CJD SUPPORT NETWORK

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Newsletter

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CREUTZFELDT-JAKOB DISEASE SUPPORT NETWORK NEWSLETTER

Is dental treatment a risk factor for variant CJD?

A dental study update

Pauline Watt and Fiona Ord, Research Dental Hygienists, NCJDSU, Edinburgh

This dental study began in April 2008 and has been funded by the Department of Health to assess whether dental treatment is a potential source of transmission of vCJD. To date, there is no evidence of transmission of vCJD through dental treatments.

In June 2008, letters were sent to families to gather names and addresses of the dentists where their relatives were registered. With the information received from families, it has been possible to contact many dentists to obtain as much information as possible about the dental treatments carried out.

This information is being collated along with the dental treatments of approximately 650 controls for comparison.

Dental practices are required to keep patient records for a number of years, however the dental treatments of some of the relatives have been destroyed. For those that have been destroyed the NHS Practitioner Services in Scotland and England/Wales are able to provide a record of treatments received, if claimed under the NHS. This information is stored as far back as 1998 and has been useful in building a history of treatments where the original records are not available. Much time and effort goes into locating dental practices and records as unlike medical records, dental records do not follow patients if they move from one practice to another. The collection of dental treatments is still ongoing and work has begun on inputting this information into a statistical database.

Informative articles have been published in dental journals to raise awareness of the study and poster presentations will be submitted for consideration at dental conferences in 2009/2010.

In the coming year, the information will be gathered and entered into a database and the results will be published. These will be distributed to committees e.g. SEAC and to organisations developing policies relating to public health issues.

NCJDSU would like to express their gratitude to all the families who have provided information. If anyone has not heard from NCJDSU regarding the study and would like to provide information on their relatives dentist, please contact Pauline or Fiona on 0131 537 2251/2345.

CJD Support Network Funding

The CJD Support Network has been fortunate in the past to have received over half of its income from a Department of Health Section 64 grant. Due to a change in DoH policy, this grant will no longer be available and in order to keep functioning, the Network is having to look for other possible income streams, from government and public sources.

The Network has always received generous donations from the public, but the current funding situation has increased the importance of these donations. One important source of donations is from fundraising activities

The CJDSN can help fundraisers with the organisation and collection of sponsorship money. We can provide collection boxes and items such as T-shirts and balloons. In addition, we can arrange for donations to be made electronically via a fundraising webpage designed specifically for the fundraising activity. This makes the collection of sponsorship money simpler and has the added advantage of increasing the money received by the Network through the tax system.

All fundraising activities no matter how small are very important and considerable sums are raised by events such as coffee mornings. We also receive donations from individuals or groups who are undertaking larger events. On the next page is a selection of activities being carried out to raise money for the Network.

NEWS IN BRIEF

vCJD in the Netherlands

A third definite case of variant CJD has been identified in the Netherlands. This was a female aged 49 with a clinical onset in 2008, who died in 2009. She had no significant history of travel to the UK or blood donation.

Clean blood transfusions

The Mail of Sunday (8 March 2009) raised awareness of minutes of a SaBTO meeting last year (The Safety of Blood, Tissues and Organs) which revealed that experts are considering limiting the 'risk reduction option' to those least likely to have been exposed to BSE in the nineties.

It is being considered that 'clean blood' sourced from BSE free countries would be offered to Vegetarians and children under the age of 16 if they needed a blood transfusion. Clean blood could not be applied to all recipients on grounds of feasibility or cost-effectiveness.

Gillian Turner, National CJD Co-ordinator of the CJD Support Network told the Mail on Sunday that she had concerns about the segmentation of clean blood.

She said that in the absence of any test in life for CJD, there is never any proof that someone is completely free from exposure to vCJD. She said people should not be discriminated against.

Judy Kenny (Trustee and committee member of the network) whose husband Deryck was the first person to die as a result of receiving contaminated blood, told the Mail that she could not understand why they ever thought blood was safe.

'The key thing should be protecting the whole blood supply. Knowing what I know, if I need an operation I'll donate my own blood.

