare conditions."

Rare Disease Day—Sunday, 28 February 2021

WDSG-UK held a virtual *Coffee Morning* on Rare Disease Day 2021 inviting all members of WDSG-UK, together with affiliated members of the Group. A report of the meeting can be found overleaf.

WDSG-UK Management Committee Meetings

During 2020-21 the management committee met twice via Zoom in September and January and our annual Support Group Meeting and 10th AGM took place via Zoom on 26 July 2020 (please see report overleaf).

WDSG-UK 11th Annual General Meeting

The 11th AGM will also be held via Zoom on **Sunday, 18 July 2021** at 1100 a.m, an invitation to which is included here in the newsletter. The accounts for 2020-21 and the minutes of the 10th AGM will be emailed to you separately. If you have any specific questions that you would like to submit to the committee, please let us know in advance. Election of officers and members of the WDSG-UK Management Committee for 2021-22 will take place then. Current members of the committee **Graeme Alexander, Mary Fortune, Liz Wood, Debbie Buckles** and **Valerie Wheater** have submitted their names for re-election for the coming year.

Next WDSG-UK Face to Face Meeting

We hope that in 2022 we may be able to reinstate our face to face gatherings in Cambridge and at last celebrate Dr Walshe's 100th birthday!

Emma Rachel Collcott 8.05.87—3.09.2020

A Poem written by her mum, Lesley

She was born, lived and died a fighter
She never gave up, no matter the challenges her illness brought
She never complained.

She touched the hearts of everyone she met
She gave inspiration to those who were struggling in their own lives
She brought hope and love to all around her.

She faced every difficulty with fortitude and bravery
She lost control over her body but not her mind
She lost old friends but made many new ones.

She knew what she wanted and was determined to live a normal life
She expected to be treated the same as an able-bodied person
She wasn't afraid to fight for the help she needed.

She wore us out with her demands
She kept us dancing to her tune
She kept us all together with her love and spirit.

Emma, we love, respect and are proud to have had you in our lives.

Wilson's Disease Support Group Meeting & 10th AGM
Sunday 26 July 2020 via zoom

The committee had planned to throw a 100th birthday party for Dr Walshe in Cambridge at the beginning of May and hold its AGM, a requirement of our constitution, immediately before it. In the event, Covid forced us to cancel the party and instead, committee member **Liz Wood** organised and hosted a Zoom meeting later in the summer. Invitations went out to all WDSG-UK members with their newsletter and we were pleased that 26 people registered to join us.

In particular, we were delighted to be joined by **Dr James Dooley** who has supported the Group since its formation in 2000 and **Dr Sam Shribman**, who is running the CROWD Study at Queen Square, London, in which many of you are taking part. One of the advantages of a video meeting is that members who find it difficult to travel to Cambridge normally were able to take part from the comfort of their own homes. This included **Caroline Barr** from Edinburgh and **Ashok Pandit**, a regular contributor to our newsletters, from Nepal.

Our new chairman, **Graeme Alexander**, conducted the business side of the meeting, giving us a report of work undertaken by the Group in the past 12 months. He told us that the BASL WD Special Interest Group was gathering apace with 85 representatives present at the meeting in London in November 2019. He also gave us an update on the work Public Health England is doing under NCARDRS into identifying the number of patients with Wilson's disease in England and he introduced us to **Debbie Buckles**, a patient he has known for 33 years, who has kindly agreed to come on to the committee.

There were no speakers as such, but Sam gave us an update on the CROWD study and his full report can be seen on *p14-15.* **Valerie** then presented us with the accounts for the year and the committee made up of Graeme, Liz, Debbie, Mary Fortune and Valerie was unanimously elected for 2020-21. Liz then divided us into separate *chat rooms* where we were able to catch up with one another more informally.

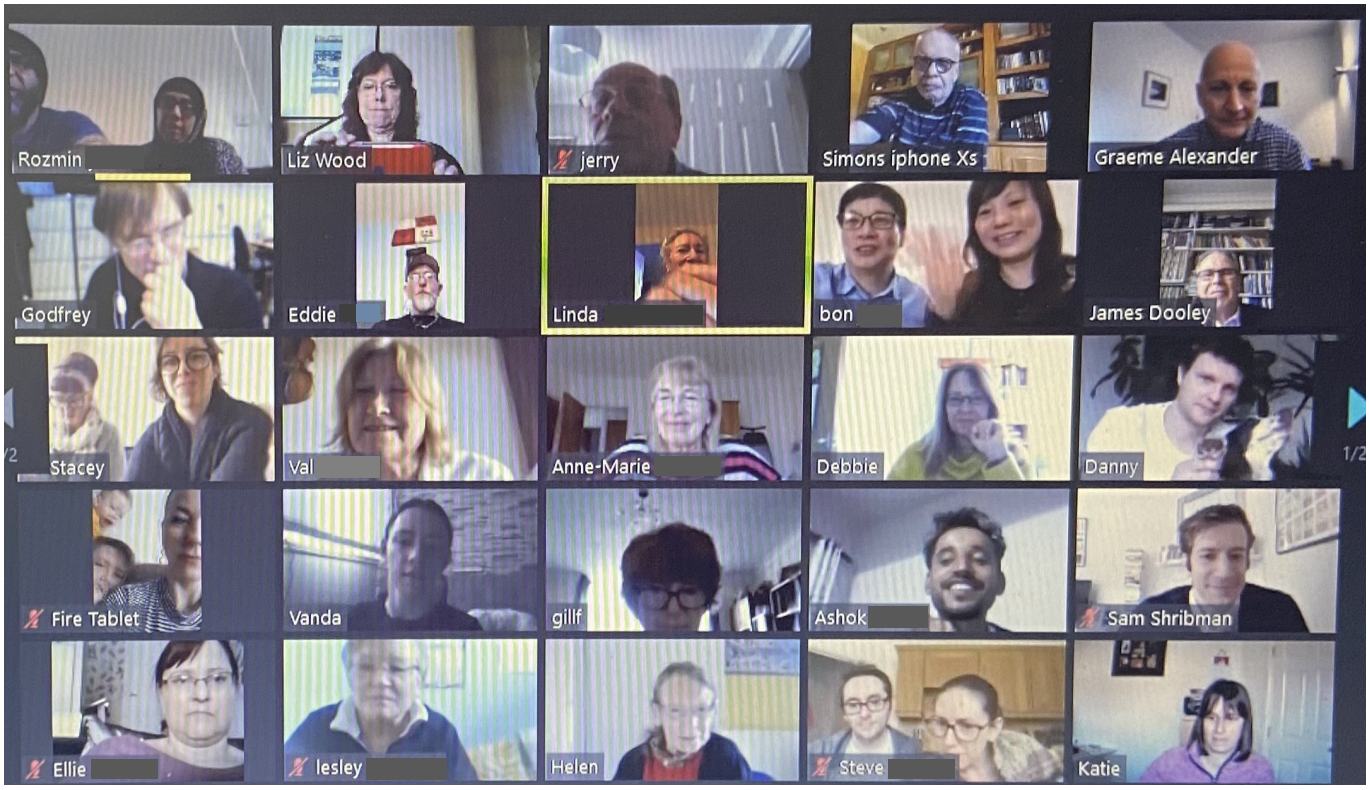
With the current restrictions in place, the committee has decided to abandon any idea of a formal annual gathering in the summer this year and will instead hold its AGM via Zoom on Sunday, 18 July 2021 at 1100 a.m. Copies of last year's Minutes and accounts will be emailed separately nearer the time, together with an invitation to attend. Joining instructions will then be sent to all interested parties.

Screenshot of some of the Attendees at the 10th AGM

Rare Disease Day—Sunday, 28 February 2021
WDSG-UK Virtual *Coffee Morning*

After the success of last year's Zoom AGM, the committee decided to hold an informal meeting to mark **Rare Disease Day 2021,** an occasion which is recognised globally every year on the last day of February. Its aim is to raise awareness of rare diseases and improve access to treatment for patients affected by them. Invitations were sent out to all current members, to doctors closely affiliated to the Group and to associated members through the WDSG-UK Facebook page. We had a fantastic response with 50 people signing up to join us including patients from as far afield as South Africa and Canada. In the former instance the time zone is the same, but in the latter Linda had had to set her alarm for 5.30 in the morning to be sure not to miss it!

Again, Liz Wood deftly hosted the event.



Some of the Attendees at The Coffee Morning with improved lighting techniques from Graeme, Debbie and Valerie!

We were keen for the meeting to be as informal as possible. However, with so many attendees we felt that splitting us up into pre-arranged groups from the outset was possibly the best way forward. So after an initial welcome by Graeme together with a speedy update on current Group business, we were divided into four *chat rooms* each headed by a doctor and somebody from the committee. This gave patients the opportunity to raise any specific medical concerns that they had or share any exciting news including the arrival of Matilda!

After twenty minutes, the screen alerted us to the fact that we had 60 seconds before we would be leaving that *chat room* and entering another—this time one which had been randomly selected by Zoom! It was a smooth transition, but we all found one or two people in our new chat room that we had just left in our previous one! Conversation continued until our hour was up and we came back together again. Graeme then wound up proceedings by inviting short summaries of any particular matters of interest that had come up in discussions.

Such was the success of this meeting, that we are hoping to hold another one in the Autumn. If you didn't receive an invitation to this meeting, then it is because we do not have your up to date email address. Please can you make sure that we do, so that you won't be missed out next time!

