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

NEWSLETTER

#wdsguk APRIL 2019

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

Climb Children Living with Inherited Metabolic Diseases

A warm welcome to the newsletter, particularly to our new members who this year come from as far afield as Aberdeen and the Shetland Islands. I hope that you will all find something of interest within. May I begin by thanking our patient contributors, **David, Helen, Dorothy, Alpana**, **Caroline** and **Ashok,** who have either shared their personal accounts of Wilson's disease or have contributed to the members' section at the back. David's story demonstrates that you are never too old for a diagnosis, and Dorothy's underlines the importance of being compliant with your treatment. These stories tend to be the most popular component of our newsletter, but might well be rivalled this year by **Katy's** appetising article on the manufacture of chocolate! It appears that the copper content of **white** and **milk chocolate** is no different from that say of white and brown rice! And for other comparisons, patients will find enclosed a revised *WDSG-UK Food Chart* compiled from the Foods Standards Agency (2002) **McCance & Widdowson's** *The Composition of Foods: 6th Summary Edition,* Cambridge: Royal Society of Chemistry.

Photo: Courtesy Anusha Joseph

We are, of course, grateful as ever to all the doctors who support us. They include **Dr Bill Griffiths** who has written a medical perspective on David's story, and **Dr Chris Harrington** who explains the complex laboratory procedure for quantifying copper. We thank **Dr Godfrey Gillett** for working closely with the Group and for always being available to advise, whenever asked to do so. And for those of you taking trientine, I am sure you will wish to thank **Dr James Dooley** who has worked tirelessly on your behalf over the past two years leading clinical discussions with NHS England to ensure that, despite its cost, trientine continues to be prescribed to patients who need it.

Jerry explains more about this and all the other work that is ongoing on behalf of the Group in his report overleaf. As chair, he is deeply committed to improving all aspects of healthcare for patients, and devotes much of his time to attending meetings, writing reports and keeping up to date with medical advancements that could be of benefit to us all. I am sure that you will join me in thanking him, too, for his time, dedication and unwavering support for the Group.

As usual we are indebted to our fundraisers who this year include **Rushden & Diamonds, Andy & Ann-Marie Collcott, Sam Panchal, Jamie Finlayson, Belinda Diggles** and **Ishbel Salt**, whose various exploits are described within, and to our generous donor, **Giuseppe Cardone,** who has again managed to raise nearly **£2,000** through his *workplace charity giving scheme* at *Google.* On a sadder note, we were sorry to hear of the deaths this year of **Keith Pereira, Jean Turley, Stefanos Kaloumenos** and **Heather Dixon.** Heather was the widow of **Hal Dixon,** the Cambridge biochemist who suggested the use of trientine as a treatment for WD. They were all closely affiliated to the Group and will be greatly missed.

Please don't forget to keep up to date with our Facebook page, through which we are able to deal quickly with your queries, such as difficulty accessing your meds. We now have 850 members worldwide and your shared experiences and advice to others are always well received.

Our annual meeting will take place on **Sunday, 21 July** (full details of which are enclosed), to which you are all warmly invited. **Sam Shribman** will be there giving us an update on his recently launched **CROWD** Study (which I hope you are all involved in!), as will **Dr Walshe**, who by then will be well on his way to reaching his **100th** birthday. We realise that it is a long way for many of you to travel and we do appreciate your efforts, but it is an excellent opportunity to support one another and show your appreciation to Dr Walshe for continuing to support all of us.

Now, all I have to do is remind you to please renew your subscriptions **promptly.** I look forward to seeing as many of you as possible in the summer, but meanwhile have a very happy Easter and may all your eggs be chocolate ones! Enjoy the Dingbats...

Valerie

IN THIS ISSUE ...

Chairman's Report for 2018-19

This year has been a dynamic and significant one for the Support Group. We have focussed our efforts on bringing the patient's perspective and voice into organisations with which we are involved, in the hope that this will eventually lead to quicker diagnosis, new methods of treatment and better long-term healthcare for our patients.

We continue to work closely with **Genetic Alliance** and **Rare Disease UK**, expressing our views on the type of services we need, especially healthcare services and improving diagnostic methods. We also retain our existing links with the **NHSBT Organ Donation &Transplantation Directorate,** the **British Liver Trust**, the **Liver Patient Group, the Liver Patient's Transplant Consortium** and the **National Institute for Health Research** (NIHR).

We could not be involved in the activities described below and elsewhere in this newsletter without the help and guidance of our committed medical advisers, **Drs Godfrey Gillett** and **James Dooley,** and more recently **Dr Bill Griffiths** (who leads the **BASL WD SIG,** about which he writes on *p.16*). I would like to thank them for all their support this year and I would also like to thank our loyal members for **their** continued support.

Donations and Fundraising

WDSG-UK does not receive sponsorship from commercial organisations and consequently we rely exclusively on our members and their families and friends for our income. We should therefore like to thank all of you who made generous donations with your subscriptions this year. In addition, we should like to thank **Giuseppe Cardone** who, together with a group of fellow Google employees, donated **£1,800** to us via Google.org support for non-profits. We also very much appreciate the contribution that **Linda Pereira, Emma Coombes** and **Alpana Kharkar,** and their families, have made to us by holding collections at the funerals of their loved ones. Together they raised over **£2,000.**

And as for fundraising, we are grateful to **AFC Rushden & Diamonds, Andy** and **Ann-Marie Collcott, Samantha Panchal, Jamie Finlayson, Belinda Diggles** and **Mrs Ishbel Salt,** who have brought in an additional combined income of **£3,000.** 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 **£230**. Any donations to us, of course, can be gift aided. Forms are available from **Valerie** direct or off our website www.wilsonsdisease.org.uk.

WDSG-UK Activities

Over the past year, as patient representatives, we have prioritised our limited resources on working with **NHS England**, with the **British Association for the Study of the Liver** (BASL) and with **Public Health England** (PHE) and I am happy to report back as follows:

NHS England—Trientine Policy for Wilson Disease

This year has been dominated by **NHS England** completing and publishing the Clinical Commissioning Policy specifying guidelines for the funding of trientine as a treatment for Wilson disease in England (Trientine for Wilson Disease (all ages): NHS England Reference:170094P). These guidelines will be effective from **1st April 2019**. It is a very important policy and safeguards the continued use of trientine to existing patients and outlines the criteria for prescribing it to new patients. The Review was a long and exhaustive process managed by Joan Ward of NHS England and carried out with great rigour. Valerie and I contributed to it on your behalf by pushing for trientine's continued use, as did medical specialists, including Drs. Dooley, Gillett, **Bandmann** and Griffiths, to whom we are indebted. Of particular mention is James Dooley who spent many hours, as lead, gathering all the necessary information and guiding the clinical deliberations and discussions through a labyrinth of data and views to arrive at the final policy document. The Policy and the supporting documentation can be found at www.england.nhs.uk/publication/trientine-for wilson disease-all ages/.

BASL Special Interest Group: Wilson's Disease

Valerie, **Mary** and I are members of the BASL WD SIG Group, representing your interests in a group of clinicians and scientists who, I believe, are passionate about improving the care and treatment of patients with Wilson's disease. They have the reach and authority to engage the NHS in bringing about change. The SIG's aims are wide-ranging and ambitious and further information about it can be found on *p.16* of this newsletter. Of particular interest is the initiative being proposed for the development of specialist services to treat Wilson's disease and we have submitted our documented specifications as to the type of service we would like to see, which would ensure consistent quality across the UK.

Public Health England: Wilson's Disease Pilot

PHE took over the responsibility for the registration of rare diseases NCARDRS (National Congenital Anomaly and Rare Disease Registration Service) in 2015. The work on collecting data on the Wilson's disease population in England is ongoing and an update by **Mary Bythell**, head of NCARDRS, can be found on *p.19*. Just to remind you, Wilson's disease is being piloted by PHE as the first rare disease population in the UK on whom data is being collected. We eagerly await the results.

Genetic Alliance and Rare Disease UK

I continue to be actively engaged with organisations such as Rare Disease UK and the Genetic Alliance Patient Empowerment Group which monitors the progress of the implementation of the UK Rare Disease Strategy. The **Department of Health** recently published a progress report and specifically mentioned the **PHE Rare disease Wilson's disease pilot** to illustrate the progress being made on one of the 'commitments' outlined in the Strategy. It is not often that Wilson's disease gets any mention in official documents, so we welcome our involvement in this type of project as it raises our profile in the wider medical community and also in the public domain.

I also attended a workshop on the advantages of neonatal testing for genetic diseases, which would be an area of great interest to the group in making an early diagnosis. Existing and new methods of testing such as genetic tools were discussed and the process for approving new tests, which is very rigorous in the UK, was explained. This is a complex subject and in the case of Wilson's disease further research is required to establish whether testing is viable, especially using new genetic methods.

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

Valerie attended two meetings in London over the past year with NHSBT, the LPG and the LPTC. The LPTC is a partnership of all liver patient support groups and charities in the UK working together on transplant issues important to patients and their families. In July, an update was given on the new Liver Offering Scheme implemented in April 2018 whereby patients anywhere in the country requiring a new liver go on a national rather than a regional list, thus ensuring that livers are given to patients with the greatest need.

Also discussed at this meeting was the introduction of a 'deemed consent/opt out system' for organ donation in England in 2020, and a new scheme of *hypothermic* and *normothermic* machine perfusion for donor livers. The difference between the two types of perfusion are in the temperature and degree of oxygenation delivered to the livers. Perfusion of donor livers is undertaken at the time of organ procurement and keeps the liver viable for transplantation longer. So far the trials have returned excellent results with more livers being deemed fit for transplantation than would previously have been. For further information visit https://www.odt.nhs.uk

Supporting Clinical Research

We were invited to participate in research being run by **UCL Department of Applied Health Research** reviewing **Co**ordi**n**ated **C**are **o**f **R**are **D**iseases **(CONCORD)**. The aim of the study is to investigate how care of people with rare diseases is coordinated in the UK and how patients and their families, as well as the healthcare professionals who treat them, would like them to be coordinated in the future. WDSG-UK was selected to attend a workshop to raise the issues our members faced. Our experience was very similar to that of other groups which illustrated that lack of coordination of services was a common problem in rare disease management. The study will collect data from published resources and elsewhere and will also review different economic models of service delivery. The study is due for completion at the end of 2020, when the findings will be shared with participants involved in the study.