Editor's note. We would be interested to have your views on the possibility that 'clean blood' could be offered to vegetarians and children under the age of 16 only. Please send your comments to the CJD Support Network, P.O Box 346, Market Drayton, Shropshire TF9 4WN.

Carleton Gajdusek

Carleton Gajdusek (1923-2008) who was awarded the Nobel Prize for his work on Prion Disease has died age 85.

PRION 1 Study

Recent findings show that Quinacrine at a dose of 300mg per day was reasonably tolerated but did not significantly affect the clinical course of prion diseases in this observational study.

Source: Safety and efficacy of Quinacrine in human prion disease (PRION 1 Study): a patient-preference trial. Prof. John Collinge et al. www.thelancet.com/neurology Vol.8 April 2009.

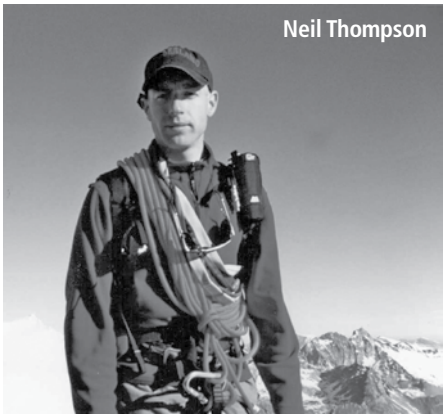
Fundraising 2009

Team Prion

Team Prion is a team of six people based at the Prion Unit in London. Members care for CJD patients and carry out research into prions and all strains of CJD. They are running the London 10k event on 12 July 2009, in support of the CJDSN. To find out more or to sponsor them please visit their website.
www.justgiving.com/teamprion09

Ultra Everest 2009

'It is not the mountain we conquer but ourselves.'


Neil Thompson

In spring of this year Neil Thompson will be part of a 10-man team climbing Everest by the South Col route. Donations may be made via his website where there are links to the expedition and Neil's blog.
www.justgiving.com/ultraeverest2009

Triathlon

William Tite's Mum recently died of Sporadic CJD. William says that the family received terrific support from the CJDSN and so they would like to make a donation to help support the work of the CJDSN.

He tells us that they will be doing a triathlon soon to help raise money for the Network. You can visit his website and donate at
www.justgiving.com/williamtite


Anna Edmonds

Flora London Marathon 2009

The CJDSN has secured a Silver Bond place in this year's Flora London Marathon on 26 April 2009. We are very pleased about this, as places are oversubscribed many times over. We have allocated our place to Anna Edmonds, who works for Firefly, a company which has generously donated a year's PR support to the CJDSN. Anna says 'Yes, it's true; I will be running the London Marathon in April 2009! As crazy as it may sound being a novice runner, I have good reason to take part in what will undoubtedly be the biggest physical challenge of my life. I'm running to raise money for the CJD Support Network, in memory of my dear dad who died with Sporadic CJD'. You can visit Anna's website for more information and to donate.
www.justgiving/annaedmond

Staffordshire Half Marathon

Rich Tittensor, his brother Eddie and one of his best friends Jackie, are running in the Staffordshire Half Marathon on 15 March 2009. They are running in memory of Richard's Dad, Alan, who died suddenly from Sporadic CJD ten years ago. Rich says that they are in full training now and hoping to beat the two hour mark! Parachute and Triathlon to follow.... You can see more and donate on his website **www.justgiving.com/richardtittensor**


Rich and his dad

Fundraising in 2008

The CJD Support Network would like to thank Mark Jones, Tim Wilson with Chris and Mark, Sarah Inwood, Simon Palmer, Nick Peters, Paul Elson, Martyn Robertson, Niall Cossins, David Wright, Joe Glammaiva, Jeremy Parker, Melanie Tong, Kevin and Donna Devlin and the O'Sullivan family for organising a variety of sponsored events during 2008, raising £15,000 between them for the work of the network.