Thank you to all the attendees, in particular Drs Dooley, Gillett and Shribman who once again gave up their precious time to join us.

Fundraising 2020-21

Valerie

Like all charites, the Covid pandemic threatened our fundraising income last year, so we were particularly grateful to the following who, against all the odds and in a socially distanced kind of way, were able to raise much needed funds for us.

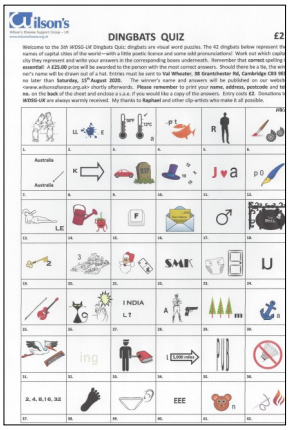
Katie Hibbard's Pebble Painting Phenomenon

Katie Hibbard took Blyton by storm last year when, at the beginning of lockdown and in the interest of entertaining the younger members of the community, it was suggested that children paint pebbles and hide them around the village for others to find. Pebble painting is something that Katie had never tried before, but wishing to join in with the spirit of the challenge, she purchased some acrylic paints and tried her hand. The results were amazing! The idea of the pebble hiding was that children could wander around the village looking for hidden pebbles and on finding them, pick them up and rehide them for others to find later. Unfortunately, Katie's were so popular that children were finding them and popping them into their pockets—never to be seen again!

A selection of Katie's hand painted pebbles

Realising there might be a commercial opening to this, Katie asked around and soon received several orders for her work. **£225** later and a house move and baby in the mix, her efforts have come to a standstill. However, should you wish to place an order, she would be happy to consider. Thanks Katie for all your hard work and for raising funds for us at this difficult time. We wish you a very **Happy 40th Birthday** for the 8th.

Valerie's Demon Dingbats - Capital Cities of the World



For the fifth consecutive year, Valerie provided us with yet more dastardly dingbats! This time the theme was Capital Cities of the World. Members receive them free with their newsletter, but as this has become a money making exercise too, it was imperative that Valerie was able to nobble the general public even in the middle of a Coronavirus pandemic! The only place that beckoned was the bank of the River Cam with people taking their daily exercise along it and also picnicking there.

When Rupert Brooke's Tea Gardens in Grantchester opened and people flocked to the deckchairs in the orchard, Valerie didn't waste a minute!! Over the following six weeks, she painstakingly collected over **£350** for Group funds, spending more time touting for sales of Dingbats than putting the whole of the newsletter together!

Of the 200+ copies distributed, 30 were returned with 5 all correct answer sheets including ones belonging to members Katie Hibbard and Stephanie Reid. Another member Ruth Morgan supervised the draw and **Andrew Jackson** from Cambridge was the lucky winner. He received a **£25.00** cash prize, together with a pat on the back!

Thank you to everybody who took part. For those of you who are still waiting for the answers, these are available on our website www.wilsonsdisease.org.uk. I had planned to take a year off this year, but with all this time on my hands during lockdown, I have somehow managed to produce another one. It is themed *Coastal Destinations in and Islands off the UK,* which if foreign travel restrictions aren't lifted this summer, we might all be resorting to! I hope you enjoy solving it; please let me know if you would like to sell some on the Group's behalf.

The Virtual London Marathon (VLM) — 4 October 2020

Another early victim of the pandemic last year was the 40th London Marathon which usually takes place in April. It was cancelled which was a particular blow to WDSG-UK as two people had registered to run in it and raise funds for us! In the end, the event was reincarnated later in the year when participants had to generate their own 26.2 mile course close to their homes.

The course...

Chris Billinghurst from Bournemouth, whose nephew, Ben, was diagnosed with Wilson's disease in 2018 at the age of 18, went ahead with the challenge on our behalf, having selected a suitable circuit near to his home. Afterwards he texted, "1st ever Virtual London Marathon. Tough conditions. Knee started aching about 11 miles in. Stopped to change socks about mile 20 as wet through, which reflects on official VLM app time. Ran past house 6 times. Adam J joined me for the last 10 miles and Jo, for last 2. Great support crew."

Chris and his wife, Jo—post run!

He completed the course in 5 hours 4 minutes and 17 seconds, burnt 2,410 calories and had an average heart rate of 141 bpm! Oh the wonders of smart phone apps! He reported only "slight aches" afterwards and is now raring to run further marathons including the Brighton marathon in September and the London one again this October.

We wish him all the luck in the world and thank him, his support team and all his sponsors for the fantastic sum of **£350+** (including Gift Aid) that he has raised for us so far through his fundraising page on *Virgin Money Giving*.

* * * * *

We should also like to thank **Seb Novell**, whose intention it was also to run in the London Marathon last April. Unfortunately, he was unable to take part in the postponed event in October, but is still in training and hopes to run later this year. He has so far raised **£300** for us—again through a fundraising page on *Virgin Money Giving*. Thanks Seb to you and your supporters!

Lynn Martin's Sale of WDSG-UK Wristbands

In 2019, before any of us had ever heard of Coronavirus let alone been affected by it, you may remember Lyndsay Kelly and her mum, Maria, climbed Ben Nevis in order to raise funds for us. Lyndsay works and is a close friend of **Lynn Martin**, whose 26 year old son, Jonathan, was diagnosed with Wilson's disease eleven years ago. She was keen to show her support to Lynn and to raise the profile of Wilson's disease within their local community in north-east England.

The WDSG-UK silicon wristbands

Lynn also felt that she would like to raise awareness of the disease and having read the article in last year's newsletter about the wristbands **Sam Panchal** had commissioned in 2017 and was selling on our behalf, asked if she could have some to sell too. Of course, this year has been a difficult year and Lynn has spent most of it working from home, but whenever she and her husband have had an opportunity to sell any, they have grabbed it with both hands raising a further **£220** for the Group. Lynn says, "Because of these lockdowns, it has been hard, but when normality returns we plan to do charity pub crawls to sell more wristbands and to raise even more awareness of Wilson's disease."

We thank them for that. Should anybody else be interested in selling any or wish to buy any (£2.00 each), please let me know.

Emma's Story

by her Mum and Dad, Lesley and Ian

Emma finally lost her battle with Wilson's disease and life on the 3rd September 2020, aged 33. We had offered to write something for the WDSG newsletter in the past, but she always refused. So, we are now putting words to paper on her behalf, with a small selection of stories of the last 16 years of her life.

Emma left school in summer 2004 after completing her GCSEs , tried college for a couple of months and then had a job in a clothes shop. She then went to work for Abbey National in Milton Keynes. Her hands had become red and she was diagnosed with Raynaud's syndrome, so her GP referred her to Milton Keynes University hospital (MKUH). Emma was also becoming very clumsy, dropping things, and stumbling. She had blood tests, a liver biopsy and an eye test. The next thing we knew a rather worried doctor was telling us she thought Emma had Wilson's disease. Ok, what's that? Our family found out over a very long period exactly what that meant for Emma and ourselves. She had cirrhosis of the liver as well as Kayser-Fleischer rings. She was referred to the Royal Free Hospital where we met Dr James Dooley for the first time.

Emma spent a couple of weeks in hospital having various tests and was prescribed trientine. She came home and went to the GP to get a sick note for work. To our surprise he gave her one for a year. He definitely knew something we didn't! That summer she started to deteriorate. She was living with her mum, as we had separated a year or so previously. Lesley was working part-time and during this period Emma was falling over more. Her arm would shoot up in the air, which she didn't seem to be aware of. Her mum told Emma not to go out alone, so as soon as her back was turned, Emma went out! Her behaviour was so odd; rumours started circulating that she was taking drugs. She was once picked up by the police who took her back to her dad's house where he had to deliver a lecture to them on Wilson's disease. Needless to say, she didn't tell her mum about this.

In September 2005 she deteriorated further and we asked Dr Dooley to admit her to hospital. She never came home to live after that. Within a couple of weeks, she could no longer walk or talk, she shook like a leaf and had problems swallowing. She had a catheter and a Percutaneous Endoscopic Gastrostomy (PEG) tube fitted. By Christmas, Emma was unable to control her body apart from her right arm which she raised up and down. The spasms stayed with her for the rest of her life though they were controlled with medication. Her legs became tight and her feet and toes curled over and twisted like a sickle, a feature of dystonia. With the exception of her final years, Emma was under the constant supervision of the Royal Free. She attended at least every 3 months and the development of what was diagnosed as the neurological version of Wilson's disease was monitored. Emma generously waived her right to patient anonymity so that her medical progress could be published and we hope this may be of some benefit to future sufferers. We also wish to thank again Dr Dooley who took a personal interest in Emma, liaising across the many specialities caught up by this illness, befriending Emma and keeping in contact with us.

In January 2006, once her condition had stabilised, Emma was admitted to the Capio Jacob Centre in Sawbridgeworth for rehabilitation. She was there for about 4 years. They succeeded in sitting her up, supporting her head with the help of botox injections administered at the Royal Free and teaching her to transfer to and from her wheelchair. She was able to eat with difficulty and was also partially PEG fed for a few years until she asked doctors to remove the tube. She was always choking - a constant worry, but Emma wanted to have a normal diet! By now Emma's manual dexterity was deteriorating and Royal Free suggested using an old medication which might help remove the excess copper from her brain. Emma readily agreed. The medication required specific approval by the National Institute for Health and Care Excellence (NICE) and is called dimercaprol. Dimercaprol, also known as British Anti-Lewesite, was developed during World War II to treat acute poisoning by arsenic, mercury, gold and lead. Unfortunately, this medication has to be administered slowly into a muscle daily, as it is suspended within a form of viscous peanut oil. There was always a risk of ulceration which Emma developed after 3 months. The treatment was stopped leaving Emma with a deep and painful wound which was slow to heal. She never complained and the treatment did enable her to use the keyboard on a modern mobile phone more easily for her remaining years.