The CROWD Study (Cohort Research on WD)

Prof. Tom Warner and Dr Sam Shribman from the Dept. of Neurology at UCL have now launched this Study to see if they can discover any clear differences between Wilson's patients who develop neurological or psychiatric problems and those who develop hepatic problems (please see update on *p.18*). We were pleased to make use of our WD Patient Register—UK for the first time and wrote to patients enclosing invitations from UCL for them to take part in the Study. 60 invitations were sent out in total and we understand there has been a good response. Thank you to everybody who has taken part and anybody still wishing to join our Patient Register can download a form off our website or alternatively approach Valerie for further information.

WDSG-UK Meetings

During **2018-19,** The WDSG-UK management committee met twice, in October and January. Our annual Support Group Meeting and 8th AGM was held in Cambridge in July 2018 (please see report overleaf).

WDSG-UK Committee & Annual Meeting and 9th AGM

The 2019 Support Group Annual Meeting will be held on **Sunday, 21 July 2019** at our normal venue in Cambridge at the clubhouse of Cambridge Rugby Union Football Club. The 9th WDSG-UK AGM will take place as a formality after lunch with the election of officers and members of the WDSG-UK Management Committee for 2019-20. All members of the current committee have submitted their names for re-election for the coming year. We welcome new committee members with new ideas and expertise to take the Group forward and are looking, in particular, for help in modernising our website.

Finally, I would like to take this opportunity to thank the current members of the committee: Valerie Wheater, Mary Fortune, **Caroline Simms** and **Liz Wood** who have served with great commitment over the last year. They have given their time and brought their skills and experience of living with Wilson's disease to guide the management of the Group. Valerie, continues to play a vital role in the every day functioning of the Group and her commitment to patients and knowledge of the wider Wilson's community ensures that WDSG-UK continues to be the leading charitable group in the UK dedicated to supporting patients with Wilson's disease, and their families.

Jerry Tucker
March 2019

Wilson's Disease Support Group Meeting & 8th AGM

Cambridge Rugby Union Football Club, Sunday 15 July 2018

With no respite from the 2018 summer heatwave, the **WDSG-UK** Annual Meeting and 8th AGM again took place at the Cambridge Rugby Union Football Club on Sunday 15 July. Fortunately for attendees the conditions inside the Turnstone Suite were altogether cooler, with the air conditioning system coming into its own and offering great relief from the constant heat of the previous few weeks.

As always, we were delighted to welcome our regular members from across the UK, together with several new members, who included **Lenka** and Jamie **Jonasova** from Edinburgh, **Rosie Faruok** and her niece from Bolton, **Joy Allen** and her husband from the West of England, **Katie Chiswell** and her husband from Leicester, **John** and **Sharon Ryan** from Liverpool, **Dorothy Patterson** and her daughters from Norfolk, **Rafal Szczygielski** and his parents from Surrey and **Gill Ford** and her husband from Hertfordshire.

Representing the medical profession and offering patients advice and support throughout the day, we were pleased to welcome **Dr John Walshe,** our ninety-eight year old President and world authority on Wilson's disease and his successor, **Dr Godfrey Gillett,** the medical adviser to the Group and a Wilson's disease specialist with clinics in Sheffield and London. In addition, we were joined for the first time by **Dr Sam Shribman**, a neurologist from University College London (UCL), who joins Dr Gillett at his monthly WD clinics at Queen Square, London, and who was to be our first speaker of the day.

Jerry Tucker, Chair of WDSG-UK, formally opened the meeting before delivering his annual report. He began by talking about the implementation of the *2020 Rare Disease Strategy*, which recommends earlier diagnosis and intervention in rare diseases, improving and standardising patient care, developing specialist services in multi-disciplinary clinics across the UK, creating Hospital Centres of Excellence and promoting research. He then spoke about *WDSG-UK's* involvement in the recently formed **BASL WD SIG** (British Association for the Study of the Liver, Wilson's Disease Special Interest Group). This Group is made up of Wilson's disease specialists, scientific staff and ourselves and its aim is to improve patient care across the UK (see *p16*)

Jerry reminded us that WDSG-UK had been representing patients' interest on a panel headed by Public Health England (PHE) in which the future prescribing and funding of trientine in England was being discussed. The situation had arisen because of Univar's massive price increase of the drug. Finally, he mentioned that Phase 3 of the drug trial of WTX101 (bis-choline tetrathiomolybdate) was now under way and that gene therapy, which could help as a treatment for some Wilson's patients in the future, was emerging and attracting much interest.

The blistering heat outside... ***R Faruok***

...cool, calm and collected inside ***B Diggles***

Sam Shribman talking about the CROWD Study ***B Diggles***

Jerry then handed over to Sam Shribman, who introduced us to the exciting new research project he and his team are planning into Wilson's disease. Called **The CROWD Study*: Cohort Research On Wilson's disease,*** the aim of the research is to discover why some Wilson's patients develop neurological or psychiatric problems and others develop liver problems. He encouraged patients to join the WD Patient Register—UK if they hadn't done so already as once the Study was launched in the New Year, WDSG-UK would be sending out letters to those on the Register inviting them to take part.

An *in house* buffet lunch was provided between 1300 and 1400 hours, giving members the opportunity to catch up with one another and talk to the doctors informally, while **Sue** and **Chris Boysons** sold the last of the raffle tickets, **Belinda Diggles** ran a stall selling home made jams and chutneys and **Samantha Panchal** found yet more customers to buy her silicon wristbands (details of which can be seen on *p7* overleaf).

Afterwards, the **8th AGM** was very quickly held. Current committee members, **Jerry Tucker, Valerie Wheater, Mary Fortune** and **Caroline Simms**, were unanimously re-elected to the Management Committee for **2018-19** and **Liz Wood, a** patient and long time member of the Group from Southend in Essex, very kindly agreed to join the committee for the forthcoming year.

Our second speaker, **Mary Bythell,** from PHE (Public Health England) then spoke to us about a new project she was heading to establish population numbers of patients with rare diseases in England. **Prof. Graeme Alexander**, a hepatologist now working at UCL, whose idea it was to set up the BASL WD SIG during his tenure as president, had suggested to her that she pilot her new scheme on population numbers of Wilson's patients. Work was now under way and Mary hoped to have some exciting results to share with us next year (for an update see *p19*).

Patients were then given the opportunity to ask questions of the doctors and also of the committee before the raffle, which raised **£116**, was slickly drawn by Chris and Belinda. Our thanks go to Belinda's husband, **Barry,** for being our official photographer for the day. He then collected us together to take Group photographs before the meeting was officially closed and we all set off into the waning heat of the afternoon to make our way home.

Mary Bythell from PHE ***B Diggles***

Katie and Rosie ***R Faruok***

The Group Photograph ***Photo: Courtesy Barry Diggles***

Fundraising 2018-19

Valerie

AFC Rushden & Diamonds Football Club —Youth Section

We are very grateful to **AFC Rushden & Diamonds Youth Section** for nominating WDSG-UK as their chosen charity for the **2016-17** and **2017-18** seasons. **Sam Fitzgerald**, a Wilson's disease patient, was an outstanding junior player at the Club before Wilson's disease took its toll on him in 2015. The Club wanted to show its support for Sam and has done so over the past two seasons through various initiatives and events led by **Mark Cullen,** Chair of the Youth Section of the Club. The events culminated on **Sunday, 29 April** last year with a Ladies' Charity football match at their new ground in Hayden Road, Rushden. A bitterly cold wind from the East greeted the players on the pitch, but this did not dampen their spirits nor weaken their resolve! In the changing rooms afterwards, **Tracy Stephen,** head of fundraising, kindly presented **WDSG-UK** with a cheque for **£1,000** which Valerie happily accepted on its behalf. We thank everybody involved - the players, supporters, organisers and sponsors—for their overwhelming generosity and we wish all sections of the Club great success in the future.

Tracy Stephen presenting Valerie with a cheque for £1,000.

Andy and Ann-Marie's MK Colour Obstacle Rush

Andy's sister, **Emma Collcott,** is well known to many of us, particularly those attending our annual meetings in the summer. She has spent a lot of time in hospital over the past year and Andy and his wife, **Ann-Marie**, wanted to show their support for her by taking part in a charity event and raising funds for the Group. This they did on **Saturday, 2 June** last year when they entered Milton Keynes' ***4th Colour Obstacle Rush*** at Willen Lake on the north-eastern fringes of the town.

Cheered on by their parents, the course covered a distance of 5km along which there were 8 colour stations (spewing out neon coloured powder), 20 different inflatable objects to crawl over, through and under and 20 music zones spurring the participants on and adding to the carnival atmosphere of the occasion. Through the WDSG-UK Charity page on the *Virgin Money Giving* site, Andy and Ann-Marie advertised the event and raised **£450** for the Group. We would like to thank their work colleagues, family, friends and supporters for their generous support.

A colourful Andy and Ann-Marie after the Obstacle Rush!

Valerie's Demon Dingbats

They're everybody's of **T**, I know, but last year's Dingbats com- petition raised a further **£300** for the Group. Based on the names of London Underground Stations, some of the answers proved very taxing indeed! This was the third competition of its kind included free in members' newsletters, but otherwise sold separately to friends and the unsuspecting public around where I live! Thanks to everybody who took part, and congratulations to **Ken Dunkerley** from Oldham who won the **£25** prize.

This year's Dingbats competition is based on the names of countries around the world—some easier than others! Entrants have until *17 June* to submit their answers. As this is primarily a money spinning exercise, if you prefer not to take part, please see if you can sell your copy to a friend or family member for **£2** and then credit the Group's account accordingly. Every penny counts!

2018 London Underground Stations

Samantha's Sale of WD Wristbands, and *Tropical* Raffle

Samantha Ryan Panchal continues to sell her blue silicon wristbands to help raise awareness of Wilson's disease and to publicise the Group. She has raised a further **£300** this year bringing the total amount generated to **£750.** These wristbands are still available, should you wish to buy one. They cost **£2** each **plus** postage and can be obtained via WDSG-UK or its Fb page, or direct from Sam with payment to her via Paypal (as gift friends/ family) (https://www.paypal.com/uk/home). Sam's brother, **Ben**, was diagnosed with neuro Wilson's in August 2016 at the age of 28. His has been a harrowing story, which his father, John, hopes to share with us next year.

