CJD SUPPORT NETWORK

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Newsletter

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CREUTZFELDT-JAKOB DISEASE SUPPORT NETWORK NEWSLETTER

Managing risk of transmission of vCJD by blood transfusion

Marc Turner – Professor of Cellular Therapy, University of Edinburgh and Associate Medical Director, Scottish National Blood Transfusion Service

Each year in the UK around 2.2 million blood transfusions are given. Of these, the majority are of red blood cells which carry oxygen and are used for patients with anaemia or major haemorrhage. Around 200,000 transfusions of platelets are also given to prevent bleeding for example in patients receiving treatment for leukaemia, and plasma to provide clotting factors for example in the context of major trauma. Plasma from many donors is also pooled in order to manufacture plasma products, these are pharmaceuticals such as Factor VIII and Factor IX used for the treatment of haemophilia, immunoglobulin for patients with immune deficiency, and albumin for emergency resuscitation.

Since the first description of variant Creutzfeldt Jacob Disease (variant CJD) in 1996, UK and International Blood Services have been concerned about the possibility of transmission of infection by blood transfusion, tissue or organ transplantation, for two main reasons: first, that variant CJD was recognised to be a new strain of a prion disease and that it should not be assumed that it could not be peripherally transmitted and second, that there was evidence of

accumulation of abnormal prion protein in peripheral lymphoid tissues.

A number of precautionary measures were taken including changes in the guidelines on who may donate blood and tissues, implementation of universal leucodepletion (i.e. the removal of white blood cells from blood components by filtration) and the importation of plasma for plasma product manufacture.

Since 2004, five transmissions of variant CJD prions have been described. In three cases the patients received red blood cells (non-leukodepleted) and went on to develop variant-CJD themselves. In a fourth case an elderly lady died of un-related causes and was found to show evidence of accumulation of prions in her spleen and one lymph node. A fifth,

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Fundraising news

Many people take part in events both big and small to raise money for the CJD Support Network. If you are considering taking part in, or organising a fundraising event, please contact the Network on 01630673993 to discuss how best to organise your fundraising.

We are very grateful to all the fundraisers and sponsors in 2009 who donated a massive £16,000 to the CJD Support Network. This money will help to fund our 24 hour helpline and ensure that we can continue to offer emotional and practical support to families affected by all strains of CJD.

In 2009 we had a sponsored runner in the **London Marathon** and a team from the National Prion Unit in the **British 10k London Run**. We are now recruiting for the **2010 British 10K London Run** in July. If you would like to take part please contact the Network.

Many fundraisers take part in, or organise smaller activities (such as **coffee mornings**) and all the money raised is most gratefully received by the Network and used to support patients, carers and families.

Many of our fundraisers find it helpful to set up a web page for on line donations at www.justgiving.com/cjdsupport. In 2009 sponsors donated £11,585.92 to the CJD Support Network through **Just Giving**. If you would like to set up a page and would like our help please telephone on 01630673993.

How to hold a coffee morning

Sarah Tomkins has kindly shared some tips for planning a coffee morning. Sarah says:

- Send out your invitations 3-4 weeks in advance – ask as many friends and neighbours as you can.
- Time suggested 10.30am – 12.30pm – expect people to come and go all morning.
- A bring and buy stall and a raffle helps the funds, a cake stall is always popular.
- Admission is usually £2 or £3 – this includes coffee and scones or biscuits or cake.
- Helpers needed, one on the door, one for the raffle, one or two for the stalls, two to make and serve the coffee and wash up!
- Preparation, have cups, saucers, plates, serviettes, sugar all ready before hand. Have plenty of change and pots for money.

Good luck to everybody who decides to have a go.

Success in the Berlin Marathon

Noeleen and Sinead Boyle with their medals after the Berlin Marathon, September 2009

Noeleen and Sinead Boyle entered the 2009 Berlin Marathon to raise money in memory of their father Hugh Boyle.

Hugh died of Sporadic CJD in Belfast in July 2009. Noeleen and Sinead with the help of their family raised an amazing £12,000. Gillian Turner from the CJD Support Network was asked to attend a cheque presentation event at their home in Amagh in December. Over £7,000 was given to the network, £3,000 to the Marie Curie nurses who helped the family care for Hugh. Their GP, Dr Richard Dorman was presented with a syringe driver for the community clinic. Lucy, Hugh's wife, said that this piece of equipment had been invaluable when caring for Hugh at home.

We would like to thank the Boyle family for this very generous donation, which will all be used in our work supporting families affected by CJD.

Sinead Boyle presenting Gillian Turner with a cheque for £7,000

Fundraising in 2010

Calling all golfers

Colin Robinson died of Sporadic CJD in 2009 aged 51. See his story on page 6. He was a keen golfer, as are his family and many of his friends, and so the family felt it was appropriate to remember Colin by playing the game that he loved.

The Colin Robinson Memorial Golf Tournament will both remember Colin and raise money for the CJD Support Network. It will be held on Friday 4th June 2010 at Warrington Golf Club.

An invitation for golfers and a letter to any business who would like to sponsor a prize can be found on our website at www.cjdsupport.net or by contacting Colin's brother in law, tim.gibbons2@btinternet.com or the CJD Support Network office on 01630 673 993.

Calling all runners

The CJD Support Network has six places in the British London Charity 10K run on July 11 starting at 9.30am. This year is the 10th Anniversary of the British London Charity 10K run and we are looking for runners to run for the Network.

Three runners have already been signed up and have started training. If you would like to join them please contact us on 01630 673 993.

Kite flying

A kite flying family fun day is being held by members of the Great Ouse Kite Flyers in Hertfordshire on Sunday 25 April to raise money for the CJD Support Network. Organiser Stephen Hodges says everybody is welcome who would like to take part and fly a kite. The event is called The Chocolate Flyin and it has been held for the past three years. Stephen can be contacted at stephenhodges_jb52@yahoo.com

A family story

John Shelley died in January. His family says of him, 'Our dad was a hardworking, generous and thoughtful man who was loved and respected by all who knew him. Dad always made sure us three children had everything we needed and took us on many great holidays.'

John's story, as told by Angie, Mindy and his partner Donna, can be read on the CJD Support Network website www.cjdsupport.net. It details the events which led to the eventual diagnosis of sporadic CJD and the devastating effect it had on his family.