She had met her boyfriend Ollie in the Royal Free and they were together for a few years. He took her on holiday a couple of times abroad, which frightened the life out of us! He also took Emma on trips from the care home. At least this gave Emma a semblance of normal life with someone of her own age.

It was a difficult time whilst Emma resided at the Sawbridgeworth care home as she was very angry. She refused to communicate properly with us and wouldn't allow anyone in authority to talk to us either. She was also a long way from home. Each journey took at least an hour each way so we weren't able to visit as often as we would have liked. Late one Sunday evening we each received a text message advising us she was 3 months pregnant, had split up from Ollie and was going to have an abortion in 3 days' time. Having verified this, we wondered if the abortion should be delayed for a few days. However, once Emma had finally agreed to let Dr Dooley talk to us, we were informed if she went ahead with the pregnancy, she may lose her life. They couldn't delay the abortion as Emma was already many weeks into her pregnancy. This was just one of many challenges we had to face over the years. She also contracted MRSA , C Diff and pneumonia, on several occasions, but always pulled through! By contrast, this last year she was sometimes on a hospital ward surrounded by Covid 19, but never caught that!

Emma was moved to another care home in Waltham Abbey where she stayed until 2015. Whilst there, she developed a twisted intestine, another feature of the dystonia which had already tightened her neck and distorted her feet and ankles. The poor girl was in the Intensive Care Unit at Queen Alexandra Hospital, Harlow where an invasive examination was undertaken daily until, on the sixth day, it was confirmed that blood supply to the twisted intestine was recovered and no further action was needed.

We were fortunate to be able to move her to a suitably modified flat in Bletchley, near to us, and we are very grateful that Emma was funded for 24 hour care throughout the rest of her life. She loved having her own flat. She liked shopping, going to the cinema, pop concerts and other day trips. We had a great long weekend break at the Calvert Trust Centre in Exmoor in 2019. Specifically set up for people with disabilities and their families, activities included cycling, canoeing, zip wiring, abseiling and bushcraft. She had a broad smile on her face all weekend. It gave us some lovely memories for which we were thankful as she started to deteriorate soon after. The last year of her life was difficult for her as she felt unwell most of the time.

Calvert Trust Centre Holiday, Exmoor—2019

Her liver had started to fail and her abdomen would swell with fluid and cause her discomfort. Over the last 2 to 3 years it was agreed that Emma's care and final days should be administered by the local hospital (MKUH). We were fortunate the lead hepatologist, Dr Prakash Gupta, who we understand studied under Dr Dooley, took the same interest in Emma and maintained a close dialogue with the family. As you may imagine, communication was challenging as Emma, the fighter, never lost hope in life and would certainly never give a medical power of attorney to anyone!

She spent most of last year in hospital having her tummy drained which caused a lot of infections. Although communication by her iPad became slower and more tortuous, Emma remained fully aware until her final week, asking why anyone voted for Trump and why no mass vaccination had started yet? Eventually she fell into a coma and a couple of days later, our beautiful daughter passed away. We had been able to visit her in hospital for the last 3 weeks of her life and we thank MKUH for allowing close contact throughout all the Covid 19 restrictions.

Everyone who knew Emma, admired her strength, determination, courage and resilience. She spent a long time in hospital but each time believed the doctors would sort her out. We wished Emma had tried to do more to regain her speech but she was always reluctant to try. However, with the use of her iPad she could command our attention and ensure we listened to her. She couldn't control her body well, but was very good at controlling everyone around her! Emma didn't want to be treated as a disabled person which did cause problems at times as she didn't want to make use of some of the aids that were available.

Her legacy will be the positive effect she had on those around her. She managed to smile and enjoy life in spite of her difficulties and inspired confidence in her parents when dealing with new people and situations. Her long term illness helped to put the trivialities of our own lives into context. All her family admired, loved and feel very proud to have known Emma.

Gene therapy for Wilson's disease?

In September 2020, Vertex, a company based in the USA, announced that they were planning to investigate a new approach to treating Wilson's disease using gene therapy. At first, they will study a small group of patients and follow them up over a long time. Vertex indicated that they planned to start in April 2021. Even if all goes as well as possible, it will still be a few years before we know if this treatment is effective. But the fact that they are willing to attempt such an ambitious and expensive study shows they think that gene therapy for patients with Wilson's disease is now a real prospect!

So what does this all mean? What are genes?

We inherit half of our DNA from each parent. The DNA is stored in *every* cell in the body as two non-identical strands. Each of the two strands of DNA contains numerous distinct patterns that determine and control over 20,000 genes that define each one of us. At any one time some of these genes will be silent depending on many things such as age— with different needs in childhood and as adults, events including infections, but also with position in the body. A cell in the skin has different needs from a cell in the liver or brain, for example. Some genes will be active continuously, others intermittently. When a gene within DNA is activated it generates a chemical called RNA. This RNA acts like an architect's plans—to create three dimensional proteins that are specific to the DNA of that individual. Mutations, or changes, in DNA within a cell (not the whole body) occur every day in response to damage and DNA is undergoing continuous repair. These mutations can cause changes to the proteins that the RNA creates. These mutations in response to injury and unchecked can drive diseases such as cancer.

Every one of us carries a small number of mutations in our DNA that arose between conception and birth and ensure each of us is unique. Some mutations have been beneficial; for example the gene that allowed humans to have larger brains than Neanderthals has been identified recently. Other mutations are lethal so the foetus cannot survive. But the great majority of gene mutations are minor and have little effect.

What causes Wilson's Disease?

Genetic diseases are a highly complex area. Some occur because an abnormal protein is produced that causes toxic damage. However others, including Wilson's disease, occur because the mutation in DNA creates RNA that then makes a protein that is missing a small but important component and so cannot function adequately.

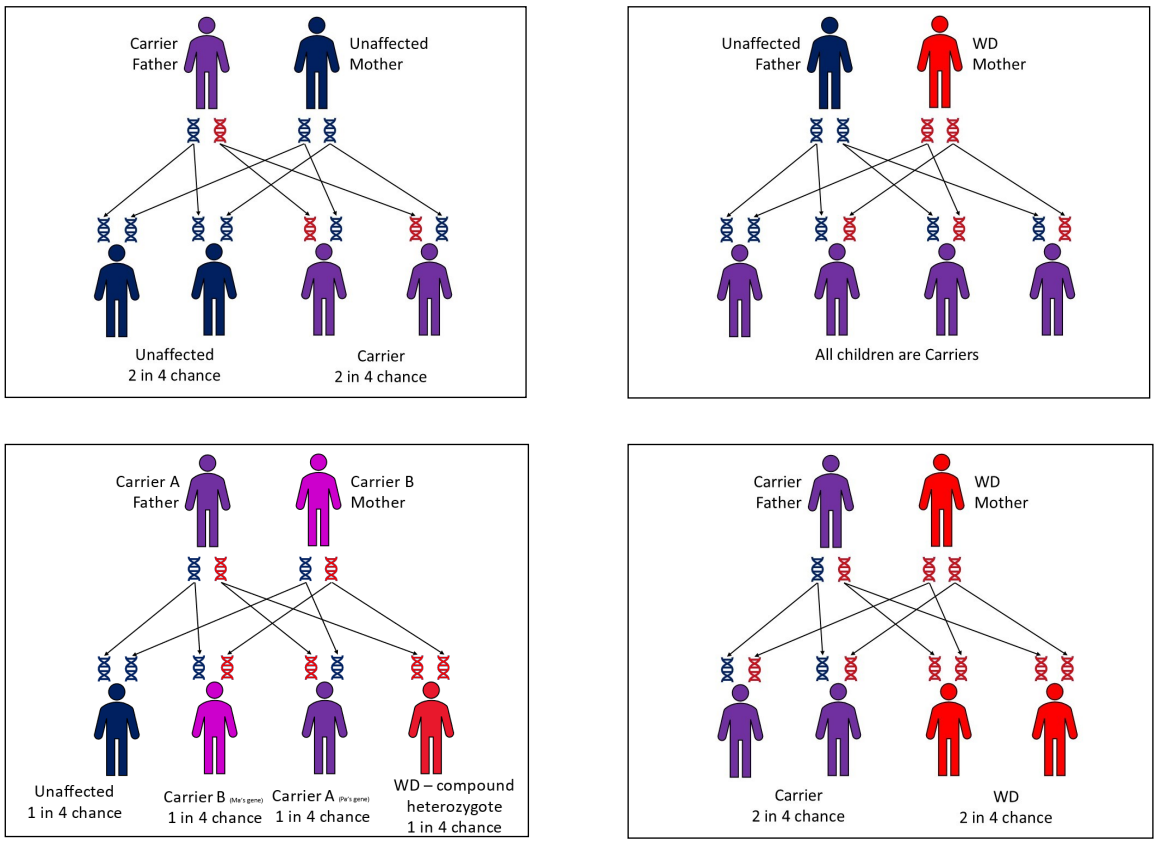
In Wilson's disease the critical gene is ATP7B. This should make a protein in liver cells that helps the body get rid of copper in just the right amount. However, in someone with Wilson's disease, the failure to move copper into the bile duct and then gut (and then down the toilet!) means that copper builds up in many organs and acts as a poison/toxin.