Every penny raised helps us to run our 24 hour helpline and website. These provide practical and emotional support to families affected by all strains of CJD and for those people who have been informed that they are at increased risk of CJD through surgical instruments and blood transfusions. Donations also help us to provide financial assistance to families in need when caring for a patient with CJD and to ensure that patient and family needs, wants and experiences are at the centre of policy making. Donations are also used to support research.

Thank you all for your support and interest in our work.

My Mother

By Andy Tomaso

Carmelina Tomaso 1946 - 2007

We lost my Mother, Carmelina, to CJD in early October 2007. She had just turned 61 years of age. This story details the events that led up to her hospital admission and subsequent diagnosis of CJD, as well as the devastating and emphatic impact it has had on our lives.

My Mother worked happily as a dinner lady at a school in North London. She had been working there for a couple of years and I had not seen her so happy for a long time. She was a valued and respected member of the team and I sensed that it gave her a real sense of purpose. From the stories she told, it was obvious that there was a great sense of camaraderie, and she always had some story or other about what had happened in that 'mad kitchen'.

After the school summer holidays came to end in 2005 however, the job that seemed to give her so much pleasure suddenly caused her a great deal of anxiety. It coincided with a general lowering of her overall mood. She seemed nervous, edgy and generally anxious. She was reluctant to go to work and became socially withdrawn. She began to experience panic attacks which became increasingly severe. This led to her being hospitalised in the Mental Health Unit of our local hospital. She also decided that she could no longer face the prospect of returning to work. She decided to leave and felt relieved when my wife phoned and informed her manager that she wouldn't be returning to work. Mum had lost all her self confidence.

Over the next eighteen months, Mum's condition did not improve. She was hospitalised in the Mental Health Unit for several weeks on another two occasions. She was prescribed various anti-depressants and anxiety controlling medication, but nothing seemed to make any difference. The specialist overseeing Mum's treatment seemed to be surprised that Mum hadn't made any real progress. 'You shouldn't be here!' she once told her. This was an incredibly frustrating time as we were desperate for her to get back to her old self, but everything that was tried seemed to fail. What exactly was going on? It was in July 2007, after returning from a trip to visit family in Italy, that I got an indication that we were dealing with something more than depression and anxiety.

It had been four weeks since I had seen Mum. Waiting in the arrivals lounge at Stansted, I spotted her and Dad and made my way towards them. Mum seemed to be shuffling rather than walking, and although no more than ten to fifteen feet away, she could not see me. I called out to her and suddenly I registered in her field of vision. I was surprised, but it was very late, so I put it down to her being tired after the journey. As we walked down to the car, I noticed however that she was definitely walking more slowly than usual. She also seemed to have her left arm tucked into her body.

My family and I went away to Cornwall in August 2007, and whilst there I telephoned my Dad to see how Mum was doing. I remember being horrified when he told me that she now couldn't see a lot of the time. Dad had some work commitments that he had to attend to so I came back early to look after her. Driving back on my own from Cornwall that Friday night I had no idea what the next few weeks had in store for us.

The next morning I drove to Mum's and rang the doorbell. I stood in amazement as I heard her shuffle to the front door and fumble with the latch. After what seemed an eternity, she finally managed to open it. Helping her get ready, I realised the true extent of her lack of vision when she couldn't even see the end of her toothbrush. 'Where is it?' she asked as I held it about two feet away from her face as she had the toothpaste in her hand. As I watched her trying to brush her teeth, I noticed the involuntary shaking of her head.

At the end of August, Mum had an appointment with a Psychiatric Consultant. It was arranged to assess her progress under her latest anti depressant medication.

During the consultation, Mum would gaze up into space with her mouth open as if she had seen an incredible sight. The Consultant became increasingly concerned and quickly reasoned that Mum's condition was more that just a psychiatric condition. We left bewildered and I think the consultant felt the same.

That weekend saw Mum unable to walk unaided. In desperation, Dad telephoned the day centre for mental health patients that Mum had been attending and they told him to bring her straight to A&E. She was admitted to hospital on Monday 3rd September 2007, two days before her 61st birthday.