In memory

Heartfelt thanks to the friends and families of those below for the donations received in 2009 in their memory:

Lynda Lewis
William (Bill) Smitherd
Catherine Whitbourne
Roy William Curry
Peter Allan
Maureen Hill
John Mellor
Nigel Robinson
Jean Godber
Carmelina Tomaso
Peter Nelson Callow
Rita Hamblyn
Colin Robinson
Oliver Gerwyn Lloyd
Hugh Boyle
Linda Fanning
Linda Young

Recent CJD figures

The number of deaths of definite and probable cases in the UK, up to 1 March 2010. Source: the CJD Surveillance Unit in Edinburgh

Year	Sporadic	Iatrogenic	Familial	GSS	vCJD	**Total**
2005	66	4	8	5	5	**88**
2006	69	1	6	3	5	**84**
2007	63	2	8	1	5	**79**
2008	87	5	2	3	1	**98**
2009	70	2	3	5	3	**83**
2010 •	11	0	0	0	1	**12**

• As at 1 March 2010

Total of definite or probable vCJD cases (dead and alive) in UK 172

CJD in the news

The Daily Telegraph reported in Scotland on the 19 December that Grant Goodwin age 30, who had died of vCJD, had belonged to a genetic group that had not previously shown signs of vCJD infection. This is clearly a matter of concern as one possible explanation is that vCJD has a longer incubation period in this genetic group and therefore more cases could be seen in the future. Our sincere condolences go out to Grant's family.

800 hemophiliacs given tainted blood are at risk of vCJD

It was reported in *The Independent* in May 2009 that Lord Darzi, a Health Minister, revealed in a parliamentary answer that more than 800 people with hemophilia have received contaminated blood products putting them at heightened risk of developing vCJD.

Lord Darzie gave these figures following a death of a hemophiliac who had received infected blood products. A post mortem showed that although the person died of an unrelated cause there were traces of vCJD in the spleen.

Chris James, chief executive of the Hemophilia Society said that this death shows that there is a real possibility of a link between receiving blood products and developing CJD. What was a theoretical risk is now a suspected causal link.

Does vCJD still pose a major public health threat?

The Daily Telegraph on 8 February reported on a case of eight men and women who had lost loved ones to vCJD who took their case to the High Court arguing that the delay in changes to the Government's compensation scheme in relation to psychiatric injury and particular hardship was 'irrational, perverse and unfair'.

They argue that it is also unfair that the proposed reforms are not retrospective and will not apply to cases where diagnosis was made before March 31st 2010 cut-off date.

The article told the story of Judy Kenny. Judy's husband Deryck, who was the first person to die in the UK from a blood transfusion. Judy, a committee member of the CJD Support Network, said that she had sympathy for those in Court last week. Judy was not told before Deryck's death about the contaminated blood transfusion. She said that she did receive some compensation, and she was grateful, but it would have been more useful while Deryck was alive so she could have given up work to nurse him at home.

An American view

Florence Kranitz, President of the American CJD Foundation, and Deana Simpson, founder of CJD Insight in America, have written an excellent article 'Using Non-Pharmacological Approaches for CJD Patient and Family Support as Provided by the CJD Foundation and CJD Insight' in the journal *CNS & Neurological Disorders*. Florence's husband died of Sporadic CJD and Deana's family is affected by Genetic CJD. The article can be read on our website www.cjdsupport.net by clicking on Carers' views on the left hand menu. If you do not have access to the internet and you would like a hard copy of the article please telephone the CJD Support Network office on 01630 673 993.

Network news

Annual General Meeting

Over the weekend of the 14 and 15 November 2009 the CJD Support Network held a very successful **AGM and family support meeting** for members at the Burlington Hotel, Birmingham. We also held a planning meeting for the members of the network committee to plan the network's work for 2010.

We are very grateful to a family member, who in 2009 kindly sponsored the family support meeting in 2009 and another similar meeting in 2010, The family support meeting is a very important day in the work of the CJD Support Network as we are able to meet and support families and share their experience in an informal and enjoyable way.

48 members attended the family support meeting and had the opportunity to talk and share experiences with families who had been affected by all strains of CJD. They also had access to key professionals working in CJD who answered those previously unasked questions.

Although we had a few tears, we had many kind words from members who said they had both enjoyed the day and that they had found it very helpful to meet and share experiences with other families.

A letter received from a member who attended the family support meeting said "Thank you and all the committee for the very warm welcome and kindness shown on the 14th November. Obviously such a meeting requires much time and thought. I am quite sure it was appreciated by all who were there."

The network continues to support families who are affected by CJD.

Between July and November 2009 we were contacted by 24 new families for information, advice and emotional support. Our 24 hour helpline received 600 calls in 2009 from families affected by all strains of CJD and the website received 58,935 unique visitors accessing our information.

In the current financial climate, we, like so many other charities and organisations, are worried about our future. Our international colleagues all tell us that funding for CJD is getting more and more difficult to acquire.

At our 2008 AGM we reported that we had lost our Department of Health Section 64 grant. However, we were very pleased to report this year that we had been successful in obtaining funding of £30,000 from the National CJD Surveillance Unit which is funded by the Department of Health and a fantastic £25,000 raised by sponsored events, in memoriam and other donations from our members. This has cemented our financial position this year and we have our fingers crossed for next year.

We would like to thank all our sponsors and the many families who have walked, cycled, knitted and a variety of other things on our behalf and even forgone birthday presents in exchange for donations to the network.

We would also like to thank you for responding to our request for a membership fee. 2009 was the first year we had asked for a fee and the money raised from this has helped us to fund the newsletter and postage. Thank you.

Please may I give you a gentle reminder that if you have not signed a bankers order or already paid, your 2010 membership fee of £10 is due in March.

Diary date

Next CJD Support Network family support meeting

The date of our next family support meeting is **Saturday 20 November 2010**. At the moment the venue has not been decided, but if you can suggest a location which is easy to get to, I would be grateful if you could let me know.

When we pick a venue we have to consider one that is easy to get to for a majority of people on our membership list. It must therefore be near a railway station and should be affordable. In the past we have held our family support meetings in Liverpool, Birmingham Milton Keynes and London. Up to-date the Birmingham venue has attracted the most families.

Contact me, Gillian Turner, at the address on the back page.

Around the world

American CJD Foundation conference

The network was represented at the American CJD Foundation conference in July 2009 by Prof. Richard Knight. This conference is jointly organised by the foundation and the National Prion Disease Pathology Surveillance Center and PrioNet Canada,

Richard gave a short presentation on our behalf on how the network works alongside the National CJD Surveillance Unit and National Prion Clinic in the UK supporting families affected by all strains of CJD.