One good ATP7B gene will generate enough capacity to keep copper levels in the body at just the right level. This means that to develop Wilson's disease you need two mutated genes. Estimates for the UK suggest that between 1 person in every 7,500 and 1 in every 30,000 people has two mutated WD genes – i.e. no good ATP7B gene. Over 800 different Wilson's disease related mutations of ATP7B have been found. Some Wilson's disease patients will have *identical* mutations of the ATP7B gene and are known as homozygotes. This is more prevalent in cultures where cousins may marry, but most patients will have inherited *different* mutations from each parent and these patients are known as *compound* heterozygotes.

A *simple* heterozygote is someone who carries one mutated gene and one normal gene so is healthy and disease free and these people are referred to as "carriers". They *carry* only one mutated gene so do not have Wilson's disease themselves. There are between 1 in 48 and 1 in 90 such people carrying just one mutated ATP7B gene in the UK. This is important to consider when someone with Wilson's disease is planning to have children; the risk of having a child with two mutated genes is small but real; for someone with Wilson's disease with a partner who just happens to carry one mutated gene each child would have a 50% chance of inheriting two mutated genes. Fortunately, the chance that a partner is a carrier is quite small.

- The gene involved in Wilson's disease is ATP7B
- There are over 800 different Wilson's disease related mutations of ATP7B
- There are between 1:48 and 1:90 WD carriers in the UK i.e. have only one mutated gene
- Wilson's disease patients have inherited two mutated ATP7B genes—one from each parent
- It is suggested that there are between 1:7,500 and 1:30,000 people in the UK with 2 mutated ATP7B genes, i.e. have Wilson's disease
- A simple heterozygote has one mutated ATP7B gene and is known as a carrier
- A homozygote has inherited two identical mutations of the ATP7B gene; one from each parent
- A compound heterozygote has inherited two different mutations of the ATP7B gene; one from each parent.

Charts Showing Inheritance Patterns in People with Mutated ATP7B Genes



Gene Therapy—Approaches and Challenges

A couple of decades ago the first hurdles to gene therapy were getting the 'treatment' into the right place in the body and then finding a way to keep it there—the human cell responds to new genes as if they are viruses and tries to eliminate them.

The first uses for gene therapy have been in the field of stopping genes where mutations produce a toxic protein and after about 25 years of research these have led to life changing developments. However, in Wilson's disease the need is the opposite—we need to introduce a gene that correctly makes a protein to transport copper out of the liver and into the bile duct and then gut.

Also, researchers had to find a way to get the treatment to exactly the place where it was needed in the body—in Wilson's disease this is the liver. Some approaches for the liver link the gene therapy treatment to a protein that the liver can absorb. This has been effective. But more often currently the gene is inserted into an Adenovirus that can get into the liver before releasing the gene correction therapy (in much the same way as the Oxford/AstraZeneca Covid vaccine which is made by inserting the Covid Spike protein into an Adenovirus). Much work has now identified the right type of Adenovirus for efficient delivery of the right gene and sophisticated techniques have been developed to neutralise the Adenovirus itself, without impairing delivery of the gene, so the Adenovirus cannot cause an infection.

There are huge hopes for medicine in general using a technique called CRISPR whereby a specific gene is targeted, the small abnormal area is removed and at the same time the correction is inserted; the early work on CRISPR deservedly won a Nobel prize. But in Wilson's disease there are so many different mutations to correct specifically that this approach may not be very easy to apply across all cases.

Over the past few years modified Adenoviruses have been used to deliver a normal Wilson's gene into mice to correct mouse models of human Wilson's disease. These experiments have gradually been improved so that now the pharmaceutical industry is poised to replicate the process in humans and will use a 'neutered' Adenovirus to deliver the normal version of the Wilson's disease genes into the livers of patients with Wilson's disease.

There are many things that cannot be predicted. No-one knows the answers to the questions: Will it work? Will it have side effects? How long will it last? Will it be good enough to stop conventional treatment and more? No-one knows with certainty because this is, by definition, research. But it is a step with huge potential.

Drs. Mary Fortune and Graeme Alexander

Debbie's Story

by Deborah

I am Deborah (some of you may know me by my maiden name Nunn). I was recently invited to join WDSG-UK by Graeme Alexander, who has not only been my Consultant at Addenbrooke's Hospital post liver transplant, but also a huge support and influence in encouraging me to live life to the full!

My story starts in 1987 when I was 22 years old. I had visited my GP on a few occasions feeling tired, breathless and generally "out of sorts." My job at that time was quite demanding providing cover for holidays and staff absence at concession floristry shops on the USAF military bases throughout the UK. Hands-on industry experience in Holland at the Dutch auction markets and a busy social life made it seem logical, therefore, that I was simply tired and "run down."

After a restful holiday, and weeks of early nights, my condition had not improved. I collapsed at home in the bathroom, and my mother insisted on accompanying me to the GP's. I had hardly made it through the door, when I collapsed again, and was moved to a side room, given a sweet cup of tea, and examined. The GP I saw (Dr. Richard Verill) did ask if we had heard of Wilson's disease, which we hadn't, but it was dismissed as unlikely and exceedingly rare!! I was sent home to rest in bed with suspected hepatitis.

Later that same day (23rd July 1987), another GP called at our home after blood tests taken earlier that day showed my blood count down to be just *4*! I was rushed to Ipswich Hospital and admitted to an isolation ward for suspected infectious diseases. After two weeks with numerous tests and blood transfusions, I was sent home; the diagnosis being *a virus or mystery illness!* My consultant described my blood results as being like stocks and shares - constantly on the move - up and down!

Although still very weak and unsteady, I managed two nights at home in my own bed before the hospital rang calling me back in, having diagnosed Wilson's disease after receiving blood results back from Cardiff. I was started on penicillamine and discharged after a week.

During the next six weeks I developed ascites (a build up of fluid in the abdominal cavity) and looked heavily pregnant! During the last few days at home, I was having to climb upstairs on my hands and knees to the bathroom. My breathing had become difficult, due to the pressure on my lungs from the ascites.

On Friday 4th September, back I went into Ipswich Hospital, where despite all their efforts, my condition deteriorated further. It was decided to send me to Addenbrooke's under the care of Dr John Walshe. There was a very tentative mention of transplantation, but at no point did I take it seriously, after all transplantation was something you read about in newspapers, and certainly not something that happened to ordinary people like me!!

On 9th September 1987, I arrived at Addenbrooke's Hospital. The treatment with penicillamine continued, but my liver was too damaged to respond. I remember attracting enormous interest from students due to the Kayser-Fleischer rings in my eyes: I had never been so popular! However, my memory of this time begins to falter now, as I was starting to lapse into coma. The last thing I remember was carrying a huge urine container back from the bathroom to my bed, as my copper excretion was being constantly monitored.

> *"They were told there was nothing more that could be done...I could die at any time"*

My story continues with help from my now husband (boyfriend then) and mum. Apparently, I was moved into a side room, so that my parents and husband could stay with me 24 hours (Addenbrooke's provided a room for them to sleep in.) They were told on 17th September that there was nothing more that could be done, my condition was terminal and I could die at any time.

At around 6pm on 22nd September Prof. Calne, accompanied by his team, arrived in my side room and examined me. He led my family out and told them there was a liver available, which with their permission he was prepared to transplant into me. He didn't want to discuss it in front of me in case I could still hear. It fell upon my father to give permission, as I had previously told my mum that I didn't want an operation and had pleaded with her not to let them cut me.

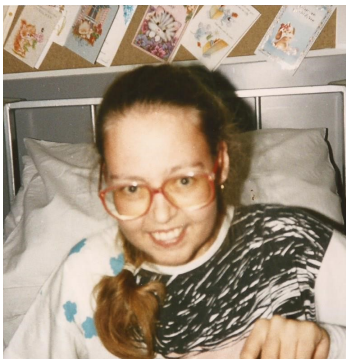
After assurances from Prof that were it his child he would go ahead with the transplant, my dad agreed to the operation, explaining to me months later that he had to give me a chance. He also dined out for years on the fact that it was the first time in his life that he'd had the final word over my mum!

I've been told that once permission was given, the task of 'building me up' for the transplant began. Obviously, I was completely unaware of this. My understanding is that my transplant started at 6am, and lasted 6 hours. My parents and husband were told in the morning that I had made it through the operation and was in intensive care. I learned later that I had to be taken back to theatre because of severe bleeding.

Back in intensive care for the second time, my recovery began. My first recollection is of muffled sound, almost like swimming under water. Gradually it became clear that I was still in hospital. I realised that something had happened due to the machinery around me. I looked down to my stomach and saw what I imagined were large black spiders. I realised that was completely stupid, and decided that I'd had an exploratory op to find out what my problem was! It wasn't until my dad, who was never a demonstrative man, clasped my hand and said, "We love you: Keep fighting!" that I realized something serious had happened. He then told me "You've had a liver transplant; but try not to think about it at the moment!" I can honestly say I accepted the situation from then, and have never felt anything other than gratitude since.