Sporting the Wilson's disease wristbands and looking Absolutely Fabulous!

Having recently become an ambassador for *Tropic* products, an all natural vegan and toxin free range of skincare and makeup, Sam who is never short of ideas decided to run a couple of raffles in aid of WDSG-UK. The prize for each was a *Tropic* beauty treatment and deluxe skin care pack worth £98. The profits from the raffles brought in a further **£130** for the Group and we thank Sam and all her friends for supporting us in this way. As an aside, for those who enjoy watching Alan Sugar's *The Apprentice* on television, the founder of *Tropic*, Susie Ma, was a contestant in the 2011 series. For further details, please visit Sam's website: www.tropicskincare.com/sampanchal.

Jamie's *White Collar Fighter* Competition

Towards the end of last year, **Jamie Finlayson** began training with *White Collar Fighter* in preparation for a sponsored boxing event which was being held in Salford on **24 November.** The idea of the event is to take beginners who have never boxed before and over an eight week period and under the guidance of world-class coaches prepare them for an experience of a lifetime! Pushed to his limits through the intense training programme, Jamie decided to use the event to raise awareness of Wilson's disease, and funds for the Group**.** His mother**, Tania,** was diagnosed with neurological Wilson's disease when she was only 13 years old and struggled with her symptoms all her life. Living in Blackpool where she brought up Jamie and his little sister, she sadly died in 2016 at the age of 35. Although his mother had never heard of the Support Group, Jamie was keen to help others with the disease and found our details over the internet. He won his fight on the night and raised **£535** in the process. Many thanks to him and all his followers who sponsored him and who encouraged him along the way and supported him on the night!

Jamie preparing for the fight

Belinda's *Home Made Produce* Stalls

In addition to the jam and chutney stall that **Belinda Diggles** ran at our annual meeting in the summer, on **2nd December** she also took a stall at the small craft fair held by the Lancashire Power Boat Racing & Waterskiing Club, which she and *Jess* visit regularly on walks from their house. As she was raising funds for charity, she wasn't charged for her table and the sale of her Christmas cakes, mince pies, jams and chutneys, not to mention the gingerbread men iced by her husband, **Barry,** brought in an overall total of **£240** for the Group! Thank you to Belinda and everybody who supports her for continuing to fundraise for us in this way.

Yum, yum!

Ishbel's *Significant* Birthday

And finally, we would like to thank **Mrs Ishbel Salt** from Cheadle in Staffordshire for choosing to ask her family to make donations to WDSG-UK in lieu of presents on her recent *significant* birthday. Ishbel's son, **Nick,** was a member of the Group before he died in September 2011. Her very generous gesture raised **£300**.

An Unusual Story

by David Redman

Hello! My name is **Dave** and I live just outside Colchester in Essex. I am married with four children and six grandchildren. My story of Wilson's disease is a little different from most. I am now aged sixty-two, but I was only diagnosed with the disease less than two years ago.

At Durdle Door in Dorset last September

I have always been athletic. I am 6'4" tall and spent fifteen years playing Rugby for my local Club. In addition, I was a keen runner and always took advantage of this pursuit whenever I went on holiday. In **March 2017**, I had taken my wife to Grasmere in the Lake District for her birthday celebration. We know the area well and I love running over the surrounding fells. However, on this occasion I noticed that I lacked my usual energy. I pushed myself, but it was difficult. When I returned home I went to see my GP to express my concerns and he ran a few tests, but nothing conclusive was found.

A few weeks later I noticed my legs were getting heavier and I had even less energy. Then one weekend in **May 2017** I was decorating the house and my legs had become so swollen I couldn't bend down. My wife had commented that she thought my face was changing colour slightly. I took myself down to the Out of Hours surgery at Colchester General Hospital and they took one look at me and sent me straight to A & E. From there I was admitted, but my condition only deteriorated. By **Thursday, 15 May,** I was confused and very ill. I have little recollection of events after that until I woke up in Addenbrooke's Hospital seven days later.

I have to rely on my medical notes and also my family's memories to fill in the gaps. Colchester had transferred me to Addenbrooke's when they realised I was in end-stage liver failure. Addenbrooke's believed that I was too ill to receive a transplant. However, I am eternally grateful to **Prof Watson**, **Dr Jacobus Prellar** and **Dr Jo Leithead** at Addenbrooke's who pushed for me to go to the top of the transplant waiting list, because within twenty-four hours they had found me a suitable match, which they gave me straight away.

I have a wonderful family who despite living far away were always at my bedside. When I came out of a coma after my transplant, I was told that they had found that I had **Wilson's disease**. I had never heard of it. However, the doctors concentrated more on the transplant side of things and making sure that my liver wasn't rejected. I was in hospital for six weeks fighting a fungal infection that I had contracted there, before finally being allowed to go home. I could hardly walk as I had been off my feet for so long.

However, being so active beforehand I was determined to get myself fit again. Slowly my strength returned and in **May 2018**, just one year later, I competed in a half marathon raising funds for the *Addenbrooke's Charitable Trust*. Also around that time I decided to look more into Wilson's disease. My sons, daughters and grandchildren have all been checked, and the next step for me was to see what support there was for patients with the disease. I found out about the UK Support Group off the internet and made contact with Valerie straight away. I have recently met her for the first time and she asked if I would be happy to write an article about my experiences.

> *"I have little recollection of events after that…"*

I am only too happy to do what I can to help others with Wilson's and also to publicise my own story to a wider audience. I understand it is unusual for a patient to reach the age of sixty without having had previous symptoms of the disease. I suppose looking back that I had noticed my handwriting had started to deteriorate, but other than feeling tired I can't remember any other unusual symptoms for me.

I am very happy also to put myself forward to take part in the CROWD Study being carried out by the Institute of Neurology at University College, London. So far I have completed the online questionnaire and sent off a saliva sample for Sam to process and have now been invited to take part in the final stage of the research. For those of you who are undecided as to whether or not to take part, may I encourage you to do so. In my case, it is a small way of giving something back for all the help that has been given to me. After all, I feel so lucky to be alive.

An Unusual Story—the hepatology perspective

Wilson's disease (WD) can occasionally present for the first time as a sudden onset, severe form of liver disease which is termed *acute liver failure*. David's case shows that you can never be 'too old' to present with WD and at 60 years of age we think he is the oldest patient on record to present with fulminant hepatic Wilson's and survive.

It is remarkable that a genetic disorder can show no signs whatsoever until this stage in life, and then manifest in such a dramatic fashion. Of note, Wilson's disease patients who develop acute liver failure by and large have already developed cirrhosis, i.e. their liver has in fact been scarred over a long time until eventually the copper builds up to such an extent that the liver can no longer cope. It is not clear why some patients present with sudden onset disease and others more gradually, although David is now contributing to the CROWD study, run by the neurologists **Dr Sam Shribman** and **Professor Tom Warner** at Queen Square, London, which aims to find out whether genetics can explain these differences.

There are certain features that are seen in acute liver failure. Typically, patients present with jaundice and prolonged clotting (*coagulation)* time. Confusion may also be present because of the failure of the liver to clear toxins, particularly ammonia, and this confusion is termed *hepatic encephalopathy.* Patients may also rapidly develop *ascites* which is fluid in the abdomen occurring when the liver fails. To make a timely diagnosis of acute liver failure due to WD in a patient with no prior history nor family history of the disease may be challenging. Initial clues include a low caeruloplasmin level, (the protein in the blood that binds copper that is reduced in WD), a low alkaline phosphatase level (one of the commonly tested liver enzymes) and a low level of red blood cells which are broken down quickly in this setting due to the toxicity of free copper, a process called *haemolytic anaemia*. A 24 hour urine copper level can take several weeks to come back, so is not always helpful. If there is time a liver biopsy can be performed and stained for copper; this is done via a neck vein under X-ray which is safer than a needle into the liver from the side of the body when the clotting is prolonged.

Sometimes urgent treatment with the drugs used for Wilson's disease can rescue a patient in the early phase of acute liver failure but more often than not patients are considered for *emergency* or *super-urgent* liver transplantation, providing they meet certain criteria. Liver transplantation can be life-saving in the setting of acute liver failure due to WD and is curative. In the UK since 2007 there have been **67** liver transplants for Wilson's disease, of which **46** were done as *super-urgent* i.e. two-thirds of transplants for WD end up being done in an emergency.

> *"...you can never be "too old" to present with Wilson's disease"*

Validated scoring systems can be used to aid decision making about transplantation because they tell us whether a patient is unlikely to survive without this procedure. The most familiar system is the 'Nazer' score developed at King's College Hospital, London, which incorporates variables related to liver failure. A benefit in having made the diagnosis of Wilson's disease is that there is a specific category by which patients can be *super-urgently* listed. This means if there is any sign of hepatic encephalopathy (confusion) along with abnormal clotting, the team does not wait for the patient to get worse before putting them on the list for emergency transplantation.

The survival following transplantation for liver failure is very good although outcome drops off above the age of 70. Figures are now not dissimilar from routine (*elective*) transplantation for cirrhosis i.e. around 95% alive at a year following transplant and 80-85% at five years following transplant. The majority of patients return to an excellent state of health and David's outcome is a testament to that. David will have to be maintained on drugs to suppress his immune system, but his Wilson's disease is cured and he certainly had life-saving treatment. We wish him many successful marathons in the years to come!

Dr Bill Griffiths
Consultant Hepatologist
Addenbrooke's Hospital,
Cambridge

Laboratory Tests in the Diagnosis and Monitoring of Wilson's Disease

Many people who have been tested for the diagnosis of Wilson's disease will have had a blood or urine sample taken for analysis and in some cases may also have had a liver biopsy, where a small piece of tissue is removed from the liver for analysis. For patients receiving treatment they may also be routinely monitored to determine whether any changes to their prescription are required.