The conference provided a combination of family meetings, scientific presentations, and updates from directors of Prion Disease Surveillance Centers around the world and reports from the CJD International Alliance of patient support groups.

NeuroPrion 2009

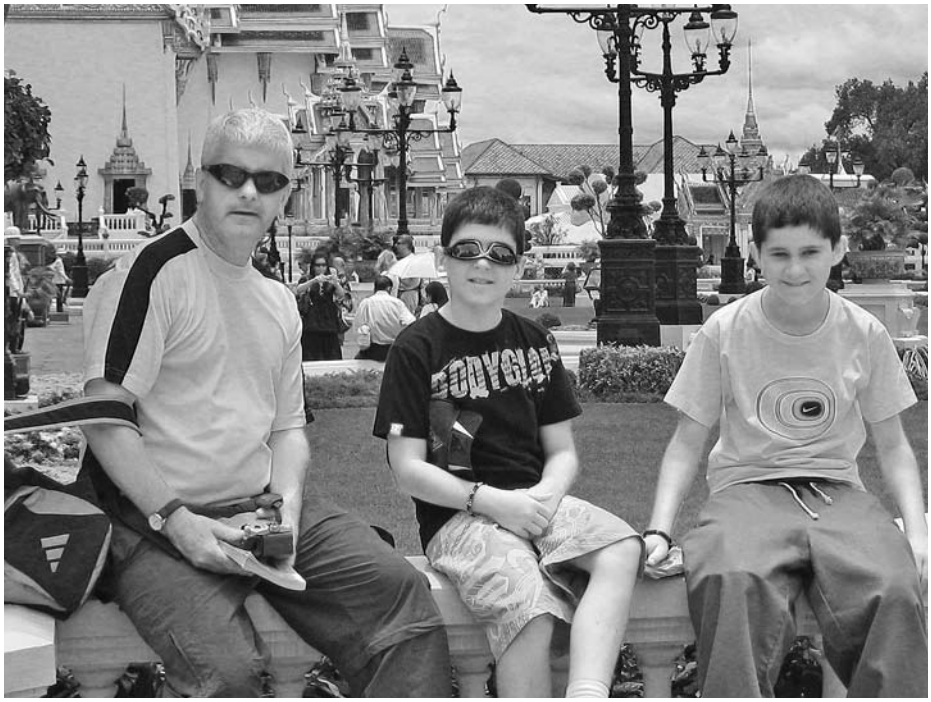
A Scientific conference, NeuroPrion 2009 was held at Thessaloniki in Greece in September. Gillian Turner attended on behalf of the network and was joined by over 800 researchers from all over the world who are working on prions and related diseases and patient support groups from America, Australia, Italy, France and Israel.

Over the four days of the conference delegates listened to presentations from researchers from around the world in the areas of functions & Cell Biology of PrP, Diagnostic, Therapeutics & Decontamination, basic Mechanisms of neurodegeneration & Pathology, Enigmatic N2 Functions & Cell Biology of PrP. Details of some of the talks can be found by visiting www.neuroprion.org.

Gillian said that she was encouraged that there was such a breadth and depth of research being undertaken by so many committed researchers. Gillian was grateful to Alliance BioSecure who sponsored her attendance at the conference and to East Willow Construction Ltd of Middlesex for sponsoring her travel to Greece.

Colin's story

by his wife Dr Ann Robinson



Colin with his children on holiday in Thailand

When I realized in early May 2009 that it was very likely that my husband Colin, was about to be diagnosed with CJD, my first thoughts were how to explain this to our children, Paul aged 14 years and David aged 12 years. Although there was plenty of information on the website and from the support network, none was written with children in mind. Through Colin's story we would like to share with you how the children managed throughout and after his illness.

It had been shortly after Christmas 2008 that Colin's memory started to fail. He would forget the simplest of things. He had worked hard for a professional exam between September and December, continually complaining that he could not remember the facts – 'old age' I presumed.

Tense atmosphere

After Christmas he appeared quite withdrawn and irritable. Conversations around the dinner table were monosyllabic. Focusing on 'the here and now'. There was no interest in planning our annual summer holiday or any short breaks. Paul and David became increasingly frustrated with their dad – he would forget any task they asked of him and then arguments would ensue as he denied ever being asked. The atmosphere was tense. They questioned me continually about their dad's memory. As a doctor myself, I was very concerned and absolutely convinced he had early onset dementia. I knew this would be devastating for the family and realised I needed to talk to Colin to agree a way forward. It was too difficult as he was edgy all the time – with me and the children. There never seemed to be a moment when he was relaxed and we could converse.

We were due to embark upon out annual skiing trip to France with some friends on 15th February – the day before I had given Colin his valentine gift – he forgot to give me mine. It was embarrassing telling the children when they asked why he hadn't bought me a present. I prayed no-one would ask me the next day and kept my head down at the airport when the conversation drifted towards Valentine's gifts.

Could not be left alone

Whilst we were on holiday it became even more obvious to myself and the children how severe Colin's symptoms were. He had no patience and became very irate with David over something so minor and grabbed him by the 'scruff of the neck.' He forgot his way back to the ski chalet and I soon realised he could not be left alone. He frequently forgot what he'd ordered from the menu and I always had to make sure I had noticed. He checked his skis, boots, and rucksack time after time, in an obsessive way.

On arriving home, I confronted him. We needed to see someone about his failing memory. He burst into tears, apologizing profusely – afraid he was letting us all down. His frustration was obvious.

Initial diagnosis

There was a speedy occupation health referral, followed by a psychiatric referral. Unfortunately Colin could not operate his mobile phone and therefore did not pick up the telephone message about the outpatients arrangements. Knowing the hospital system, I was quickly able to track down and speak to the consultant. He arranged to visit Colin at home and diagnosed severe depression. He was commenced on antidepressants.

The children wanted to see an immediate improvement. I tried to explain that the medication usually took at least three weeks

before showing any effects. At the back of my mind, I still questioned whether his severe memory loss was characteristic of depression. He had forgotten to renew the car tax and insurance and very soon had to stop driving. Sausages became his favourite snack – he would cook them at any time of the day or evening – it was a joke amongst family and friends. Paul and David were finding that they were taking on more and more responsibilities at home whilst I was at work. Roles were reversed – Paul looked after the cooking as Colin wanted to prepare bacon and sausages for every meal!

Two friends, our GP and his psychiatrist wife, were concerned that there may have been an organic reason for Colin's illness and pushed us to agree to a neurology referral. This was arranged around the middle of April and by this time Colin had started to trip slightly whilst walking. Paul was due to go on a rugby tour with school and couldn't wait. I remember saying how much he was looking forward to a rest – the responsibility was hard for him.