Mum was taken to the Clinical Decision Unit (CDU), where we nervously awaited news as to what Mum was suffering from. Parkinson's disease was mentioned, as was Porphyria, but all the tests came back inconclusive. Her condition deteriorated significantly during her week-long stay on CDU. Within the space of seven days, Mum

began hallucinating, lost the use of her arms and legs (apart from involuntary jerky movements), lost the ability to swallow, and perhaps most devastatingly, the ability to talk. What also caused a great deal of distress for me personally was seeing her flinch backwards several times in complete and utter fear, as if she had been given the fright of her life. I still wonder what she had been so scared of.

Mum was transferred to a main ward, and for two weeks we desperately awaited a diagnosis. Finally after many tests, and by what seemed a process of elimination, a Doctor informed us that they were almost certain that Mum had developed CJD. Sadly, we were told there was no cure. Our world fell apart.

We were visited by the CJD Surveillance Unit who sent somebody down from Edinburgh to interview us and examine Mum. We were also visited by members of the Prion Unit in London, who did the same. They also asked our consent for a blood sample to be taken from Mum for analysis. The reason was to see if Mum's CJD was an inherited form. Surely not? There was no history of this illness in our family. I signed the consent form with a certainty that it was only a formality and that the results would be negative.

Mum's condition continued to deteriorate. She developed huge blisters on her arms and legs in what appeared to be a completely unrelated skin condition. She developed a chest infection and was rushed to ITU where we almost lost her. Bravely, she fought back with the help of antibiotics and was returned to the main ward where she was given her own room. The palliative care team were assigned to Mum and they were fantastic in keeping her as comfortable as possible. She had a syringe driver fitted and it was agreed that the tube feeding her through her nose be removed, as this seemed to be causing her more discomfort. Her breathing became increasingly heavy as the effects of Pneumonia took hold.

After bravely holding on for several weeks since the physical symptoms of this cruel and devastating disease took hold, Mum passed away in the arms of my Dad and me on the afternoon of Thursday 4th October 2007.

Several weeks later, in mid November 2007, my wife and I travelled into London for a meeting with the people from the Prion Unit to discuss the results of Mum's blood test. We sat in utter disbelief as we were informed that Mum's CJD was genetic. My blood ran cold as the doctor also explained that there is a 50% chance that both my sister and I have the defective Prion Protein gene. I asked if I could find out and was told that there is a blood test available, but that a counselling process would have to be followed first.

'How could this be?'. I thought about my Mum's side of the family and couldn't think of one family member of who had suffered from CJD. It was very difficult to take in and digest. The enormity of finding out this news hit me harder when I thought about my two children. If I am 50% at risk, then they are both 25% at risk. Should I have a test and discover I am positive, their level of risk would increase to 50%.

As I write this, it is November 2008 and a year has passed since Mum passed away and we discovered that Mum's CJD was genetic. I was due to have the test earlier in 2008, but I postponed it as one of my children has been unwell and has spent most of the year in various hospitals. However, I have now decided to go for the test once again and am awaiting my appointment to see the genetic counsellor.

My wife and I have attended several seminars over the last year given by the Prion Unit in London. The information we have received regarding research into possible treatments has been extremely encouraging. The National Cohort study that the Prion Unit is undertaking is critical to this process, and I have recently enrolled onto it as somebody who is 'asymptomatic without confirmed disease'.

I am not sure how long it will take for me to find out the results. Fifty-fifty, red or black, heads or tails. I am certain I want to know. Some day soon I will.