So what happens to these samples once they have been taken and how do the results contribute to a diagnosis or not and how do they inform the multidisciplinary team that are treating the patient?

Sample Types Used

In the case of a **blood sample** this will usually be taken by a trained phlebotomist or other healthcare professional, using a procedure called venipuncture, whereby a sample of blood is taken into a special tube that draws the blood out of the vein via a vacuum in the tube. The blood is allowed to clot to form two separate layers, the red blood cells and either plasma (if the blood is allowed to clot naturally) or serum (if an additive is present in the tube to trigger the clotting process). Both serum and plasma can then be used for the determination of copper and/or the copper containing protein ceruloplasmin.

In the case of **urine samples**, the clinician would like to know how much copper is being excreted via the kidney and because the flow of copper varies with the time of day, due to how much liquid is consumed or what foods have been eaten, the most accurate way to do this is to perform a 24 hour urine collection. This averages out any fluctuations in copper excretion, but is only useful if the timing of the collection is accurate and does not stop early or go on longer than expected. Once again the copper level is measured.

The final sample type is the **liver biopsy** which is obtained using a more invasive procedure with more risk involved, which the clinical team will discuss with the patient prior to obtaining permission. Either a needle is inserted into the liver to remove a small core of tissue, or the liver is sampled via the vascular system to obtain a tiny piece of tissue. This sample is then sent for tissue copper analysis.

Methods for Diagnosis and Monitoring

The aforementioned samples are then sent to the clinical laboratory carrying out the tests. For the measurement of copper in the liquid samples, they are diluted for analysis. The solid tissue samples need to be converted into a liquid to facilitate analysis. This is done by dissolving the tissue in an acid solution by heating it in a controlled way using a laboratory microwave oven.

There are a number of analytical methods for the measurement of copper, but the state-of-the-art uses a technique called **inductively coupled plasma mass spectrometry** (ICP-MS). One such instrument is shown in ***Fig. 1.***

Put simply, the plasma, which is like a very hot flame at about the temperature of the surface of the Sun, turns the contents of the liquid sample into dry particles, then into atoms and finally generates ions (atoms with a positive charge), these will include ions corresponding to the 2 isotopes of copper which are measured in the instrument at mass-to-charge ratio ^{63}Cu and ^{65}Cu. Sample copper concentrations are measured against standards of a known concentration and quality control (QC) specimens of serum and urine, which have a known content of copper. By ensuring that the instrument determines the correct value for the copper concentration in the QC, we have confidence that the sample concentration will be correct.

Fig.1. Inductively coupled plasma mass spectrometer for the measurement of copper.

To show that a laboratory is proficient at the tests undertaken and provides accurate results to clinicians, it needs to be accredited by the UK Accreditation Service (UKAS) and participate in a Quality Assurance (QA) scheme. In the UK the QA scheme is based at the Trace Element Centre in Guildford and sends out serum, urine and liver tissue to laboratories around the world who undertake the measurement of copper. Each participant determines the copper content and returns their result, which is collated for each laboratory. Depending on how close to the assigned copper value for each specimen the laboratory is, it is awarded a score. These scores show the laboratory performance over time and if the test is not being performed accurately helps the laboratory to improve its performance. If over time the performance does not improve then the laboratory may be advised to stop performing the test.

Interpretation of Patient Results

Once the concentration of copper in the serum, urine or liver has been measured a report is issued to the requesting clinician. This will contain the patient result followed by a "Reference Range", which will put the result into the context of what the laboratory considers the copper concentration should be for a healthy population of people; the values are derived from studies on a suitably large population. In the case of copper, because it varies with age the Reference Range will be split into age groups. ***Table 1*** shows the copper Reference Ranges for serum/plasma, urine and liver tissue used in our laboratory.

Whilst the biochemical tests described can aid in the interpretation of an individual's copper metabolism, the diagnosis of Wilson's disease is made by the requesting clinician and will take into account the clinical presentation as well as tests carried out by other healthcare professionals (Kayser-Fleischer rings, neurological assessment, liver function, haematology etc.).

Serum/Plasma	µmol/L
<4 months	1.4 to 7.2
4 <6 months	3.9 to 17.3
6 months <19 years	11.0 to 27.4
>19 years & adults	11.0 to 25.1
Ceruloplasmin	0.2 to 0.45 g
Ceruloplasmin (WD)	<0.25 g
Urine	**µmol/24hr**
Adults	<0.70
Adults post-penicillamine	<12
Wilson's disease	>1.57
WD post-penicillamine	>25
Liver Tissue	**µg/g**
Undiseased liver	<55
Wilson's disease	>250

Table 1: Reference Ranges used by the Royal Surrey

The Reference Ranges in ***Table 1*** highlight why it is difficult to make the diagnosis in younger patients as it can be seen that a low serum copper may not differentiate between the healthy and those with the disease. For this reason, clinicians may choose to use a "dynamic function" test in children, which is a test carried out over a period of time and which can show differences between each group. In the case of Wilson's disease this involves determining a baseline urine copper excretion, prior to the administration of penicillamine, followed by a subsequent urine collection after 24hr. *Table 1* shows that there is a marked increase in excretion of copper in Wilson's disease patients because the penicillamine chelates the excess copper that has accumulated in the tissues and makes it available for excretion by the kidneys. In healthy individuals copper is excreted via the bile into the faeces and there is no accumulated copper in the tissues, so the urine excretion is much lower.

New Tests are Being Developed

Whilst some cases of Wilson's disease can be diagnosed using the aforementioned methods, in more subtle presentations none of the current routine tests on their own is able to do this.

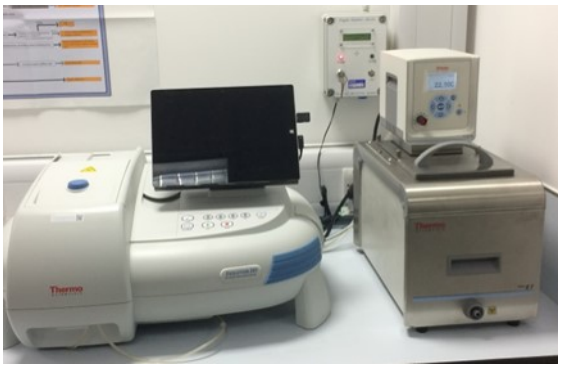


Fig 2. Ultra-violet/visible spectrophotometer used to measure the oxidase activity of the copper containing protein ceruloplasmin.

Wilson's disease patients have a gene that lacks the ability to make a membrane bound protein known as ***ATP7B***. Absence or reduced function of this protein leads to a decrease in the normal liver excretion route of excess copper via the bile and the failure to incorporate copper into ceruloplasmin, a protein that oxidises iron (termed a ferroxidase) so that iron can be incorporated into haemoglobin. Functionally these changes can lead to an increase in copper levels in the liver, a low ceruloplasmin in the serum and a disrupted haematology in some patients.

Wilson's disease patients have a low circulating concentration of ceruloplasmin and this has also been used in the diagnosis of the disease. However the methods used for this measurement are not specific to the copper containing form, but include the non-metallated form and hence overestimate the level of the protein of interest. It is for this reason that pioneers such as **John Walshe** recommended that it is better to measure the activity of the protein (how much it can oxidise another substance) which gives a much more accurate concentration of the metal containing form (without the metal it is inactive). This enzyme activity based method has been developed in Guildford following funding by the Wilson Disease Association (WDA) US to purchase a ultra-violet/visible spectrophotometer (**Fig. 2**), which can measure the change in absorption of a coloured complex as it is oxidised by ceruloplasmin.

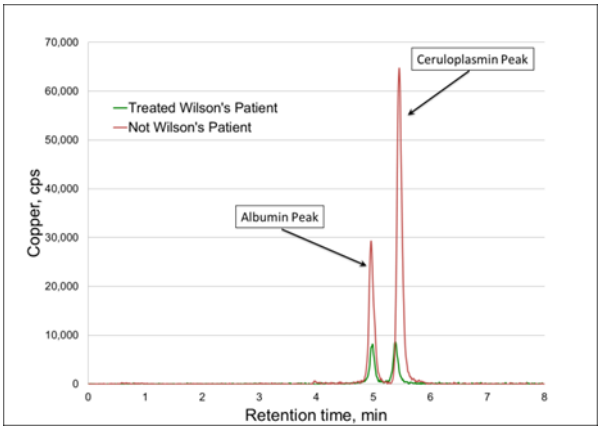


Fig.3. Separation of copper containing proteins in serum from treated Wilson's patient and a patient without disease.

The Supra-Regional Assay Service (SAS) Laboratory at the Royal Surrey County Hospital in Guildford has been involved in the development of new biochemical tests related to Wilson's disease for the last 2 years and also offers more specific tests for the measurement of ceruloplasmin than those routinely offered. Some recent studies funded by WDA have focused on measuring the exchangeable copper which is the amount of serum copper that is not held within ceruloplasmin and is in effect the fraction that causes the damage in patients. Copper circulates in the body bound to proteins which include ceruloplasmin, albumin and transcuprein **(Fig. 3)** and by measuring each form separately it is possible to determine the amount of toxic copper present. When the exchangeable copper is divided numerically by the total serum copper concentration the derived ratio clearly differentiates between Wilson's disease patients, all other genetically related forms and healthy individuals. This approach can also be used to monitor the effectiveness of treatment.

By studying patients with the disease using the new methods we hope to improve patient treatment.

Dr Chris Harrington FRSC FRCPath. Deputy Director, SAS Trace Element Centre, Royal Surrey County Hospital.

Helen's Story

Hi there everyone. Some of you may recognise me as a Facebook member of the Group, through which I have been approached to share my story.

I was born in a small town called Rawtenstall which is about 20 miles north of Manchester. I have just turned 44 years of age and was diagnosed with Wilson's disease in the year 2000.

I have always been a happy, positive soul. I had a lovely childhood growing up with my Mum and Dad, two younger sisters and lots of friends. In fact the girl who lived next door to me is still my best friend to this day. I loved school and I loved to learn. When I was in the 4th year (year 10) aged 15 at high school, I became very lethargic: I had no energy and didn't want to do anything. I had a lot of time off school. After numerous trips to the doctors I was told a year later that I may have had glandular fever that wasn't diagnosed. Even after all the time off school I still did very well in my GCSEs gaining all As and Bs with one C.