David and I took Colin to London for a few days whilst Paul was away but we could not leave him alone. We were watching a performance of 'Oliver' at the Drury Lane Theatre. I was a few rows behind Colin and David and Colin turned around, saw me and then commented to David that 'his nanna' was sitting at the back of the theatre.

The boys were truly perplexed by his actions – his continual checks for his keys and his wallet when he went out. He was stumbling more and more by now and we were anxiously waiting for the results of his MRI scan, EEG and lumbar puncture. Paul at this time was studying hard for his GCSE ICT exam and I was aware, having had preliminary discussions about prion disease with Colin's consultant neurologist, that we might be expecting a diagnosis of CJD. The boys continually asked me what was going to happen and whether or not their dad would get better.

CJD confirmed

On 22 May 2009, we received the confirmatory diagnosis that Colin had CJD – the SSP40 protein was positive in his CSF with a very abnormal EEG and MRI scan. By that stage Colin was having periods of time when he was totally unaware of what was going on, other than performing his activities of daily living. There was no purpose to his life, he had stopped working six weeks before and could no longer drive, operate his mobile phone, or use money. He frequently went for walks and on several occasions we couldn't locate him. Fortunately, on most occasions, one of our local support network of friends who lived close by, usually found him and brought him home. One time he dressed himself in his 'best' navy blue suit with white trainers. What a sight!

On the day we went to outpatients to receive his results, he was perfectly lucid. The neurologist informed us he had CJD and Colin obviously wanted to know what that meant with regards to his future life. Between myself and the neurologist, we explained that he was likely to die in the next three to six months and that he would be totally physically dependant.

The full truth

We sat in a private room for an hour or so – crying – Colin understood perfectly and we discussed how we would tell the children. He kissed and hugged me – something he hadn't done for months and months. We drove home in time for the boys returning from school – about to finish for their half term holiday. We agreed that they must know the full truth – so we sat with them on the sofa and related what we knew. We told them that Colin was dying and that it was likely to happen in the next six to twelve months. There were a lot of tears shed that night. Paul had lots of questions and soon went and lay down on his bed with his door shut, and cried. I had some of the Support Network newsletters so he was able to read some of the stories which he found helpful. David had a few questions and then went off to watch television to distract himself from the heartbreaking news. Within a couple of hours, Colin was oblivious to his diagnosis – so frustrating.

Adapting to a wheelchair

Soon after, we had visits from the Surveillance unit in Edinburgh and the Prion Clinic in London. I kept the children fully up to date. By this stage, it was obvious Colin needed a wheelchair. Their reactions to its arrival were miles apart. David, enquiring and mischievous, was already propelling himself in it around the house. Paul felt ashamed his dad needed the wheelchair and didn't want me to bring him to school events in the chair. David had told all his friends about his dad, CJD and the fact he was about to die in the next six to twelve months. They both became adept at dismantling Colin's chair and storing it in the boot of the car.

The house needed adapting with ramps and rails. Paul felt so very conscious about the huge ramp to the back door – David was immediately skateboarding up and down it.

The boys help with care

July brought the summer holidays – no prospect of a holiday away but initially lots of days out with

family and friends. We had the use of a wheelchair accessible vehicle which made life so much easier. Paul lost his awkwardness as his friends and their parents became totally engaged in helping with Colin's care. David tried out the hoist and regularly he assisted in transferring his dad from bed to chair. Both boys became expert at feeding him and David loved to make him laugh. They both knew all the community nurses, who by this stage were coming in twice daily to assist with Colin's care. The holidays were fine, going well until Colin developed a sore at the bottom of his spine. Suddenly we were restricted to the house until we had a suitable wheelchair for him. It seemed to coincide with most of our friends' holidays – the boys were getting a little frustrated, it had been a long summer holiday at home.

Difficulties with eating

We celebrated our 20th wedding anniversary with lunch at 'What's Cooking?' at the Albert Dock in Liverpool, followed by a trip to the bug museum. So much for the wonderful dreams I'd had a year earlier of a weekend away in Paris. Colin's eating was becoming more difficult – he was only taking in small amounts of very soft food and each meal would take the best part of an hour.

By this time he was sleeping downstairs on a hospital bed with the highest pressure-relieving mattress available. As his eating became more difficult I realised that he was probably nearing the end of his life. The children were back at school – a new academic year. It was important they knew he was deteriorating and we talked about what should happen if he became very ill whilst they were at school. Both Paul and David wanted to be called and brought home. They wanted me to stay with Colin whilst someone else came to collect them. They also didn't want to be with him when he died, but they were happy to be in the house.

Within a week or so, he had completely stopped swallowing and was started on a syringe driver and kept comfortable in bed. Friends and family came to say goodbye. The boys came and told him their news from school. 'How long do you think he's got?', they wanted to know. It was impossible to say, I told them on Wednesday evening. If he deteriorates you won't go to school tomorrow.

Colin died at 5.30am the following morning. I was by his side and the boys asleep in bed upstairs. I woke them gently and told them that their dad had died. They each in turn came to hold his hand and say goodbye. We had discussed what would happen after that, so there were no surprises when the undertakers arrived.

We had a beautiful day, the day Colin died – 24 September 2009. Friends and family visited and we all reflected on what a peaceful death it had been at home – the most perfect you could wish for and the one he had desired most.

The funeral

Paul and David took the next few days off school. We planned Colin's funeral carefully and chose the hymns and readings together. We sorted out the venue for the reception afterwards, the boys choosing the menu. They were both adamant they wanted to wear suits so we enjoyed a mammoth shopping spree while other children were at school. Paul was conservative in his choice – a grey pinstripe suit with a pale blue shirt and tie. David was his usual flamboyant self – a bright red and grey check shirt with a grey tie and black suit.

We visited the cemetery at Fox Covert where he was going to be buried so they understood what was involved. Their school was most supportive. They were each allowed to choose five close friends who came with the staff in the school minibus. Their friends helped to give out the orders of service and taking the collection afterwards for the CJD Support Network.

Thank goodness for Facebook! – I never thought I would ever hear myself say that. The boys each had at least fifty friends send their thoughts and best wishes. Paul and David thanked each one in turn – I was so proud of them for that. Paul even posted a photograph onto Facebook of the three of us on the morning of Colin's funeral with the accompanying words 'a sad morning but a great party afterwards.'