During my recovery, I had a stroke and a tracheotomy (incision in the windpipe) so my movement was limited and I had no speech! I was fortunate to have a ward orderly who was deaf and could lip read. She acted as translator for me on several occasions. Visiting was limited to family only and a visit every 2 hours: mum each time, dad and husband alternately. Sometimes a visit would only last 2 minutes depending on how I felt, at which point my visitors would receive the royal wave.

I had the aid of a child's letter board, for spelling out words—no computers in those days! Communication for me at this point was so important. The facial expression of my nurse could change my mood, as I would interpret the look that she gave as an indication as to how I was that day. A smile, and a stroke of the arm would be so reassuring that all was well.



Ward C9—After the transplant

After 4 weeks in intensive care, it was decided I could be moved to C9 despite contracting chickenpox. I had hoped that it would keep me in intensive care for a few more days! It was a great step foward but a huge wrench for me as I was leaving the secure and safe environment of ITU. I moved on the day of the great storm 15th October 1987! Once on Ward C9 the long road to recovery began, and my family's role changed to force feeders, and the mantra "one more drink and I'll make you another" became almost mechanical! That and the regular visits from *physioterrorists* as I called them, helped and encouraged me every day to get stronger, and work towards the goal of going home.

> *"It's like being given a top of the range sports car…"*

As part of the Transplant programme, I had to go to King's in London for part of my recovery. I spent a week there, during which time my mum stayed in the hospital with me. My appetite continued to improve to the point that my husband's friend claimed he'd never seen anyone eat a bag of crisps so quickly!

I finally came home two weeks before Christmas, making it the best Christmas present any of us could have hoped for. I have often been asked what it's like to have been given a liver transplant. My response is that it's like being given a top of the range sports car: you want to take it to its maximum, but you know you're looking after something precious, which should be taken care of and treated with respect.

I do my best to get the most out of every day. I work full time, still in the florist industry, and pack each day as full as possible. I try to be an ambassador for the Transplant programme, and although there were only 7 weeks between diagnosis of WD and my transplant, I am looking forward to learning more about this rare disease as a member of the WDSG-UK committee.

I am often asked how has it changed my life? It has given me a life! Were it not for the courage and conviction of pioneering surgeons and dedicated medical staff, and the decision of my donor family, I simply wouldn't be here today.

I have enjoyed many special times that my transplant has made possible including getting married, sand surfing in New Zealand, a USA road trip, holidays and days out with my niece and nephew, and the closest relationship and friendship that I could have with my mum. There have been hiccups along the way, but it's thanks to all the Transplant team for their constant support and positivity that I am here today. I am forever grateful and look forward to celebrating my 34th *Tranniversary* on September 23rd - God Willing!

WDSG-UK Research, Notices & Updates

Research

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

The CROWD (Cohort Research On Wilson's Disease) Study was launched by **Prof. Tom Warner** and **Dr Sam Shribman** in December 2018. In the first part of the study, participants from around the UK have been completing an online questionnaire and sending in saliva samples for genetic research to understand why Wilson's disease affects the brain in some people but not others. The team is still looking for more people who might be interested in completing the online questionnaire but have temporarily stopped collecting saliva samples due to the pandemic. More information about this part of the study is available at www.thecrowdstudy.com for those who are interested and thanks to all those who have taken part so far through WDSG-UK's Patient Register.

The second part of the study is focussing on how to monitor the effects of Wilson's disease on the brain. Forty participants have already attended the National Hospital for Neurology and Neurosurgery for clinical assessments, urine and blood tests and an MRI scan of the brain. This part of the study is now closed. Below is an article describing some of the initial work developing blood tests to monitor neurological involvement in Wilson's disease and the team is currently submitting its initial work on brain imaging for publication in medical journals and hopes to share some results with you soon.

Can blood tests measure neurological involvement in Wilson's disease?

How do we currently measure neurological involvement?

The extent to which the brain is affected in Wilson's disease is highly variable and we currently measure this by performing a bedside examination. This involves assessing speech, coordination, walking, balance, etc. In research studies and clinical trials, a scoring system such as the Unified Wilson's Disease Rating Scale (UWDRS) can be used to give these examinations a numeric score. However, any improvement or deterioration in the bedside examination usually lags behind the underlying changes in the brain. In the CROWD study, one of our aims is to identify novel approaches to monitoring the effect of Wilson's disease on the brain. Our hope is that brain-specific tests could give us much earlier confirmation that a treatment is working, predict how patients will respond to treatment and test the effectiveness of new treatments in clinical trials.

How can a blood test be used to measure disease activity in the brain?

One approach to measuring the effect of a disease on the brain is to look at proteins released from neurons, i.e brain cells, when they are stressed or damaged. Each neuron produces thousands of different proteins and specific proteins can usually be measured in the cerebrospinal fluid, which sits around the brain. However, a procedure known as a lumbar puncture is required to get a sample of cerebrospinal fluid and this is not practical on a regular basis. In the last decade, it has become clear that some brain-derived proteins can be detected at tiny concentrations in blood. Measuring the quantity of these is extremely challenging given they make up less than one billionth of the mass of all proteins in a blood sample! An ultrasensitive test known as a single molecule array, or SIMOA, assay is required.

How have we investigated this in the CROWD study?

We tested blood samples for four brain-derived proteins including NfL, tau, GFAP and UCH-L1 using the SIMOA assay in a highly specialised laboratory based at the UCL Queen Square Institute of Neurology. We used samples collected from 40 people who had visited the National Hospital for Neurology and Neurosurgery as part of the study. The study included adults aged 16-68 years, the majority of whom had been treated for Wilson's disease for many years.

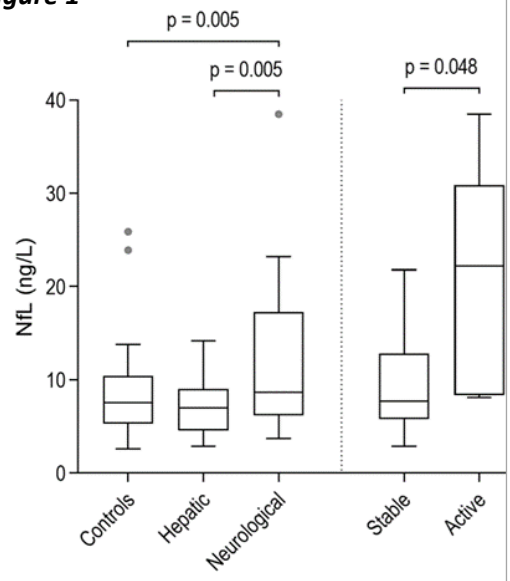
What did we find?

Firstly, (***Figure 1***) we compared the concentration of these proteins between 23 people who had neurological symptoms at the time of diagnosis (neurological group), 17 people who did not have neurological symptoms at the time of diagnosis (hepatic group) and 40 healthy individuals (control group). We saw clear differences in the concentration of NfL but not tau, GFAP or UCH-L1. NfL levels were higher in the neurological rather than the hepatic or control group. NfL is known to increase with age in healthy individuals and the differences between the groups became much clearer when we factored in the age of participants.

We then compared 5 patients with active disease to 35 patients with stable disease, defining those patients who had recently being diagnosed with neurological symptoms or who had recently deteriorated neurologically, as having active disease. Here, we controlled for age and neurological severity. Again, we saw clear differences for NfL with higher concentrations in the active group.

Finally, (***Figure 2***) we compared protein concentrations to the UWDRS neurological examination scores, a measure of neurological severity in stable patients. We found that there was a close correlation between neurological severity and NfL, tau and UCH-L1 concentrations.

Figure 1



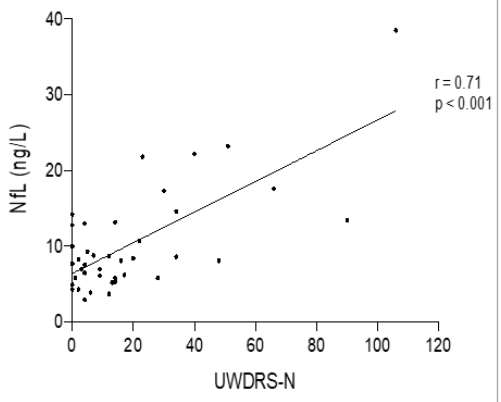
Box plot showing differences in NfL concentrations between groups. *The horizontal line in the middle of each box is the median (average) NfL concentration and the rest of the box indicates the interquartile range (spread).*

What is NFL and what does this mean?

Neurofilament light (NfL) is a scaffold protein, which means that it supports the internal structure of neurons. It has previously been studied in several neurological conditions over the last few years and has been shown to be increased in blood samples from people living with multiple sclerosis, head injury and some causes of dementia. It is not therefore specific to Wilson's disease.

Our findings suggest that NfL measured using the SIMOA assay is a promising blood test for monitoring neurological involvement in Wilson's disease. However, there are several unanswered questions and issues that need to be addressed before we could consider using this in everyday clinical practice. Firstly, we need to understand how NfL concentrations change over time in individuals who are starting treatments like Penicillamine, Trientine and Zinc. Secondly, NfL concentrations increase with age and we don't currently have reference ranges that allow us to say whether a specific value is normal or abnormal for a given age. Thirdly, the test requires specific equipment that is, to our knowledge, only available in a few, highly specialised laboratories.