The following term I enrolled in my school's 6th form. After six months I found I wasn't enjoying it and told my parents I wanted to leave. They said that I could, but that I had to find something else to do, so I got myself a job. I was earning money and lots of my friends were working also, so I became a bit of a party animal. I was out on Thursday, Friday and Saturday nights and sometimes even Sunday! Then out of the blue I just got extremely tired again, so I took time off work. One week became two weeks, then the weeks turned into months. I sank into a depression; I hardly left my bedroom, I hardly even got out of bed. I used to pretend to be asleep so I didn't have to see anybody.

When I was 20 the eldest of my two younger sisters was involved in a car crash on her way home from school. Tragically, the young lad driving the car died and all the passengers in the car were injured. This incident shocked me so much, it made me leave the house. I needed to know my sister was okay. I went to the hospital; it was nerve racking and I felt like I was going to pass out all the time, but she was my little sister and I had to know she was okay. I left the house and nothing bad happened to me. I knew I could do it again.

Over the next few months I got out more. I had some panic attacks when there were a lot of people about but on the whole I was okay. By the following September I felt confident enough to enrol in college. I started to study Media Studies, Film Studies and English (films have always been a passion of mine). One lunchtime during college my friends and I were in the pub having a game of pool. I got up from my bar stool and fainted. I was out cold for about 4 or 5 minutes.

When I finally came to, my speech was slightly slurred and I didn't feel at all right. My friends then made sure I got home safely. A few weeks later I went to the doctors, who sent me straight to the local hospital. The doctor there said I'd had a mild stroke and that my speech would never go back to how it was. He arranged for a speech therapist to come and see me. I felt so stupid as I'd always been so clever and now I couldn't even talk! I sank into a depression again and isolated myself from everyone and everything.

> *"I hardly left my bedroom; I hardly even got out of bed"*

Over the next five years the more common symptoms of Wilson's disease began to surface. The symptoms that made my family extremely concerned, the symptoms that I had no control over and didn't understand. My whole body shook. I had paralysis in my mouth, would often choke on food and I was drooling all the time (which I found very embarrassing). I constantly felt tired and light-headed and often passed out. I walked as if I was drunk. In the end I couldn't do anything for myself: I couldn't fasten my own buttons on clothes, tie my trainers, clean my own teeth, brush or wash my own hair. The only thing I could do was feed myself, but I couldn't cut my food and often choked. I was dying but I didn't know why.

Finally, in 2000, after numerous trips to the doctors with my concerned mum, my GP got in contact with Manchester Royal Infirmary about me and I was asked to go to hospital straight away. I was admitted and stayed there for three weeks. I hated it. I didn't understand what was happening to me or why I had no control over it. I felt completely lost and alone. Whilst in hospital I had numerous tests: CT scans, electrodes attached to my head, a nuclear scan, MRI scan, an endoscopy, a liver biopsy and slit lamp test. During this

eye examination, the doctor was getting trainee doctors from all over the hospital to come and look at my eyes because I apparently had textbook Kayser-Fleischer rings around the corneas; you could actually see them with the naked eye. I absolutely hated my stay in hospital but at least doctors now knew what was wrong with me. I had Wilson's disease and would be started on treatment straight away. It was a lot to take in. I had a disease. Was I going to die? What was going to happen to me? Did my younger sisters have it, too?

Once my medication had been sorted out, I left hospital and went home. It was so good to be home with all my own things around me. I started to take my medication but I didn't know what to expect as I didn't know what damage had been done. After about 3 - 4 weeks of taking penicillamine, my body tremors stopped and I didn't drool at the mouth anymore. My speech was still very incoherent and my walking was terrible, but at least the things I was most embarrassed about had stopped. And only one of my sisters had been given a probable diagnosis of the disease.

Obviously, as I was so ill I was unable to work so I looked into which benefits I might be able to claim. After about 6 months a doctor came to assess me at home to decide if I was fit for work or not. He decided that I **was** even though my parents had to translate what I was saying for him. I kept banging into things around the home and struggled with the tasks he gave me to do. When I got the letter from the Department of Work and Pensions stating that I was fit for work, my dad and I went to my doctor's and my doctor got all the reports from the hospital and the report that this other doctor had written and sent copies of them all to me highlighting the differences between them. This was a huge help and a few months later the decision was overturned and I was awarded Disability Living Allowance.

> *"...you could actually see them with the naked eye."*

Over the next few months my cognitive capabilities started to return. My memory improved dramatically, my balance began to return and I stopped falling over or banging into things everywhere I walked. I knew then that having Wilson's disease wasn't going to define me as a person; I wouldn't let it.

Although I wasn't well enough to get a job, I still wanted to do something so I did some volunteer work at a local charity shop and also volunteered at my old primary school. I absolutely loved helping at my old school as it gave me a sense of purpose. I was helping these children to learn.

With each month that passed my symptoms were diminishing more and more. I started to socialise again with my friends. On one such occasion in January 2003, I met Aidan on a night out and we *clicked* straight away. He was so easy to talk to and he made me laugh. That night he walked me home and we arranged to see each other the following day. We haven't been apart since. I told Aidan that I had Wilson's disease from the start, so when I moved in with him four months later, he bought me bottled water in case his water pipes had copper in them, which I thought was very sweet.

> *"Wilson's disease wasn't going to define me as a person; I wouldn't let it."*

I enjoyed volunteering at my old primary school so much, I decided to see if I could get some qualifications to help me get a job. I gained a level 2 and 3 in Support Teaching and Learning with a distinction grade in both! In December 2004 our daughter was born after an uneventful pregnancy. The hospital actually kept my placenta for histology testing as they said it's rare for a Wilson's disease patient to give birth naturally. When our daughter was about one year old I got a job as a teaching assistant supporting a child on a one to one basis. I still have the same job now supporting a child with special needs. I truly love my job and find it very rewarding. In 2008 our son was born, but this pregnancy wasn't as straight forward and I was advised not to have any more children.

I have everything I need: a loving family, my own home and a job I love. Oh and I have Wilson's disease too! Wilson's disease doesn't define me as a person; it's just part of me that makes me who I am. If I had a choice, would I want things to be different? It's something I don't tend to think about as I feel there's no point. In my eyes I could be a lot worse off. So what if my speaking voice isn't the same as it used to be, I can still communicate and be understood. So what if I walk with a slightly uneven gait, I can still get about. And, so what if my co-ordination is slower and more awkward, I've adapted the way I do things but at least I can still do them.

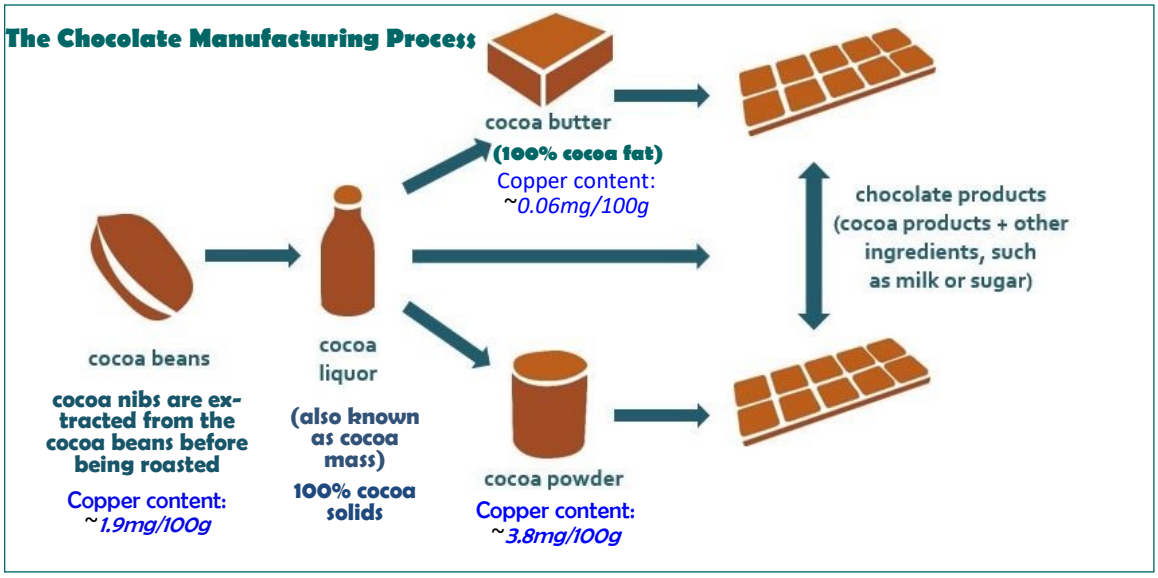
Don't get me wrong. I have bad days when I'm clumsy and my speech can be quite difficult to understand, but these are few and far between. Wilson's disease doesn't control my life. I don't consider myself to be *suffering* from Wilson's disease. I think of myself as a person *living with* Wilson's disease and as such I love my life. Thank you for taking the time to read my story.

The Chocolate Manufacturing Process

by Katy Quinlan

Having Wilson's disease itself is bad enough but it also means that you have to watch your diet! Copper is present in most foods and limiting its intake is advised, particularly for newly diagnosed patients. For me, the worst part of having a reduced copper diet is limiting the amount of chocolate I eat!

My name is Katy and my story appeared in the 2017 newsletter. I work for a chocolate manufacturing company in the UK and I thought I could share with you how chocolate is made and why some chocolate contains more copper than others. Here is a simple diagram of the chocolate making process with a written explanation afterwards.



1. The cocoa nibs (from inside the cocoa beans) get roasted and then go through a grinding process to be turned into cocoa liquor (also called cocoa mass). This is 100% cocoa solids.
2. This cocoa liquor/cocoa mass/100% cocoa solids is then pressed to extract the cocoa butter (cocoa fat). The remaining part of the "solids" may get pulverized into cocoa powder or remain as cocoa liquor/cocoa mass/cocoa solids to be used in the chocolate process.