Their father's gifts

I reminded each of them in turn during my eulogy of the gifts their dad had given them. They had both inherited his sharp mathematical brain and sporting prowess. Paul has a sharp eye for a bargain and searches out the best deal, continually reminding us of Colin's thrifty ways. David has Colin's sense of humour and mischievous grin – his one-liners frequently catching us all.

As we celebrated Colin's life on that day, I felt happy that we had prepared the boys as fully as we could for his death. We miss him lots and talk about him often, sharing our happy memories. We are looking forward to his memorial golf day in June – let's hope the sun shines!

The Colin Robinson Memorial Golf Tournament takes place on Friday 4 June 2010 at Warrington Golf Club. *See page 2 for details.*

A day in the life of…

Liz Ford, a clinical nurse specialist

Tuesday

As a Clinical Nurse Specialist at the National Prion Clinic I find no two days are the same. Everyday is a journey into the lives of people who are affected by the devastating changes that prion disease has on their lives. It is a privilege for me to be taught by patients about their unique needs and share with them the challenges that they and their families individually experience every day. The week usually involves being out and about around the country in hospitals, hospices and the homes of our patients. However on a Tuesday the whole team gets together for a review of the weeks work and to plan for the next week, let me take you through a typical Tuesday.

I arrive at our offices in the Institute of Neurology at 08:30 having cycled in from home. Working in the National Prion Clinic involves a lot of travel which means that all staff need to stay fit (although biscuits and cakes do make a regular appearance in the office!). Every Tuesday we have our Multidisciplinary Team meeting where we discuss all the patients we have seen over the last week and planned visits for the week ahead, as well as new referrals and any patients we have particular concerns about. It is an opportunity for us to relay feedback from patients, relatives and local healthcare professionals, discuss symptom cause and management, diagnostic investigations, and medical, social and nursing care issues. These discussions help us to ensure that all options for medication and care are assessed, and the appropriate advice and support is given back to carers and professionals, ensuring best practice and evidence based quality care.

We also use the meeting to formally review quality standards through audit presentations, review journals and current literature, learn more about the research projects being run by the Clinical Research Fellows and staff in the MRC Prion Unit, and as a time for teaching and staff training.

We all leave these meetings with plenty to do so the next hour or so involves phone calls and emails to carers, patients and relatives to feedback outcomes of meeting discussions, arrange visits and cohort investigation days, or sometimes just for a quick catch up. As the clinic works nationally it is not possible to see people face to face as much as we would like, so we have to rely on the telephone or an email with detailed information for local teams to ensure people get as much support and information as they need.

Next up is a trip to The National to review a lady affected by Inherited Prion Disease who is currently an inpatient. As an NHS service we have the ability to admit our patients to The National if our assistance is needed with diagnosis, specialist opinion or treatment, and can refer people on to other specialist services within University College NHS Foundation Trust as required. The medical review is followed by a meeting with other professionals from The National, such as the physiotherapist who has been working with our patient, to review her progress and to start planning her discharge home.

Then its back to the office for a team meeting about the National Prion Monitoring Cohort, the observational study we are currently running to learn all we can about all forms of prion disease. Our aims are to establish a greater understanding of disease progression, so we can better inform and support patients, families and carers, and have an idea of who will benefit most from treatments once we start a formal clinical trial. So far 133 people have been enrolled into the study and virtually all eligible new referrals we see have opted to take part. We are also recruiting control participants and if you would like to know more about what this involves please contact the clinic.

Once the meeting is over it's time to prepare for the next days trips. We have three mobile doctor and nurse teams which enables us to respond quickly to new referrals or when there is a change in a patient's condition, and keep a regular follow up schedule with patients. Nursing staff also attend case conferences, funding meetings and provide training and support sessions for carers and families. I'm off on a new referral visit so its my responsibility to get together all the necessary equipment for safely taking blood samples, while the doctor packs assessment forms and a neurological and cognitive testing kit. We offer all new referrals the option having a blood sample taken to test for inherited forms of prion disease, and at the same time we ask to take a few extra samples for the CJD Surveillance Unit in Edinburgh, and for the MRC Prion Unit's research programme. This is entirely voluntary and formal written consent by the patient or their next of kin is always taken. Blood samples are extremely valuable as we aim to develop a simple blood test for diagnosing variant and sporadic CJD, thus removing the need for weeks of investigations and hospital admissions that some patients go through to reach a diagnosis. I also pack my BlackBerry and laptop so I can get plenty of work done on the long train journey and stay in touch with the team.

The day finishes with a nursing meeting to run through our planned trips for the next week. Often Tuesdays are the only days we are all in the office together so it's a chance for us to coordinate our workload, catch up about our cases and plan cohort visits. All of a sudden its 6pm so its time to get back on my bicycle and off home for the evening.

Managing risk of transmission of vCJD by blood transfusion

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plasma related transmission has recently been identified in a patient with haemophilia who also died of other causes and was found to have evidence of abnormal prion protein accumulation in his spleen.

One of the difficulties in assessing the current risk of transmission of variant CJD by blood and other tissues in the UK is the uncertainty around three key issues: prevalence, infectivity and susceptibility.

Prevalence

It is not the incidence of clinical variant CJD which is the main determinant of risk with regard to blood and tissue transmission, but the prevalence of sub-clinical variant CJD (that is people who are infected with variant CJD prions but do not have any symptoms or signs of disease) amongst the healthy population. This is of course essentially unknown.

The most recent position statement from the Spongiform Encephalopathy Advisory Committee (SEAC) estimates the prevalence at around 1 in 4000 with a range in the order of 1 in 1,500 to 1 in 20,000, based on the study of Hilton et al which showed three positive samples in appendices from a study of just over 12,500 individuals.

The National Anonymised Tonsil Archive study has thus far shown no positive samples from over 70,000 patients studied, but SEAC do not feel it appropriate to amalgamate these two estimates because of differences in methodology between the studies.

It seems unlikely that a more accurate prevalence estimate will be achieved until or unless a blood test can be developed for large scale population studies.

Infectivity

The level of infection present in the peripheral blood of individuals with subclinical variant CJD infection is unknown as is its distribution both spatially (i.e. between different blood components) and temporly (i.e. over time during the incubation period). This is because whilst the infection is clearly transmissible by blood it cannot be detected either by biochemical tests or infectivity studies in animals at present.

The data we therefore have available is from animal studies in hamsters and in mice. In aggregate, they suggest that there might be around 10 infectious doses (ID) per ml in blood (range: 1 ID to 300 ID), being sufficient by definition to infect another member of the same species. This would mean that in a typical 450 ml human blood donation there would be around 4,500 infectious doses.