Figure 2



Scatter plot showing the association between NfL concentrations and the UWDRS-N score, a measure of neurological severity.

What next?

Our findings have recently been published in *Movement Disorders*, an international medical journal for neurologists. The full scientific paper is available here (https://doi.org/10.1002/mds.28333) for those who are interested. We have also given presentations on our findings to the Association of British Neurologists and the British Association for the Study of the Liver Special Interest Group. The next steps are to study how NfL concentrations change over time in people living with Wilson's disease. This will require further research studies in the next few years. Clarification of age-specific reference ranges and whether SIMOA assays could be used in the NHS is likely to be driven by research on more common neurological diseases.

Dr Sam Shribman, Miss Maggie Burrows and Prof. Tom Warner
UCL Queen Square Institute of Neurology

Research (cont.)

University of Sheffield, Dept of Neuroscience

Prof. Oliver Bandmann and his team have had to put on hold their 31P-MR spectroscopy imaging work investigating mitochondrial biomarkers in Parkinson's disease and other neurodegenerative diseases, including Wilson's disease because of Covid. Access to the MRI scanner is still restricted and so is overall research capacity. However, things are slowly improving and their aim is to have all the scans, including those funded by WDSG-UK for Wilson's disease patients, completed by October 2021.

The idea of the study is to compare brain imaging in patients with neurological WD with patients with hepatic WD. It is predicted that there will be a more marked impairment of the cell batteries, the mitochondria, in the brains of patients with neurological WD. Several Wilson's patients have already agreed to participate in the study, but others may be needed. **Please let us know if you are at all interested in taking part.**

Meanwhile, Dr Tom Payne, a clinical fellow, has been using the lockdown time to analyse the many 31P-MRS scans which have already been done in Parkinson's disease patients and age-matched controls. His work has helped optimise the analysis strategy.

Drug Trial — Alexion, London

Copper Balance in Participants with Wilson's disease treated with ALXN1840

Alexion Pharmaceuticals is trialling an alternative treatment for Wilson's disease which is known as ALXN1840 (bis-choline tetrathiomolybdate; formerly known as WTX101). It is a novel copper binding agent in development for the treatment of Wilson's disease. It has recently embarked on a study in the UK called *The Copper Balance Study,* which is open to patients over the age of 18 and which runs for six weeks. During this time, patients will stop taking their usual drugs for Wilson's disease and take ALXN1840 instead. Patients will be required to stay in a research facility, Richmond Pharmacology, in London, under the care of dedicated healthcare professionals, for two periods of 17 days each. During this time patients will be served a set menu which they must eat (there are choices) and copper excreted in their urine and faeces will be collected and measured. Blood samples will also be taken. In between these two inpatient stays, patients will be expected to spend 14 days sticking to a low copper diet. Not everybody will be eligible to take part. Patients will need to be put forward by their Wilson's disease specialist before further screening by Richmond Pharmacology.

If you are interested in taking part in this study, please contact WDSG-UK in the first instance. There is considerable recompense offered for your time and inconvenience.

Supply of Meds in the UK (April 2021)

D-Penicillamine

We are not aware of any current manufacturing or distribution issues with penicillamine in the UK. Please let us know if you have any problems obtaining it.

Cufence® 200mg previously known as Trientine Dihydrochloride capsules 300mg

In November 2020 Univar Solutions changed the name of trientine dihydrochloride capsules 300mg to Cufence® 200mg after the granting of a European licence from the European Medicines Agency. The capsules are all but the same but the weight stipulated on the packaging now only refers to the weight of the active ingredient, trientine. Trientine dihydrochloride 300mg is made up of 200mg trientine plus 100mg of the salt base dihydrochloride. There is one subtle difference, however. Univar has found that Cufence® does not need to be stored in a refrigerator **UNLESS** or **UNTIL** the seal on the bottle has been broken. This should be helpful to patients in the future. For further details, please refer to the literature that accompanies the capsules. There are no production or distribution problems with this product to our knowledge.

Cuprior® (Trientine Tetrahydrochloride) made by Orphalan Ltd

For patients in the UK taking trientine, it may be of interest to you to learn that an alternative trientine preparation, trientine tetrahydrochloride (Cuprior®) has been licensed across the UK. It does not need to be refrigerated at all and is supplied as scored splittable tablets in blisters, each pack containing 72 tablets. There are no production or distribution problems with this product either.

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

Through the British Association of the Study of the Liver (**BASL**) a number of Special Interest Groups (**SIGs**) has been established in certain areas of liver disease in order to bring relevant expertise together from around the UK. The **WDSIG** was one of the first to get going and 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 Public Health England (**PHE**). Over 100 individuals from across the UK are registered on the WDSIG database and the SIG has now approved 15 specialist WD adult centres across England. For further information on the remit of the WDSIG and minutes of its meetings, please visit the BASL website www.basl.org.uk. Additionally notes on the SIG are documented in the 2019/20 copies of this newsletter (*p16 & p11* respectively) archived on our website www.wilsonsdisease.org.uk.

5th WDSIG Meeting—20 November 2020 via *Zoom*

The 5th meeting of the WD SIG took place remotely. In fact it was the best attended so far (45 people) which might say something about our new found friend 'Zoom'!

The first topic, and perhaps the most important topic of the day, was a discussion around production of **UK guidance/ guidelines for WD** which has always been a desired output from the SIG. It was felt that practical guidance for the 'jobbing' physician would be simpler and faster to produce, based on UK expert opinion and available evidence. The guidance will be offered for dissemination and publication by the two main specialties - Gastroenterology (Hepatology) and Neurology. Since the SIG meeting, Prof Oliver Bandmann has kindly chaired the guidance working group which consists of three main subgroups - adult hepatology, adult neurology and paediatrics - with primary input from our excellent trainees across all subgroups. Progress has been rapid and we anticipate a working draft soon after Easter for review by the SIG.

The WD Public Health England (PHE) NCARDRS project continues to progress really well. Dr Osob Mohamed, Hepatology senior fellow at Royal Surrey Hospital, presented the latest data across 27 Trusts (with many more to come on board) which since 1997 included 470 patients with WD, 372 of whom are still alive. In addition and over the same period, preliminary data on 84 patients who underwent liver transplantation for WD were also presented. Further data sources are in evolution which will enhance the capture of patients across England. Mary Bythell (PHE) has been instrumental in driving the WD project forward here and our thanks go to her and her team.

Research into WD was a continuing theme with an update from Dr Sam Shribman on the CROWD study where he has found a potential 'biomarker' (a test which distinguishes different types of WD patient). We congratulate Sam on the publication of this work in the journal *Movement Disorders*. Dr Tom Marjot, a hepatology trainee from Oxford and new to the SIG, eloquently presented the latest international research and Prof Aftab Ala updated the SIG regarding the various clinical trials in WD which are either currently open or in set up/planned for the future. Included in the latter group are gene therapy studies which are a promising way of correcting the inherited abnormality in WD directly.

We invited the paediatricians to highlight some of the issues they face and Dr Tammy Hedderly from Guys & St Thomas Hospital presented cases illustrating delay in diagnosis, a possible link with autism, compliance and the need for psychologiocal/psychiatric input. Prof Anil Dhawan gave an overview of the treatment of children at Kings College Hospital and highlighted their concerted multidisciplinary approach which may reduce the need for transplantation (full details of his clinic appear on *p23*).

Dr Miranda Durkie from the Sheffield Children's Hospital Genetics department updated the SIG regarding the new **National Genetic Testing Service** now up and running in England. The service will streamline the whole process, improving the quality, consistency and timeliness of reporting as well as providing easier access to testing for physicians. This should be seen as a welcome step forward for diagnosing suspected WD.

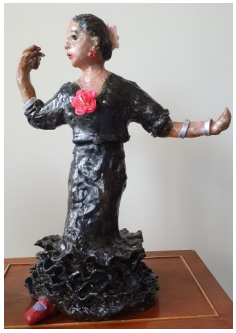
The meeting concluded with a discussion regarding the various trientine formulations available. Rupert Purchase elaborated on this topic for the benefit of the SIG. Paul Selby, lead liver pharmacist in Cambridge, gave a helpful overview of the new pathways which allow smaller centres to link in with the Specialist Centres in England for the prescribing and oversight of patients on trientine.

Bill Griffiths,
Consultant Hepatologist at Addenbrooke's Hospital, Cambridge, and WDSIG lead.
21.03.21

Members' News 2020-21

Our President and world renowned Wilson's disease specialist, **Dr John Walshe**, has finally got over the excitement of celebrating his 100th birthday during the first lockdown last year and now looks forward to being 101 in a couple of weeks time! To relieve the monotony of staying at home throughout the pandemic, he had a short spell in hospital just before Christmas.

Asked how he is now, he replied, "ticking over quietly". He remains living in the family home in Hemingford Grey and is grateful to his daughter Susan and son-in-law Phil who look after him extremely well. He is now embracing the convenience of technology and enjoys Skyping every Monday evening with his younger daughter, Clare. In July he will become a great grandfather for the first time which he is very much looking forward to. His genes live on!



Linda Asher has been exceedingly busy. Diagnosed with neurological Wilson's disease in 1957 at the age of 13, she has always been good at art. Most recently, she received a 2nd class honours degree in Creative Arts at the age of 60! She has done a lot of painting in the past and you may remember that last year she was waiting for her clay modelling class "The Crackpots" to reopen so she could finally get to use the kiln and fire the Spanish dancer she had so lovingly created. Since then she has made other clay models including one of her orthopaedic surgeon's dog, a parrot for her grandson and Doris Day as a little girl for herself!