The heavy metals (including **copper**) are concentrated in the "solid" part of the cocoa products rather than the butter/fat. The average copper content in the cocoa nibs is **1.9mg/100g** and in cocoa powder it is **3.8mg/100g.**

Any chocolate products with **high** levels of cocoa solids e.g. dark chocolate with ***70% Cocoa (****or more)* written on front of packaging, are likely to be a **high** source of dietary copper.

However, when making **white** chocolate, **only** the "fat" portion (cocoa butter) of the cocoa liquor/cocoa mass/100% cocoa solids is used as a "cocoa" ingredient. Cocoa butter only has a trace of copper in it, meaning that any white chocolate is suitable for patients on a **low copper diet**.

The following table shows the typical ingredients in different types of **100g** bars of chocolate. At the bottom, I have given a range of values of copper content in each. Quantifying copper is not an exact science, but this does at least give you some guidance and can act as a comparison to other foods on the WDSG-UK Food Chart, which is available to download off the website www.wilsonsdisease.org.uk. *Happy Eating!*

Table of Ingredients in Different Bars of Chocolate (*typical composition)		
Dark/Plain chocolate bar 100g	**Milk chocolate bar 100g**	**White chocolate bar 100g**
Cocoa Liquor/cocoa mass **Cocoa** Butter Sugar – Extras e.g. **cocoa powder**, emulsifiers, flavours, vegetable fats Copper: 0.5 – 1.8mg/100g	**Cocoa Liquor/cocoa mass** **Cocoa** Butter Sugar Milk ingredients Extras e.g. emulsifiers, flavours, vegetable fats Copper: 0.18 – 0.43mg/100g	– **Cocoa** Butter Sugar Milk ingredients Extras e.g. emulsifiers, flavours, vegetable fats Copper: 0.01 - 0.12mg/100g

Dorothy's Story

by Dorothy Patterson

My name is **Dorothy**. In **1967** I was living in Gibraltar with my mum and dad and my sister, **Yvonne**, who was only 5. My father was in the RAF and I had just sat my 11+ exam and passed with flying colours. I had a place at North Walsham Girls' High School in Norfolk, near where my father was about to be posted next.

One holiday, we set off from Norfolk to visit our grandparents in Scotland. I was always car sick, so dad spoke to the Medical Officer on the base and he gave me a travel sickness pill to take, cutting it into four and suggesting I take a quarter every four hours. We travelled overnight, so I only needed one pill. On arrival we girls went to bed to rest.

After lunch, mum, my sister and I went for a short walk and I collapsed. Mum managed to get me home and back to bed. By the evening, I was feeling fine again, so we set off to visit our other grandma. She opened the door to us and I smiled at her. Everybody was busy chatting when I realised that the smile had set on my face and I couldn't get rid of it. My parents immediately rang a doctor who sent us to the Aberdeen Royal Infirmary for tests. At first, they thought I might have *lockjaw,* but then decided I had probably had an allergic reaction to the sickness tablet I had been given.

Back home, I began to speak more quickly and was slurring my words. I wasn't aware that I was doing it, but people were continually asking me to repeat myself which I found very frustrating. I hated hearing my voice on a tape recorder. I was having difficulty with my balance, and my gait was getting stiffer when I walked.

I was referred to the local hospital in Norwich and after several months of tests including bone marrow tests and a lumber puncture, nobody could diagnose what was wrong with me. When exploratory brain surgery was mentioned, mum flipped and instead agreed to me going to St Bartholomew's Hospital in London for a second opinion. After various tests, a **Dr Paul Currie** asked if he could look in my eyes through a slit lamp.

By this time I was shaking spasmodically and beginning to drool. It was a year since symptoms first began. I had no idea what was happening to me only that I seemed to have no control over my body. Having looked at my eyes, Dr Currie announced that I had Kayser-Fleischer rings and said, "You've got Wilson's disease, Dorothy!" That was a Eureka moment! At last I had a name for what was wrong with me. My family all had to be tested and Yvonne was told that she had it, too. We were both started on penicillamine.

I continued to be seen at Bart's, but as we were now living in Norfolk, Yvonne was being seen by **Dr John Walshe** at Addenbrooke's which was much nearer. Shortly afterwards, it was decided that I would see him too. We were told at that time that there were only **52** patients with Wilson's disease living in the UK and my mum was proud that she had **2** of them!

School was difficult as some of the girls called me names and were generally horrible to me. I had missed so much school, I was made to repeat a year. I hated that. At the same time I kept getting fractures. At one stage I had both wrists in plaster at the same time! One day I fainted getting off the school bus and Dr Walshe decided to change my meds and put me on TETA (triethylenetetramine) instead. TETA is the technical name of trientine and Dr Walshe and **Kay Gibbs** used to make the capsules themselves in their laboratory.

On several occasions in the early days I would stop taking my meds. I convinced myself that I didn't need them any more. Such a mistake. My speech would suffer and mum always picked up that there was something amiss. The drooling came back once and as a teenager that was horrendous.

I married at 18 and had the first of my two daughters by my 21st birthday. Again I thought I could stop taking my medication. Mum realised straight away and took me to one side. She said that if I didn't look after myself, I wouldn't be around to see my daughters grow up. Dr Walshe said the same thing. This shook me so much that I haven't stopped taking them since and not only have I seen my daughters grow up, by my five grandchildren, too.

I am quite a determined person and am always smiling. I like to help others. My husband and I travel a lot thanks to my awesome mum, **Sheina**, and we are now accompanied on our travels by our 6 month old puppy, **Bailey**. I haven't let Wilson's disease stop me in any way. I find a way round any problem, unorthodox as it sometimes is, but I always get there in the end. My mum was once told before my WD diagnosis that my life expectancy would be 37 years, but I am pleased to report that I am still here at 63. I would like to thank Dr Walshe for never giving up on me and also, my mum, whom I love to the stars and back!

THE WILSON'S DISEASE SPECIAL INTEREST GROUP (WDSIG)

What is it?

Through the British Association of 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. WD lends itself well to this new initiative, being a rare disease that has several types of clinician involved in its care.

Who is in it?

The WDSIG comprises clinicians in hepatology, neurology, clinical chemistry, paediatrics and psychiatry as well as research scientists and representatives from **WDSG-UK** and Public Health England (**PHE**). At this time there are nearly 100 individuals from across the UK included in the WDSIG member database.

What is the remit of the WDSIG?

The WDSIG functions primarily to improve care for patients with WD. The group recognises that care and access to expertise in WD is variable across the UK. WD suffers at times as with any rare disease in delay in diagnosis, lack of multidisciplinary input and inconsistent monitoring, which can result in unnecessary harm. Research is difficult to coordinate without collaboration and new therapies are coming online for WD. The WDSIG hopes to improve research into WD as well as work towards a better model of healthcare for WD which can optimise outcome. Through PHE the WDSIG hopes to answer questions about how WD is spread across the country and how patients are being treated.

What has happened so far?

Three meetings of the WDSIG have been held (all in London) - 14 December 2017, 14 June 2018 and 14 February 2019. Between 24 and 34 participants have attended these meetings. Minutes from each meeting have been uploaded to the BASL website along with some of the slide presentations.

The WDSIG has developed four core themes:

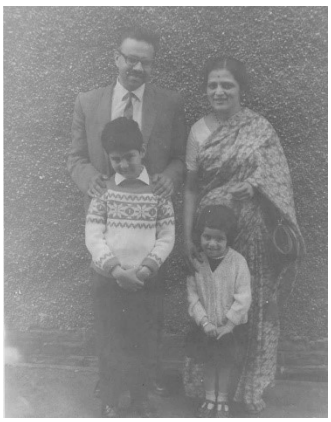
- **Service delivery** – in accordance with the UK Strategy for Rare Disease a preliminary proposal to NHS England for a 'hub and spoke' model for WD has been accepted for further evaluation. This document outlines how a number of Centres of Excellence would function to deliver care for WD via multidisciplinary clinics, networking with neighbouring hospitals. As a consequence of trientine dihydrochloride being now commissioned by NHS England due to its price, WD centres in England have needed to be defined so there is a precedent there now in place.
- **National Registry** – with the support of the WDSIG, Public Health England (PHE), specifically NCARDRS (National Congenital Anomaly and Rare Disease Registration Service), is establishing a Rare Disease registry which aims to capture relevant clinical data on all WD patients in England. NCARDRS has a special privilege to collect hospital episode data from any Trust, although patients can opt out if they wish. PHE is also collecting prescribing data, mortality information, data from trace element and genetics laboratories, and data from the national Transplant database. From these sources PHE hopes to build up an accurate picture of WD across the country.
- **Clinical care** – in bringing together the clinical and laboratory expertise the WDSIG aims to specify standards by which patients should be diagnosed and monitored. The hope is that no patient with WD is left without proper monitoring of the various strands of their condition. Dissemination of 'best practice' by Centres of Excellence will be key here.
- **Research** – the WDSIG will play an important role in WD research. The group recognises the scientific and clinical research on WD occurring in the UK, and provides a forum to discuss novel research ideas and foster collaboration. The WDSIG acts as an interface for Pharma where there are several new drugs for WD currently being trialled or in development. The WDSIG has worked to include the UK along with our European neighbours in these studies which hope to address some of the unmet needs in WD.

The group is planning a 4th meeting in the Autumn and has plenty of work to do in the meantime! Further information is available at**:** https://www.basl.org.uk/index.cfm/content/page/cid/31

Bill Griffiths,
Consultant Hepatologist and WDSIG lead.

Alpana's Story

My WD story starts in **1973** when I was 8. I was living in Bradford, West Yorkshire with my parents and my older brother Ketan, aged 12. We had been on holiday that summer to India. My brother started feeling ill a few weeks after returning from India and he then visited his GP. Sadly he deteriorated rapidly and due to a delayed diagnosis and medical negligence he passed away in January 1974 due to WD.



Ketan and I with our parents, May 1968

Following his diagnosis my dad and I visited Addenbrooke's Hospital in Cambridge and met **Dr Walshe** and **Kay Gibbs** for the first time. It wasn't clear whether I had WD so I underwent a series of lab tests, urine tests and imaging studies under the caring and watchful eye of Dr Walshe. That led to the confirmation that I did indeed have WD. I was started on penicillamine.