In addition the animal data suggest that infectivity is mainly associated with the white blood cells and the plasma rather than with the red blood cells or platelets themselves. However blood components as transfused contain elements of other cell populations and plasma. The applicability of these rodent data to the human setting of course remains uncertain.

Susceptibility

This is the extent to which a person exposed to prion infection (for example through blood transfusion) will themselves become infected and thereafter their risk of developing clinical disease. The studies carried out in sheep infected with scrapie or BSE show that 40-50% of recipients are infected through blood taken either during clinical phase of disease or in the latter half of the incubation period. The extent to which these data can be extrapolated to the human setting is uncertain. An assumption has been made that somewhere between 10% and 100% of people exposed to infection are at risk of being infected and of developing clinical disease. Again, only time will tell whether this assumption is correct or not since most UK adults will have been exposed to infection through the blood chain to a greater or lesser extent.

Risk management

In the face of these levels of uncertainty the Advisory Committee on the Safety of Blood Tissues and Organs (SABTO) which is charged with advising Ministers and Departments of Health on all aspects of blood, tissue and organ safety has chosen to continue to investigate further

precautionary measures. Two potential measures in particular have drawn interest: blood tests and prion filtration.

Blood tests

The first is the possibility of development of prion tests. The challenge is very significant because unlike 'normal' microbiological agents such as bacteria and viruses there is no DNA associated with transmission of the prion agent and also no immune response. Therefore the standard serological (antibody based) and DNA based screening tests used for agents such as HIV, Hepatitis B and C are not applicable in the context of prion diseases. To date, there is no blood test that can detect CJD in the blood.

Many research groups and companies have worked on the development of potential screening tests but unfortunately most of these have now dropped out, either because they were unable to achieve the required levels of sensitivity or because of commercial considerations.

The sensitivity required of such a test (i.e. its ability to detect PrP^{TSE} in the blood of people who are infected) is estimated to be in the order of 0.1–0.01 picograms (that is 10-100 million millionths of a gram) per ml of PrP^{TSE}. There is also the challenge of specificity (i.e. the ability of the test not to give rise to false positive tests in people who are not infected with the disease). When one considers that the UK Blood Services test between 2 and 3 million individuals per year a test with a specificity of 99% would give rise to 20-30,000 false positive tests per annum. Unfortunately these people would have to be told that they have tested positive.

Finally there is the problem of validating a test. Normally one would validate a test on large numbers (several thousands) of people who are known to be infected with the infection in question. With variant CJD this is not possible because there are relatively speaking, a smaller number of clinically affected people and also because it is difficult to take large amounts of blood from such patients. Therefore most of the test validations have to be carried out on infected brain homogenates spiked into blood and also on blood from infected animals raising again the issues of applicability of such models to the human setting.

Currently there are three tests under evaluation. Two of these are commercial tests and because of confidentiality relating to the companies themselves it would not be appropriate to share the current data, although one company at least does produce regular press releases. UK Blood Services are also working on the application of a test called Protein Misfolding Cyclic Amplification (PMCA) to blood, which is thought to be extremely sensitive but does have problems with specificity and is very labour intensive. It could not therefore be used as a general blood screening test (because of the large number of tests required in a very short timeframe) but it could be used a confirmatory assay or be offered to those people who wish to be tested for other reasons.

Prion filtration

There is considerable interest in the possibility of prion filtration and at least two companies (MacoPharma / PRDT and Pall Medical) have been working on a development of prion filters. These effectively work by an affinity ligand on the filter which binds normal and abnormal prion proteins.

One filter has been under independent assessment both in terms of its effectiveness in prion removal and in terms of potential negative impact on red blood cells. In its October meeting, as an initial step, SaBTO decided to recommend implementation of this form of prion filtration for blood transfusions given to children born since 1996.

SaBTO have also made recommendations about moving forward with platelets donated by a single donor through a process called apheresis (most platelets are derived from the blood donations of 4 donors), and importation of plasma for all patients. These recommendations have now gone to Ministers and Departments of Health for further consideration.

In conclusion

Whilst there remains uncertainty over the risk of transmission of variant CJD by blood and tissues it is important to continue to work closely with research groups and manufacturers on the development of new technologies whilst continuing to support further research to clarify the extent of the continuing risk. ■

How to decide if a blood test will work

Phillip Minor, Head of Virology and Deputy Director
of the National institute for Biological Standards and Control

For a number of reasons there is an urgent need for a blood test for variant Creutzfeldt-Jakob disease (vCJD)

Perhaps the most important reason is that a test will help people with symptoms to know if their disease is due to vCJD. A test may also help those who have been told they may be infected, for example through blood transfusions, to know whether they are actually infected or not. If, as we all hope, potential treatments are developed, a blood test might be used to help to see if they work. A reliable test could also help prevent new infections occurring as a result of blood transfusion.

Most blood tests are developed to diagnose diseases that develop fairly quickly, happen fairly often and are caused by known bacteria or viruses that can be easily studied. vCJD develops slowly, is thankfully rare and is caused by an agent whose exact nature is still controversial, which makes the development of a test problematic.

As part of the development of a new test it would first be tried out by the individual or company in their own laboratories, if successful they then move on to national authorities or organisations such as the Blood Transfusion Service for independent testing.

There are very few samples of any kind from people who have had vCJD, and because of the very small number of new cases there is very little opportunity to collect more. As there is a lot of demand for samples from companies, it is essential that precious samples are used for the most important purposes. This is decided by a panel of experts.

The CJD Resource Centre was set up by the Department of Health to support the evaluation of possible blood tests in such a way that precious specimens were used in the most important part of the test development, namely the final evaluation of a test. False results could have very serious consequences both for individuals and the blood transfusion service. It is crucial to the evaluation of a test that the developers do not know which samples are from patients and which from healthy individuals. The CJD Resource Centre can prepare samples independently so that the companies are 'blind' to which samples came from patients.

Before a developer is provided with a sample from a person with vCJD, they are required to provide evidence that the test is likely to work, for example, by examining blood from animals infected with prions, or showing they can detect tiny amounts of CJD brain tissue mixed with blood. After this, developers may be asked to show that their test doesn't give a positive result in more than a small proportion of the healthy population. If it did, we would know that these were likely to be false results and the test would not work.

If a test is shown to work well, we might then be in a position to test vCJD samples. These would be hidden in a large number of healthy samples, The developer would have to identify which came from patients and which didn't. Only one company has got to this stage of the evaluation process, however, at the final stage they were unable to correctly identify the vCJD patients.