In August last year, Linda bought herself a simple wooden dolls house kit from *Minimum World* and then rang her twin brother, David, to ask if he would mind *quickly* assembling it! Two months later and over 250 hours spent building it, he finally finished the project! The work involved wallpapering, flooring and wiring to Linda's design. In addition, he constructed a pull out garden section underneath and Juliet balconies on two of the windows, he added internal doors, skirting board and window sills, banisters and balustrade, carpet trims, plug sockets and picture frames and to top it off, he added *ridge tiles* to the roof and *lead flashing* to the chimneys! The house now has 41 light fittings with 61 bulbs! Asked how he enjoyed the experience, he replied "Never again!"

Linda, on the other hand, claims that **she** had the hard bit in designing and furnishing it all! So pleased with the results, Linda posted the finished product on Minimum World's Fb site <https://www.facebook.com/MinimumWorld/posts/3748482118541821> and it has now become one of the most commented on posts they have ever had! Do take a look: it's simply breathtaking!

Juliet balconies and pull out gardens

With the front hinged open

All mod cons, including a tv made by David!

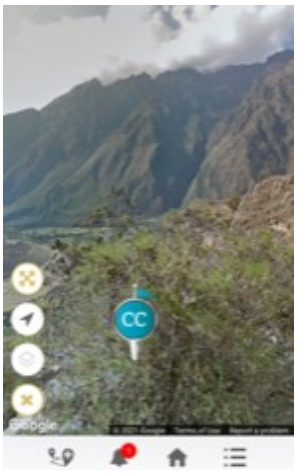
Caroline from Edinburgh, whose adventures on the high seas were featured in the 2019 newsletter, now brings us up to date with what she has been doing during the pandemic. She writes,

My lockdown—About a year ago my mental health took an enormous crash which ended up with me being hospitalised for two nights. Lockdown was scaring me; I was on my own and my support framework was crumbling before my eyes.

Mum and dad came to my rescue and my cat, Sam, and I moved in with them back to the village where I grew up! Not easy for a person who is usually very independent. It was supposed to be for a few months, but it has ended up being a year!

What has saved the day is a *Zoom* Book Group which I have joined. I have read books that I never would have read and enjoyed them all. It has given a focus to my day. The book group is run by the *Workers Education Association*.

I have also signed up with the *Conqueror*, which is an exercise app which virtually takes you on a trail of a length of your choice. I have chosen the Inca trail in Peru through the Andes mountains to Machu Picchu. It is 42km long (26.2 miles) and I am walking it at 0.5 km a day. At the end of the challenge, I get an impressive medal!"



Not looking forward to the ascent!

Laura from Shetland whose harrowing story of diagnosis was featured in last year's newsletter, writes

Hello all, I thought I would update you on my progress. You might remember that I developed nephrotic syndrome from taking penicillimine so had to change to trientine ten months into my treatment. My kidneys have taken 14 months to recover, but I am happy to report that I have now been discharged by my kidney specialist and am feeling better than I have felt for many years!

Meanwhile, I have secured a full-time permanent job as a Learning Support Worker, working with children that have autism. I have a boyfriend now too, so I am very happy. I hope to travel down to London in May to meet Dr Gillet again, but it will depend on the Covid situation. I have had my first Covid vaccine and am awaiting the second. I hope everyone is doing ok and is looking forward to getting back to some sort of normal. All the best, Laura.

A bouquet of cupcakes for my birthday last year!

Olivia is our youngest member and her mum and dad write,

It has been quite a challenging year for Olivia as she prepared to sit her 11+ exams and take academic and music scholarships to various schools here in London. While her friends were spending time with one another (as Covid permitted) or went on holiday, Olivia was busy studying at home and playing her musical instruments (piano and violin), which she loves. It was made particularly difficult as many of the exam dates were changed at the last minute because of the pandemic, making the whole process even longer!

Olivia arriving for one of her exams

Tilly

However, her hard work has paid off and Olivia has accepted a place at one of the Grammar Schools in North West London that she particularly wanted to go to. She will be starting there this September. We are so proud of her achievements particularly as she was diagnosed with Wilson's disease when she was only two and has had to live with her meds regime, diet restrictions and hospital appointments ever since!

Since taking her exams, we now have an addition to the family by way of Tilly, a Yorkshire Terrier, whom we all love. She arrived just before Christmas and has been a constant companion to Olivia throughout this latest lockdown. We look forward to seeing you all again on the next *Zoom!*

Sue was diagnosed with Wilson's disease 38 years ago. She takes penicillamine and has spent most of her working life in the NHS. She writes,

Surviving Covid: I retired over 4 years ago but after a couple of years travelling, decorating, and gardening I needed something for my brain and currently work as a bank staff nurse for our local hospice. I live on the south coast of England and have spent most of the pandemic walking on my days off. In September I walked the *South Downs Way* with a friend. We covered 100 miles in 9 days and saw some stunning scenery. We had willing husbands dropping us off and picking us up on each section and finished in Eastbourne on the most glorious of days.

Nearly there...

During the pandemic we have had very restricted visiting at the hospice and although we are wearing full PPE, this January we sadly had a Covid outbreak and 36 of us went down with it like ninepins. The hospice had to close to new admissions for a couple of weeks and now has even tougher visiting restrictions. Prior to the outbreak we had just started doing lateral flow tests and weekly PCR testing. Despite 4 negative lateral flow tests, I tested positive on 19th January 6 days after my first vaccination.

Initially I felt as if I had a cold virus starting and locked myself away in our spare room for 10 days. My husband Chris delivered everything I needed to my door. I had to text him as I had lost my voice! I didn't have a fever or much of a cough and only a mild loss of taste. However, I did have the most awful sore throat, no voice and an incredible chill. All my bones ached, and I had no energy. I am usually a busy, active person but had to spend the first week in bed. I was off work for about 5 weeks and gradually my energy came back and my voice returned. I am still very breathless on exertion, but I climbed my first hill yesterday so a definite improvement. I have had both vaccinations now as a frontline worker and am looking forward to getting back to tennis soon.

I will recommence weekly PCR tests and twice weekly lateral flow testing in mid-April as you must wait 90 days after testing positive, but I just wanted to let everyone know there is light at the end of the tunnel. Stay safe everyone - get vaccinated and keep washing your hands!

Joan's sunflower cataracts made the papers back in 1963, when she was first diagnosed with Wilson's disease at the age of 13. She has subsequently had a liver transplant and recently moved in with her daughter, son-in-law and two grandchildren into their brand new house. She writes,

Joan sitting in front of the plans of the new house: Joan enjoys her own self-contained suite...

I have hardly been out of the house in the last twelve months. One day blends into the next. The most exciting thing I have done was going up to Cambridge, a few weeks ago, to the Park and Ride Car Park to put my arm out of the car window for a phlebotomist to take blood from me! I could have gone to Southend Hospital, but that would have meant going inside into the clinic at a time (mid-January) when Southend was verging on a crisis situation having the highest number of Covid cases in the region. Besides, going to Cambridge gave me an excuse to have a trip away from my familiar surroundings so the journey was welcome. I haven't driven for over a year, so my daughter offered to take me. All the results were good!

At home, I am very interested in politics so I have found all the political shenanigans very interesting. Reading all the on-line comment boards has kept me entertained. But having to shield for so long has made me quite unsteady on my feet . Now that the weather has improved and I've had my first vaccine, I try and take short walks every day, with support. The idea is to get my strength and confidence back, ready to return to some level of normality. I get my second dose of vaccine in another couple of weeks, but won't be mixing outside of the home until there is a clearer picture of the effectiveness of the vaccine on those of us who are immunosuppressed.

When all this is over I hope the powers that be recognise how shielding and isolation has affected people's physical wellbeing as well as their mental health.

Anne-Marie is a retired Modern Languages lecturer and former WDSG-UK committee member. Diagnosed in 1977, her story can be found in the 2011 newsletter. She writes:

Our Pre-Lockdown Trip to California: How very different the last year has been. Before then, my husband and I had been fortunate to be able to spend our time travelling in the UK and abroad. It's just over a year since our last big adventure and I thought I would do some armchair travelling looking back on some of the many highlights.

Mid-February last year we left the UK to begin an exciting month-long road trip in mid California. Along the way we discovered how diverse this part of the US is, how interesting its history and culture and how amazing its landscapes and scenery.

Air Force One

We flew to LA and spent a week in Pasadena with my brother who was spending a year at the university of Cal Tech as a visiting history professor. One of the highlights of that week was meeting up with a friend from my university days who I hadn't seen for over 30 years. We also discovered the delights of the Huntingdon Museum gardens, the Norton Simon art gallery and the fascinating Gamble House, a superb example of American arts and crafts style of architecture. LA certainly isn't just about Hollywood and Disneyland.


Baby Sea Otter

Leaving LA we headed north taking the famous coastal road. On the way we stopped off at the Ronald Reagan Library where we could board Air Force One used in Reagan's day, now housed there. We stayed at picturesque Santa Barbara (near where the Sussexes now live) with its Spanish colonial heritage, then on to Pismo beach where I remember the delicious spare ribs in barbecue sauce. Further up the coast we saw the sea otter nursery where the sweet little babies floated around asleep on their backs. The most outlandish place we visited has to be Hearst Castle, which looks like a Spanish cathedral on the hillside, conceived and built in the 1920s and 30s by the publishing tycoon William Randolph Hearst and where the guest list comprised most of the Hollywood stars of the day.