Following that stay, I began my regular trips to Cambridge every 1 -2 years for testing and monitoring. At that time my parents were not allowed to spend the night in the hospital room with me so my dad would stay in an adjacent B & B, and I would be cared for by the excellent staff at Addenbrooke's. I remember the special times I spent with my dad in Cambridge. In between my tests we would often go out to the nearby pond and feed the ducks. During those visits to Addenbrookes I was so inspired by Dr Walshe and I vowed to go into medicine; my desire to become a doctor began.

Although the next few years were uneventful for me medically, our family was blessed with the birth of my younger brother, Kedar. Eventually I was old enough to attend Addenbrookes by myself. In fact I remember meeting other WD patients and hearing their stories.

In 1988 I graduated from Newcastle Medical School and became a GP. After marrying my husband, a US citizen, I moved to California in 1995 and continued to work as a GP. That same year, two weeks after I left the UK, my dad passed away. We tried to find solace in believing he was reunited with my brother, Ketan.

My WD story has not been without its ups and downs. Although I have been mainly well, I was not the most compliant in taking my penicillamine during my pregnancies in 1996 and 1998. I became ill and that led to my 1st liver biopsy in 2000. After an increase in the penicillamine and the addition of zinc I improved and stabilized over the next few months.

> *"my desire to become a doctor began"*

The story is ongoing with the onset of abnormal labs again in June 2018. My dear mum passed away during one of my frequent visits to the UK in May 2018 and I was unexpectedly without my medication for two weeks. I had a recent liver biopsy that shows a large amount of copper in the liver, and my penicillamine dose has again been increased. I am also continuing to take zinc.

The next few months and years will continue to be challenging at times and I certainly don't know what is in store. But I have been extremely fortunate to have had exceptional health largely due to my early diagnosis and the diligent medical care I have received. As a mother myself, I can't imagine what my mum and dad went through losing their sweet 12 year old son and I will forever be indebted to my brother Ketan, Dr Walshe and Kay.

IN MEMORIAM

Alpana and her younger brother, Kedar, sadly lost their mother, **Anjali,** in May 2018. The family had remained in touch with Dr Walshe since Ketan and Alpana's diagnoses in 1974 and had always taken an interest in the plight of other patients with the disease. To this end, Alpana and Kedar kindly decided to ask friends and family to make donations to WDSG-UK in memory of their mother and in so doing raised the very generous sum of **£1,200** for the Group.

Alpana and her mum, July 2012

WDSG-UK Notices & Updates

In this and subsequent newsletters, we hope to keep you updated on current research projects, drug trials and general information on matters relating to Wilson's disease.

Research

University of Sheffield, Dept of Neuroscience

Prof. Oliver Bandmann and his team continue their work investigating mitochondrial biomarkers in Parkinson's disease and other neurodegenerative diseases, including Wilson's disease. If abnormalities are found in mitochondria, there is the possibility of using so-called "mitochondrial rescue" drugs as treatment. Cells obtained from patients by skin biopsy will be investigated for their mitochondrial function, and will also be processed to convert them into neurones (brain cells) on which further studies can be performed.

National Hospital for Neurology, Queen Square, London

Further to their article in last year's newsletter, **Prof. Tom Warner** and **Dr Sam Shribman** of the National Hospital for Neurology, Queen Square, London, are delighted to announce the launch of the CROWD Study. Sam writes,

"**An update on the CROWD study: A UK-wide research study on Wilson's disease**

It was a great pleasure to be invited to the annual meeting in 2018 to present our plans for research on Wilson's disease and meet so many members of the Wilson's Disease Support Group—UK. For those who were not able to make it, our team at the UCL Queen Square Institute of Neurology is working with neurologists, hepatologists, metabolic medicine specialists and paediatricians from across the UK on a research study on Wilson's disease called the **CROWD** study. We have two main goals with our research:

1) In the first part of the study we are looking at why some people develop liver problems but others develop neurological or psychiatric problems. Everyone with a diagnosis of Wilson's disease, who is age 16 years or over and lives in the UK is able to take part. It involves completing an online questionnaire, talking to us on the phone and then sending us a saliva sample for genetic research in the post. More information about how people can take part is included below.

2) In the second part of the study we are trying to understand how to monitor the effect of Wilson's disease on the brain using tests, such as MRI scans. A smaller group of people will be involved in this part of the study, which involves coming to the National Hospital for Neurology and Neurosurgery, in central London.

The Study was launched in December 2018 and we are very grateful to all of those people who have already decided to participate. We have now received more than 50 responses for the first part of the study and more keeping coming in each week. While it may be some time before any results become available, we will continue to keep you updated on our progress and let you know about any findings over the next three years. I am looking forward to giving members an update in person at the annual meeting in Cambridge on 21 July 2019."

If you are interested to find out more about our research on Wilson's disease then please visit www.thecrowdstudy.com and feel free to contact us directly at crowd@ucl.ac.uk or on 07739 751200 if you have any queries.

Drug Trial — Birmingham, Cambridge and Guildford

Efficacy and Safety of WTX101 Administered for 48 Weeks Versus Standard of Care in Wilson Disease Subjects

Prof. Ala from the **Royal Surrey County Hospital** writes: The 'Phase 3' FOCuS multi-centre clinical trial evaluates WTX101 (bis-choline tetrathiomolybdate), a copper-protein-binding agent with a novel mechanism of action, for the treatment of Wilson's disease. It is anticipated that the FOCuS study will enrol at least 100 Wilson's Disease patients, aged **18** years or over, to receive once-daily WTX101 oral therapy or standard of care across North America, Europe, UK and Israel.

The drug has high affinity and specificity to copper detoxifying excess copper in the liver and in the blood by forming stable complexes with copper and copper carrying proteins in the blood stream called albumin that are then cleared through bile, the natural elimination route of copper. This unique approach to copper control has the potential to improve symptoms and associated disabilities in Wilson's disease patients, which was demonstrated in the earlier 'Phase 2' trial. The current Phase 3 FOCuS study is the first randomised controlled trial to support approval of a new treatment

option for Wilson's disease and to evaluate the different profile of **WTX101** in this head-to-head study versus standard of Care (SoC).

The profile of this investigational drug at a previous early Phase 2 stage shows the relatively rapid control of clinical symptoms in combination with the simple once-daily dosing regimen and its promising side effect profile which could help improve compliance to therapy.

The Phase 3 FOCuS clinical trial assesses the efficacy and safety of WTX101 administered once daily for 48 weeks, compared to standard of care (SoC), in patients with Wilson's disease. The study is currently enrolling patients with liver and/or neurological symptoms, who are *treatment naïve* and will shortly consider those patients who have previously received SoC therapy at centres from Birmingham (**Dr D. Nicholl**), Cambridge (**Dr B. Griffiths**), Guildford (Prof A. Ala). It is anticipated other centres across the UK may also open the trial this year and the age criteria for entry will be from **12+**

Public Health England—Wilson's Disease Pilot Study—Update

Mary Bythell, Head of Rare Disease Registration, is working on a project to identify people with Wilson's disease in England thereby providing a national prevalence rate for the disease. At the same time, information will be collected about diagnosis and treatment to help commissioners make the right healthcare decisions and improve patient care. For more detailed information on the project, please refer to Mary's article on *p19* of the 2018 newsletter archived on our website: <wwwwilsonsdisease.org.uk>. This is the first project of its kind that NCARDRS is carrying out and Mary writes:

"The National Congenital Anomaly and Rare Disease Registration Service (NCARDRS) continues to work to register cases of people with Wilson's Disease to support clinical care, policy development and research. We now have **163** people with Wilson's disease registered with NCARDRS. We have been working to collect this information from several sources. These include:

a) Working with the Supra-regional Assay Service to set up a system of reporting new cases to NCARDRS. We have had the first new patients reported to us this way.

b) Data sharing with the CROWD Study based at University College London (http://thecrowdstudy.com/), who are sharing patient NHS numbers and their doctors with us, if participants agree to this.

c) We are also continuing to contact hospital trusts to ask them to let us know about their cases.

Dr Michelle Camarata, who is based at the Royal Surrey County Hospital, will be joining NCARDRS on an honorary contract to work on Wilson's disease. She will work with us to register more cases, support new ways of reporting new cases like genetic test results and use the data collected by NCARDRS to write reports about Wilson's disease in England. NCARDRS looks forward to giving the Wilson's Disease Support Group– UK an update at its annual meeting in July.

If you would like more information about NCARDRS, please contact Mary Bythell or **Jeanette Aston** at phe.ncardrsrd@nhs.net."

Supply of Penicillamine in the UK (March 2019)

We understand that there are two pharmaceutical companies in the UK that supply penicillamine: **Mylan** and **Kent Pharmaceuticals**. Since **January 2019** Mylan has had a "manufacturing issue" with the **250mg** tablets, although their **125mg** tablets have been widely available. We understand that the issue should be resolved by 10 May and we hope therefore that the **250mg** tablets will be back with the wholesalers, then. Kent Pharmaceuticals have not experienced any such problems and continue to supply their wholesalers with their **250mg** tablets.

Supply of Trientine in the UK (March 2019)

We are delighted to inform you that after exhaustive and exhausting discussions, NHS England has approved the continued use of trientine for patients in England who need it. Univar, the UK manufacturers of trientine, foresee no difficulties with its supply whatever the Brexit outcome. There is currently plenty of stock in the UK. If you are having difficulty getting hold of your trientine, please contact **Valerie** direct.

The Wilson's Disease Patient Register—UK

WDSG-UK strongly supports research into all aspects of Wilson's disease. If you are a **researcher** and would like to advertise any studies that you propose, or if, like Sam, you wish to make use of our Patient Register, then please contact either **Jerry Tucker** or Valerie Wheater (details on the back cover). Alternatively, if you are a **patient** and are interested in taking part in research but have not yet joined our Patient Register, please consider doing so. A pamphlet and information sheet about the Register can be downloaded off our website <www.wilsonsdisease.org.uk> together with a registration form. We already have **65** patients on the Register and would very much like to have you, too!

Members' News 2018-19

Our President **Dr John Walshe** is always interested in news about the Group and will happily give advice, when requested. Having devoted his professional life to the treatment and management of over **300** patients worldwide, he has a wealth of experience from which to draw. At the end of the month he will have reached his **ninety-ninth** birthday and he very much hopes to make his century next year.