Although there is no blood test for vCJD yet, companies and academic groups are working extremely hard on this problem, and we hope to see positive developments soon.

Magnetic resonance imaging in Creutzfeldt-Jakob disease

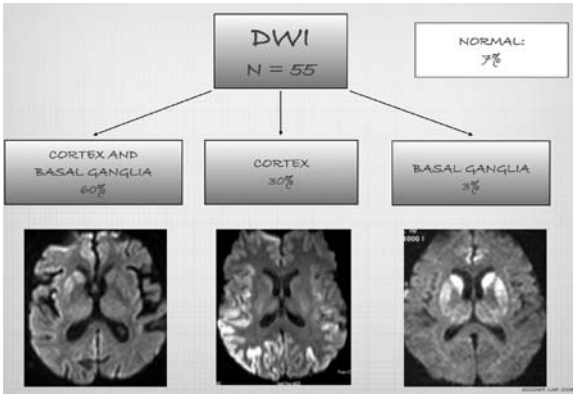
Dr Inga Zerr, National Reference Center for TSE, Georg-August University, Göttingen, Germany

Clinical diagnostic criteria for sCJD were first formulated 30 years ago, using a combination of distinctive clinical features and best available non-invasive investigations, which at that time was EEG. In recent years, there has been progress in developing other specialist investigations, including useful surrogate biomarkers (proteins, released from the nervous tissue in the cerebrospinal fluid), and clinical diagnostic criteria have been amended. Today, magnetic resonance imaging of the brain has been proven to be an important noninvasive tool in the clinical setting.

First reports on imaging findings in sporadic CJD (sCJD) were published in the 1980s and focused on brain volume reduction (atrophy) in brain imaging (computed tomography, CT and magnetic resonance imaging, MRI). With wider use of MRI, abnormally bright signal in some structures of the deep gray matter of the brain, caudate head and putamen, which are important parts of the brain's learning and memory system and higher-order motor control system, were reported in the late 1980s and early 1990s.

Subsequently, systematic studies of this bright signal changes were performed. With the emergence of more precise techniques, such as FLAIR (fluid-attenuated inversion recovery) and DWI (diffusion-weighted image), signal abnormalities were detected not only in the deep gray matter as described above, but also in the cortical structures. In variant CJD, the most typical finding represent bright signal in the posterior thalamus, another structure of the deep grey substance of the brain. This abnormality was also termed 'pulvinar sign'.

A series of recent publication has dealt with various clinical and pathological presentations in sporadic CJD. It became clear there is no single sporadic CJD type and



MRI scans for diffferent parts of a brain

affected individuals can present with various problems at onset. Some might complain memory disturbances, the others have visual problems, another group of patients might display prominent coordination deficits and gait disturbances. Some tests 'worked' in some disease subtypes, like the changes in the EEG are frequently seen in patients presenting with memory problems, but might be negative in those who develop problems with coordination and gait disturbances at the very onset. Thus, different tests have to be applied in the diagnostic workup in the clinic if the diagnosis has to be confirmed also in 'non-classical' disease types.

Utilising CSF 14-3-3 protein detection provides a higher sensitivity for less typical subtypes of sporadic CJD especially in individuals who are homozygous for valine at codon 129 of the prion protein gene. However, even for CSF 14-3-3 protein detection with an overall sensitivity of 85-95%, biological variables such as age at onset, codon 129 genotype and type of the abnormal PK resistant prion protein, modify the test results and cases with longer duration or younger age at onset may be missed by this investigation.

Acknowledging the limitations of the clinical criteria, the search for additional sCJD diagnostic investigations has continued. Magnetic resonance imaging has become increasingly important in the clinical diagnosis of sporadic and variant CJD. The use of sensitive FLAIR and DWI sequences allows the detection of the high signal in various brain regions. Characteristic lesion pattern have been reported for patients with different molecular disease subtypes.

The MRI is an extremely valuable technique in the evaluation of suspected CJD diagnosis, both by excluding other disorders and by demonstrating features considered typical of human prion disease. In a worldwide multi-centre collaborative study, MRI scans were analysed in a large number of cases of sporadic CJD and controls.

The study demonstrated that high signal in caudate head, putamen and cortex represent the most frequent MRI findings in sCJD and contribute to the clinical diagnosis in more than 90%. Characteristic brain MRI lesion patterns are helpful in establishing a diagnosis of sCJD and may help to identify less typical disease courses. As a result of this, defined MRI pattern were included in the clinical diagnostic criteria for sporadic CJD.

Genetic CJD and children

One family's story

When we were told in late 2007 that my mother died of a genetic form of CJD, we were deeply shocked. We had no previous experience of any other family members succumbing to this cruel and devastating disease, and we struggled for some time to come to terms with the impact the news had on our lives.

The doctor had explained the situation in clear and simple terms – I had a 50 per cent chance of inheriting the rogue Prion gene, and my children were at 25 per cent risk. Should I be tested and be tested positive, their risk would increase to 50 per cent.

We slowly began to digest this information and realised we faced a difficult decision as to whether to tell our two children, a 14 year old girl and 11 year old boy, about this risk.

Our initial instinct was to say nothing. As parents, we have a natural inclination to protect our children, and telling them they were at risk of developing the disease that so devastatingly killed their grandmother was definitely off the agenda. There was no conceivable way in which we would tell them. It would be irresponsible and even reckless.

No way – absolutely not, never in a million years.

Like most families who have more than one child, our two children are very different. Our daughter is quite deep, reflective and sensitive and our son is very happy go lucky whose sporting interests and social life gives him a healthy focus.

We felt that this 'protectionist' approach was for the best. We all want the best for our children. We want them to enjoy their childhoods and be worry free. The news we had was surely too heavy a weight to thrust upon their young shoulders. We were extremely wary of the news having a detrimental effect on their lives.

Over the next few months, we remained steadfast in our commitment to protect them. It was only when our daughter began to fall ill did we start to contemplate the possibility of changing our minds.

In early January 2008, our daughter was hospitalised in an adolescent unit. She was desperately unhappy and depressed. We naturally assumed that my mother's death had had a profound effect on her and that this was the cause of her unhappiness. Whilst it undoubtedly had affected her, it wasn't the sole reason and she spent the next eleven months in various hospital adolescent units whilst we tried to ascertain what was wrong with her.