Hearst Castle

Leaving the coast at Monterey we made for Yosemite National Park to admire its magnificent scenery: soaring granite cliffs , waterfalls and giant sequoia groves. After a few days exploring, we went north to Lake Tahoe to come back down Route 395 through breathtaking mountains and wild west country and into Death Valley, where the temperature in summer is intolerable, but very pleasant in wintertime. There we came across an oasis of palm trees surrounding what looked like Hotel California. We took a chance and decided to have lunch and fortunately were able to leave without trouble. Travelling on, we picked up the western end of Route 66 through the Moave Desert where you find plenty of weird-looking Joshua trees, a kind of Yucca.


Yosemite National Park

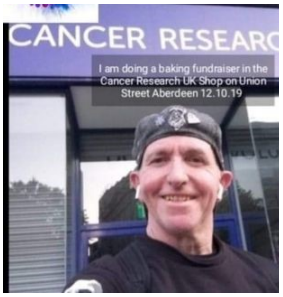
Finally, we drove high into the San Bernardino mountains coming full circle back towards LA where we were met with freezing temperatures and snow. It was here that news of Covid spreading through Europe was making President Trump start closing the country to planes from home. We were due to fly back three days later but decided to curtail the trip as we didn't want to find ourselves stuck, and BA proved impossible to contact. We managed to catch an earlier flight and were home a week before lockdown.

Sadly, Covid has put paid to more travelling, so instead, we took advantage of being at home to move house! Having talked about moving to a modern property, but still in the countryside, we actually did so this February. We have bought a house an hour's drive north from where we used to live, in north Cumbria close to the Eden Valley, not far from Carlisle and the Scottish border. In fact it feels very much as if we have gone away on another holiday!

Eddie's Story

by Eddie Watt

I was born on Friday, 13th—was that an omen? The youngest of three children, our family remained in Scotland until shortly after I started primary school in 1975, when my father, a printer with the *Sunday Post,* was offered a job working for a similar national newspaper in Rhodesia. And so started our new life so different from the old, with hot weather, a large house with a swimming pool and animals such as elephants, lions and rhinos practically on our doorstep! My only memory of attending a hospital as a young boy was being rushed to A&E after managing to lodge a bead up one of my nostrils!! And my only recollection of copper being mentioned was in relation to the many copper mines found in that part of Africa!



Eddie—2019

When I was about 10, my parents split up and my father returned to Scotland. Soon afterwards, I was sent 350 miles and some 7 hours away from home to a colonial style, private boarding school where both physical and mental abuse from the staff and senior boys were an every day occurrence. It was tough! Academically, I would say I was an average student, preferring the arts to the sciences and gaining 7 O'levels including a top grade in English. We spent many holidays in Scotland, but I missed my dad and at the age of 17 decided to return to live with him back in Glasgow leaving my mother, step-father and two older sisters in what had then become Zimbabwe.

Once back in Scotland, I enrolled on a 2 year Hotel Management course at the Glasgow College of Food and Technology. My career got off to a good start and by the age of 23 I was married. Two years later my son, Adam, came along. At around this time I found a lump in my neck which was drained, but it reappeared and was later diagnosed as thyroid cancer, for which I was and continue to be effectively treated.

Meanwhile, gaining experience in all aspects of hotel management meant moving from job to job, including 2 x four year stays in Bermuda. We were living very comfortably but I was away working horrendous hours at top end hotels like Turnberry and Gleneagles, and the marriage started to falter resulting in divorce ten years later. I was devastated and turned to drink. Looking back, I would say that I operated as a *functioning alcoholic* for around 14 years. However, my last job was in Aberdeen and in the end I could no longer function. I was admitted to Aberdeen Royal Infirmary in 2017 and was told that I had cirrhosis of the liver and if I didn't mend my ways I would only have 6 months to live. I was placed in a rehab unit for 3 months during which time I stopped drinking altogether.

Since the divorce I had continued to visit Adam in Ayrshire, when I could. On one such occasion in March 2018 I was out shopping when I was subjected to an unprovoked attack by a hooded youth who struck me in the face with a glass bottle. My injuries necessitated a trip to the local A&E department. Here, an ophthalmologist examined the damage to my eyes and said he could see copper rings called Kayser-Fleischer rings in them. I had no idea what he was talking about and I left, not giving it another thought.

My road to self-destruction gathered apace. Soon afterwards, I was arrested for leaving a sushi bar without paying. I was drunk and put into a police holding cell. While there, I was charged with an alcohol induced non-violent crime against a Police Officer which resulted in an 8 month custodial sentence—and a lifetime of shame! It was the first time I had ever been in trouble with the Police and the experience sobered me up rather quickly!

> ***At the time, it seemed like the worst day of my life...***

Being on thyroxin as a result of my thyroid cancer, I was having monthly blood checks while in prison. One day I was summoned to the health centre to be seen by a doctor. She said, "I'm not going to beat about the bush, but you have a rare genetic disease called Wilson's disease and it is too late to treat." It was the 4 November 2018. It came without any warning and I was devastated. At the time it seemed like the worst day of my life but it has proved to be my salvation. I was released two months later after starting on penicillamine and I resolved never to drink again, and I haven't. My new found freedom from alcohol has allowed me to raise funds for *Cancer Research UK* and up to the beginning of the pandemic I had collected over £1,250. I am now living in supportive accommodation as I have also been diagnosed with early onset dementia. I can't wait for restrictions to be lifted later this month so that I can return to the things that I enjoy most like church fellowship, swimming, dancing and Zumba classes!

Looking back at my lifetime's mistakes, and some triumphs, I am comforted in the knowledge that I have become a better person since the days of alcohol addiction, self-destruction and hurting others. Cancer, alcohol addiction, a diagnosis of Wilson's disease at the age of 48 and early onset dementia will not deter me and I will continue to do all I can to improve myself physically, mentally, spiritually and emotionally. I hope you won't judge me for my past life. I firmly believe that there is good in all of us. I am grateful to the support I have received from WDSG-UK and I wish all other patients well.

A Date for your Diary 2021-22

Date	Time	Event
Sunday, 18 July 2021	1100	WDSG-UK 11th AGM – Zoom Virtual Meeting — Details to follow

WILSON'S DISEASE MULTIDISCIPLINARY CLINICS

Adult Clinics

The Birmingham WD Clinic

Dr Andrew Holt (Consultant Hepatologist) and **Dr David Nicholl** (Consultant Neurologist) hold a one-stop Wilson's disease clinic at **University Hospital Birmingham** on a Friday morning four times a year. This clinic offers patients the opportunity to have their management reviewed by a hepatologist and a neurologist at the same time and is intended to supplement otherwise established care. With the pandemic, we are doing more remote (video/phone) consults as face to face consults are still limited. Referrals must come from the clinician looking after the patient and should be addressed to **Cheryl.Scandrett@uhb.nhs.uk.** She is the WD Clinic Coordinator at Queen Elizabeth Hospital, Mindelsohn Way, Edgbaston, Birmingham, B15 2WB

The Cambridge WD Clinic

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

The Leeds WD Clinic

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

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

A dedicated Wilson's disease clinic has been held at University College Hospitals for the past 30 years. It was set up in 1987 by Dr John Walshe after his retirement from Addenbrooke's Hospital, Cambridge and at the invitation of Dr Gerald Stern. **Dr Godfrey Gillett** started to attend in the mid-1990s and has continued the clinic under the kind auspices of Dr Robin Lachmann at the National Hospital for Neurology and Neurosurgery, Queen Square. The clinic has 60 patients and is held on the third Friday of the month with the close involvement of **Professor Tom Warner, Dr Sam Shribman** and **Mrs Maggie Burrows**. Referrals from across the UK are welcome, addressed to Dr GT Gillett, Laboratory Medicine, Northern General Hospital, Sheffield Teaching Hospitals NHSFT, Herries Rd, Sheffield, S5 7AU or to Dr GT Gillett, Charles Dent Metabolic Unit, Internal Mailbox 92, NHNN, Queen Square, London WC1N 3BG.

The Sheffield WD Clinic

The Sheffield clinic is jointly run by **Prof Oliver Bandmann** (Consultant Neurologist), **Dr Godfrey Gillett** (Consultant in Clinical Biochemistry, Inherited Metabolic Disease, and Adviser to WDSG-UK) and **Dr Barbara Hoeroldt** (Consultant Hepatologist). Clinics take place every six months at the **Royal Hallamshire Hospital, Sheffield** on a Tuesday morning. 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, South Yorkshire S10 2JF. Either GPs or hospital specialists may refer to this clinic.

Children's Clinic

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.

Wilson's Disease Support Group – UK

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Debbie Buckles:	Committee Member Email: **debbie@wilsonsdisease.org.uk**
Dr John Walshe	Honorary President, World Authority on Wilson's disease
Dr Godfrey Gillett	Group Adviser, Honorary Member
Caroline Simms	Group Co-Founder
Linda Hart:	Group Co-Founder
Rupert Purchase, DPhil	Group Adviser on trientine
Webmaster:	**Blackcat 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 or patients & families' correspondence lists are required, please contact Valerie.

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