As we live close to one another, we keep in touch and I also facilitate the occasional visit to him of doctors, former patients and other interested parties. In **March 2018** I received one such request for a visit. It came from a patient in the US who was embarking on writing a book about Wilson's disease and felt that it would not be complete without interviewing *the great man,* himself. She wanted to talk to him about the evolution and developments of treatment for Wilson's disease, in particular his discovery of penicillamine and later, with **Hal Dixon,** the introduction of trientine as an alternative chelating agent.

After numerous email exchanges between us, fine tuning the visit, **Rhonda Rowland** made her way to Cambridge five months later. Arriving here on Tuesday**, 7 August,** together with her friend and former colleague on CNN in Atlanta, (where Rhonda had been a medical correspondent for over 16 years), I returned from London just in time to share a nightcap with them both. Having compared medical histories and discussed the following day's events, we set off back to their hotel before the light failed, stopping at some famous sites on the way. These included the Cavendish Laboratory where Crick and Watson discovered the structure of DNA in the 1950s and the site not far away, from where Dr Walshe (and his assistant, **Kay Gibbs**) had treated many patients over the following two decades and where also they had purified trientine before posting it off to patients anywhere in the world.

Dr Walshe's former office & lab on the Downing Site in Cambridge

Sitting on the terrace, Dr Walshe is interviewed by Rhonda

I collected Rhonda the following morning to take her to meet him. The interview went very well and Dr Walshe was as articulate as ever. Rhonda had asked in advance if he would be prepared to be videoed, to which he had agreed, and by the time we arrived at his house the production team had already transformed the garden terrace into a film set for the morning. Taking pride of place on the table was the paper that Dr Walshe had published in *The Lancet* in 1956, *Penicillamine, a new oral therapy for Wilson's disease*, which documented the successful treatment of **Shirley Wylie,** the world's first ever patient to be treated with penicillamine. Shirley will be 80 this year and has been taking penicillamine continuously for the past 64 years. Rhonda, having read her story in our 2017 newsletter, had arranged to interview her in London two days later.

After lunch, we and the camera crew made our way back to Cambridge to the home of **James Kinnier Wilson,** James being the son of **Samuel Alexander Kinnier Wilson**, whose seminal work describing our disease appeared in *Brain* in 1912 and to this day holds the record of being the longest medical article ever published in this Journal. Also a nonagenarian, James has followed the Group's fortunes over the past few years and has attended several of our summer meetings. Rhonda was keen to interview him to find out more about his father after whom, of course, Wilson's disease was named.

Rhonda is still editing some of the video footage taken in the UK. Her aim is to reach and educate a wider audience and to this end she plans to submit a short film called *The Story of Wilson's disease* to the Rare Disease Film Festival in the States. Meanwhile, for patients who are interested in her endeavours so far, please visit https://www.facebook.com/RhondaRowlandR2/videos/598110020615574/ where you can see Dr Walshe holding forth on the subject of today's cost of WD meds. The *YouTube* clip has already had over 12k views!

Caroline has been a member of WDSG-UK since the Group was formed in 2000. She is also an active member of our Facebook Group. Living in Edinburgh, she was diagnosed with Wilson's disease in 1980 at the age of 12, having had progressive neurological problems over the previous two years. Wilson's disease has left her with poor concentration, poor motor skills and poor mobility and her speech is also slightly affected. Because of these problems, she has become a bit risk averse! However, she retains a great sense of humour and says that she loves her body and her WD because it takes too much energy to hate it!

She writes, "Last September, I had the adventure of a lifetime. I have been working on the Bronze Level of the Discovery Award, an achievement award for people aged 50 or over, since June last year as a way of exploring new interests. I decided to approach the ***Jubilee Sailing Trust*** (JST) which is based in Portsmouth and apply to sail with them. The JST is an international charity whose aim it is to "promote integration through the challenge of tall ship sailing." It has two vessels, ***The Lord Nelson*** and ***Tenacious*,** both of which have been built to accommodate a crew of 40 of all physical and sensory abilities. They are the only two tall ships in the world that have been specifically built for this purpose.

Aboard ship every crew member has a vital role to play and teamwork is essential. And so on 12 September last year, I arrived in Portsmouth to join *SV Tenacious* on a four day voyage along the English channel and up to the West India Docks in London. I was greeted by this magnificent ship which quite took my breath away.

I knew two other people on board, which was great. On arrival, I was introduced to a "buddy" called Karen who made sure that my individual needs were met. Having settled ourselves in, we then had lots of safety briefings which was quite reassuring! We were also told that there would be a fire drill at some point which would happen without prior warning.

All shipshape & Portsmouth fashion!

There were several tasks assigned to us besides pulling ropes and tying endless knots! These included *Watch Duty*, which as you might expect, involved watching out for other ships and *Mess Duty* which involved setting the tables at mealtimes and washing up some 150 items of crockery and similar quantities of cutlery, afterwards!

SV Tenacious, the biggest Tall Ship in the world - Sept. 18

In addition, there was *Deck Duty* every morning ("Happy Hour") when we came together to help maintain the ship. One of the highlights of the trip was climbing the rigging. Every member had the chance to do this and hoists were provided for wheelchair users so that nobody was left out. For me, it was extremely challenging physically, but when I reached the top, it was the best feeling in the world.

Enjoying a taste of the high life!

The worst part of the entire trip was on day 1 when I went below deck to change into warmer clothes for my *Watch Duty*. I was practically naked when the fire alarm suddenly went off. I have never put my clothes on so fast in all my life! Fortunately, Karen came to my assistance and helped me get dressed and we raced to our muster point, kitting ourselves out with life jackets on the way, just in time for inspection. I was last, but I **was** within time!

Life on board was great and nearly everybody had some disability or other. The shipping in the Channel was challenging, but there was plenty of time to rest or chat between duties. The views were great, and expansive, with spectacular sunrises and sunsets. I will definitely do it again if I can. It left me with an "I can do anything" feeling, something that we, as disabled people, don't often experience."

For more information about the Discovery Award or JST, visit (https://www.discoveryaward.org.uk or www.jst.org.uk)

Ashok Pandit is a 28 year old Wilson's disease patient from Nepal, who has become familiar to us all through his articles in the newsletter. He was the youngest of three children but in 1993, when he was only three, his older sister who was 9 and brother who was 7 both died within a month of one another and nobody knew why. Four years later, Ashok also became ill. He was admitted to hospital and as luck would have it a visiting American doctor saw him and suggested a diagnosis of Wilson's disease. He has been treated with penicillamine ever since, although obtaining his tablets has been challenging from time to time. Yet another misfortune befell the family, when in 2015 their house was badly damaged in the country's devastating earthquake and needed to be rebuilt.

He says, "Greetings to everybody from Nepal. I have been writing for the newsletter since 2012 and am grateful for the platform it has given me to connect with others with Wilson's disease." He continues, "After the quake hit Nepal, we suffered a lot for two years. I am very excited to say that at last we have built my dream home. Everyone said that I would understand my responsibilities if I built my own house. I was confused. Now I have that answer.

The foundation of the house was laid in March 2017. We had hired a contractor to build the house out of modern materials and we were all very happy. I shouldered the responsibility of designing the house, myself, which was exciting. We sold some land and the government gave us money towards its cost. Also, John Ross from Scotland, who helped me get my medicines when I was first diagnosed with WD and still supports me now, helped us out. The total budget of the house came to 29 million Nepali rupees (Nrs **29,000,000)** the equivalent of around **£193,000** in the UK.

Ashok's new house in Kathmandu

I left my job so that I could be there during the building work. I changed the map of the house while it was going up. The contractor thought I was ruining everything and the house would be the worst house ever designed, which made me nervous. Finally, when the house was built it looked so beautiful. Everyone who has seen the house praises its beauty. They say, "Wow; what a wonderful house!" Even the contractor finally told my mum that I was really talented.

So how has the building taught me to understand my responsibility? Well, it has helped me understand the value of wisely spending money on things. You must never be fully dependent on people. You have to discuss things before making any decisions, because listening to other people helps you gather information and make the right decision."

Members' Photo Gallery

Linda and David

Rosie and her niece, Amina

Ashok and his mum

A Date for your Diary 2019-20

Date	Time	Event
Sunday 21 July 2019	**1100 - 1530**	**WDSG-UK Meeting and 9th AGM – Cambridge Rugby Union Football Club (52) Grantchester Road, Cambridge CB3 9ED.**

WILSON'S DISEASE MULTIDISCIPLINARY 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. 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 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.

Royal Surrey County Hospital NHS Foundation Trust & University of Surrey, Guildford

The **Royal Surrey County Hospital** (RSCH) hosts regular multidisciplinary clinics to assess and manage patients with Wilson's disease. The team includes established consultant experts in Wilson's disease with focus on liver (**Prof. Ala**) and movement disorder (**Dr Jan Coeberg**), and has access to international clinical trials and patient registry programmes. Referrals should be addressed to the Centre Clinical Director, *Professor Aftab Ala, Dept. of Gastroenterology and Hepatology, Royal Surrey County Hospital, Egerton Road, Guildford, Surrey GU2 7XX*, email aftabala@nhs.net.

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.*

Children's Clinic at King's College Hospital, London

There is a 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) and a psychologist. The clinic is directed at patients who are complex with both liver and neurological involvement and referrals should be made via the Paediatric Liver Centre at King's or enquiries sent to **Kathleen Meader, (**kathleenmeader@nhs.net), PA to Prof Dhawan.

IN MEMORIAM

It was with particular sadness that we learnt of the passing of **Keith Pereira** on 27 March 2018, aged 70. Keith and his wife, Linda, had been members of WDSG-UK since its inception in 2000 and had, over the years, attended many of our patient meetings. A former patient of Dr John Walshe, Keith was a well-liked, vivacious and highly respected member of the Group and will be very much missed by us all.
His family very kindly held a collection in his memory and raised over **£700**. We offer our sincere condolences to them all and thank them for thinking of us at this time.

Wilson's Disease Support Group – UK

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Dr Godfrey Gillett Group Adviser, Honorary Member
Linda Hart: Group Co-Founder, Adviser
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