It was the darkest period of our lives as she self harmed and took several overdoses, and we came perilously close to losing her on one occasion. After a long struggle, she received a diagnosis of body dysmorphic disorder (BDD), a condition where the sufferer is convinced they have physical defects, real or imagined. They become unhealthily preoccupied with their appearance and it has a detrimental effect on their lives.

Talking with the doctors, psychiatrist and psychotherapists, an emphasis was placed on trying to gauge how life was in the family. We spoke privately with the doctors about the genetic condition we were facing. We were told by one particular doctor that our daughter often felt that there was something other than her illness 'bothering' her parents and she often felt worried that she was to blame.

It was evident just how perceptive some children can be. As far as our daughter was concerned, it was apparent that her sense of something bothering us was adding to her overall feeling of anxiety. We naturally worried that this could possibly hamper her recovery. We started to contemplate telling our children.

We agonised over the decision for a while. We quickly decided not to say anything to our son.

Despite the turmoil of his sister being in hospital he seemed to be coping with life reasonably well, so why spoil things for him? He had a lot of healthy sporting and social distractions. The decision whether to tell our daughter was far tougher, as it was being considered against the backdrop of her having a quite serious psychiatric condition.

After much soul searching, and discussion with the doctor overseeing her care, we decided that not telling her could harm her recovery if she continued to blame herself for our perceived worries. It felt like we were damned if we did and damned if we didn't.

So, it was with a certain degree of trepidation that my wife and I sat her down in her room in a private adolescent unit last autumn and calmly told her that her grandmother died of a genetic form of CJD. We explained the statistics regarding inheriting the gene. We told her that I was going to have a genetic test sometime in the near future.

I felt we had plunged the handle down of a detonator box, crouched down behind a barricade, and were waiting for the explosion to occur.

To our surprise our daughter did not become hysterical or need sedating or any of the other things we feared. She had had an 'inkling' it was possible, she said, as she had done her own research on the subject. She was calm. She was rational. There seemed to be a mature acceptance that certainly surprised us.

Over the next few weeks, we were relieved to see that the news had not had any detrimental effect on her recovery, and general progress. Blips that occurred along the way were in spite of, and not because of, the news we had informed her of. In fact, at the end of November 2008, she came home and started going back to school. She had a lot to focus on, and the genetic issue seemed to have been put to the back of her mind.

In the spring of 2009, I was preparing to have my genetic test, and it was refreshing to be so candid and up front about it with my daughter. We spoke about it, more often than not briefly, and I sensed a certain amount of pride in herself because we had thought her mature enough to deal with this information, despite her problems. She continued to make great progress in her overall recovery and in generally managing her condition.

In the summer of 2009, my blood was taken for analysis and my wife and I waited nervously for three weeks for the result. My daughter waited too.

I tested negative.

The relief was palpable and the phone call immediately after to my daughter was a special moment in our lives I will never forget. She had been tormented with worry for three weeks but kept it from me as well as she could. She was overcome with relief.

My joy and relief was tempered somewhat as I thought about my sister, who fears having the test and anything involving medical procedures since CJD entered our lives. I thought about my mum. I also thought about people I've met through CJD who have tested positive. I still think about all of these people a great deal.

Since then, my daughter has continued to manage her condition really well. She is now 15 and currently studying hard in her final year at school for her GCSE's. We even recently told my son the whole story. He asked few tentative questions and then turned away to play on his Xbox, the way only 12 year old boys can do. I'm sure he'll ask a few more questions someday.

Telling our daughter was a huge risk, given the circumstances. She was very ill with a psychiatric illness that continues to affect her and sometimes I still can't believe we did tell her. However, sometimes in life I think your perception of reality after the event is very different from what actually happens, and that was certainly the case with us. She travelled along the journey with us, felt trusted, and I like to think it helped her recovery in some small way.

There is no right or wrong way to deal with something like this and my heart goes out to any family who may be going through something similar. There are strong reasons to tell and not to tell, and no amount of deliberating can make reaching a decision any easier. Our story is our story and unique to us. My hope is that it may help any family in a similar position reach a decision that is right for them as a family.

Good luck whoever and wherever you are, we're thinking of you. ■

CJD Support Network Management Committee 2010

Dr Angus Kennedy – Chairman
Consultant Neurologist

Mike Curtis – Treasurer
Mike, a former bank employee whose wife, Joyce, died of sporadic CJD in 2006

John Gilbert – Assistant Treasurer
John's brother in law died of sporadic CJD

Anita Tipping
Anita is a state registered nurse, RSCN, whose son David died of CJD through growth hormone injections

Roger Tomkins
Roger's daughter Clare, died of vCJD

Sarah Tomkins
Sarah's late husband Edward died of sporadic CJD

Alison Kenny
Alison's father died as a result of a contaminated blood transfusion. She is a RGN, nurse practitioner.

Judy Kenny Judy's husband, Deryck, was the first person to die of vCJD through a blood transfusion. Judy is a retired nurse

Derrick Biggs is our social services adviser and an operations manager with Cambridgeshire Social Services. He is the Association of Directors of Social Services link person for CJD

Professor Richard Knight
Richard is a Consultant Neurologist at the National CJD Surveillance Unit in Edinburgh

Andy Tomaso
Andy's mother Carmelina died of Genetic CJD in 2007

Eve Buckland
Eve's son Mark died of vcjd through a contaminated blood transfusion

Dr Andrew Smith Andrew is a Senior Lecturer in Microbiology at Glasgow Dental School

Gillian Turner – CJD Support Network co-ordinator

CJD Support Network membership

Becoming a member of the CJD Support Network adds to our strength and enables you to take a full part in the decision-making process and the work of the Network.

If you would like to become a member of the CJD Support Network and receive free regular copies of our newsletters and any other information we produce, please send £10 annual membership to the CJD Support Network, PO Box 346, Market Drayton, Shropshire TF9 4WN. Please make cheques payable to *CJD Support Network*.

However, if you are caring for someone wih CJD and would appreciate free membership, please tick this box ☐

Name ..... Title .....

Address .....

..... Postcode .....

Telephone .....

Email .....

I am caring for someone with CJD: ☐ at home ☐ in residential care

I am: ☐ a concerned relative/friend ☐ former carer
☐ professional ☐ interested

Recycle your mobile

The CJD Support Network has launched a 'Recycling your old mobile phone' scheme. Many larger charities use this scheme to raise money.

You will have received a freepost envelope with the newsletter; all you have to do is put your old phone in the envelope and post it. The CJD Support Network will be credited with £2.50 for each phone, however old.

Alternatively you can send them directly to the CJD Support Network, PO Box 346, Market Drayton, Shropshire TF9 4WN

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