Recent CJD figures

The number of deaths of definite and probable cases in the UK, up to 2 March 2009. Source: the CJD Surveillance Unit in Edinburgh

Year	Sporadic	Iatrogenic	Familial	GSS	vCJD	**Total**
2005	66	4	8	5	5	88
2006	68	1	6	3	5	83
2007	62	2	7	1	5	77
2008	78	5	2	3	1	89
2009*	3	1	1	0	0	5

- As at 2 March 2009

Total of definite or probable vCJD cases (dead and alive) in UK 168
Total of definite or probable vCJD cases (dead and alive) rest of world 44

National Prion Monitoring Cohort study

Professor John Collinge and Dr Simon Mead from the National Prion Clinic

The National Prion Clinic is pleased to announce that enrolment has started in the Department of Health funded National Prion Monitoring Cohort study. The purpose of this research is to carefully study patients diagnosed with, or at high risk of developing, prion disease to try to develop improved ways of reaching a diagnosis earlier and to understand how best to monitor patients receiving new treatments in the future as these are developed. All patients, regardless of what type of prion disease they suffer from and whether or not they are receiving any treatment, will be able to participate if they wish.

Drug testing

When new drugs are tested, this usually involves 'randomisation' where some patients receive the drug and others, selected at random, receive an inactive dummy drug or placebo. The two groups of patients are then compared over a period of time. This is the best way scientifically to see if a new drug offers overall benefit to patients. Understandably, with such awful diseases, for which there is currently no effective treatment, many patients and families may wish to try the new drug straight away rather than be randomised and this was our experience in the recent PRION-1 trial. As the prion diseases are quite rare, come in many forms and can be very variable in how rapidly they progress, it can be difficult to tell if a particular drug has overall benefit. For this reason, it is very important to build up a detailed picture of the progression of the different forms of the diseases and to determine how useful various types of tests are to monitor patients. The aim is to build up a very detailed understanding of disease progression that can then be used to compare with patients receiving new drugs in the future.

DEFINITION **A cohort is a group of people with something in common - in this case, being diagnosed with, or at high risk of developing, prion disease.**

This cohort study involves monitoring people over a period of time, to see how their condition develops.

Diagnosis

Learning how to make the diagnosis much earlier is important now, but will be increasingly important when we do have effective treatments as we will want to treat patients at the earliest possible stage, ideally before irreversible damage has occurred to the brain. Currently, many patients are diagnosed at quite a late stage when extensive damage has already occurred. Developing a blood test for CJD remains an important research goal and blood samples collected in this study will be made available anonymously to a National collection that will be used to help develop and validate such tests.

The study

The study will monitor changes in symptoms and a range of memory and other tests and brain scans through the course of the illness. We will work out which are the most appropriate tests for use in drug trials to measure whether drugs have an effect and assess potential laboratory tests that may also help us understand if a drug is working. In addition to getting ready for future trials of drugs we hope will slow down or stop prion diseases, we will also be studying drugs now that are used to treat some of the symptoms of the diseases and work out which are best. For example, we aim to study which drugs may be best to treat sleep disturbance or distressing jerky movements that occur commonly in prion disease.

Participation

A better understanding of how the individual prion diseases progress will help provide better information to support patients, their carers and families. Data will be collected in a standardised and systematic manner, like in a clinical trial, but at a level and frequency appropriate to the different patients. There are different levels of participation, the minimum level being one that simply allows the data collected as part of routine NHS clinical care to be recorded for research, together with a standardised basic neurological examination. For those patients able and willing to participate more fully, additional clinical information based on detailed doctors examination and memory tests, brain scans, and recordings of brain waves will be obtained and the neurological examination will be digitally recorded. A description of the expected rates of change through defined stages of human prion infection will be invaluable to support the testing of new drugs for prion disease in the future.

Additional Research

Much work is ongoing internationally to develop treatments for prion disease and two major projects are underway at the MRC Prion Unit in London

aimed at developing both antibodies and conventional drugs to treat prion disease. The National Prion Monitoring Cohort aims to pave the way for efficient testing of these and any other future treatments.

Prion diseases are, thankfully, relatively rare, and so to obtain sufficient information to make these advances requires a high level of participation. To facilitate this, participants unable or unwilling to visit the NHS National Prion Clinic (NPC) at the National Hospital for Neurology and Neurosurgery in London can be visited in their local hospital or at home as they wish by one of the NPC teams (consisting of a doctor and nurse) for assessment.

For an information sheet about the study or to ask any questions please call the NPC on 020 7405 0755 or visit www.nationalprionclinic.org

Two cases of variant CJD in the same family

Prof Bob Will and Prof Richard Knight, National CJD Surveillance Unit

In September 2008 the Spanish CJD Registry reported that two cases of variant CJD had been identified in the same family.

A man, aged 41 years, died of vCJD in February 2008, with the diagnosis confirmed after post-mortem. His mother developed a progressive neurological disorder at the age of 64 years in August 2007 and died in August 2008. The diagnosis of vCJD was again confirmed at post-mortem. This is the first time that two cases of vCJD have occurred in the same family.

Investigation showed that the mother and son had not travelled to the United Kingdom and that they must have been infected with BSE while living in Spain. There was no history of potential exposure to infection through medical or surgical treatments, for example previous blood transfusion. The mother and son were reported to have regularly eaten (and, indeed, shared) cattle brain and both lived in a region of Spain where BSE has been identified. Measures to protect the human population from exposure to BSE were introduced in Spain around 2000/2001 and it has been concluded that the mother and son were infected through eating cattle brain, probably before the measures to protect the public from BSE were introduced. This conclusion has been endorsed by the European Centre for Disease Prevention and Control (ECDC) and by the UK SEAC committee. The fact that the mother and son developed symptoms of vCJD within a short time of each other suggest a shared exposure (i.e. both eating infected cattle brain) and makes any other form of transmission (such as from one person to the other) very unlikely.

An important question is whether the occurrence of two cases of vCJD in the same family is relevant to the UK and other countries such as France in which cases of vCJD have been identified. The ECDC pointed out that large numbers of family members of vCJD cases have not developed the disease and this suggests that the risks to family members must be low. In the UK there are 450 family members of 166 cases (123 children and 327 siblings) none of whom have developed vCJD to date. Furthermore the first case of vCJD in the UK was identified in 1995 and this means that even after 14 years no family members have developed vCJD. The evidence suggests that vCJD in the UK (and probably other countries) was caused by exposure to food products containing high levels of infectivity, perhaps through the use of mechanically recovered meat. This is quite different from the probable exposure in the Spanish family directly to bovine brain and this may explain why there has been only one case of vCJD in each affected family in the UK, France and other countries.

The precise type of dietary exposure that caused vCJD in the UK is not known and may never be known. However, the fact that there has been only one case of vCJD in each family indicates that, despite a common diet in many cases, the risk to family members is very low and perhaps negligible. Why? One explanation may be that there was only rarely enough infectivity in food to cause infection with BSE and this was not evenly distributed in foodstuffs. There may have only been enough infection in occasional portions of food to transmit the infection. This is hypothetical, but the authorities in the UK and Europe have not suggested any change in policy since the identification of the case in the Spanish family and family members of vCJD cases have not been classified as at greater risk of vCJD than the general population.

Creutzfeldt-Jakob disease, prion protein gene codon 129VV, and a novel PrPSc type in a young British woman

Dr Simon Mead and Professor John Collinge, NHS National Prion Clinic

This article is an outline of a recent paper from the MRC Prion Unit in the medical journal *'Archives of Neurology'*. It was widely reported on.

Readers of the CJD Support Network Newletter will be well aware that much of the UK population has been potentially exposed to Bovine Spongiform Encephalopathy (BSE, commonly known as 'mad cow disease') by eating contaminated food in the 1980s and early 1990s.

For over a decade we have known that the infectious agent or prion that causes BSE may also infect humans, causing the fatal brain illness variant Creutzfeldt-Jakob disease (vCJD). While fortunately numbers of patients developing vCJD have been relatively small so far, and indeed numbers of patients seen have been falling in recent years, we do not know how many people might be silently infected with BSE prions. It is known that it may take several decades for signs of the disease to show and that our genetic make up is important in determining this. We also know from research in laboratory mice that some may become 'carriers' of the infection and do not themselves develop the disease. People with such a 'subclinical' infection would still pose a potential risk to others by blood donation or contamination of surgical instruments for example.

We have known for many years that a common variation in a gene known as the prion protein gene is very important in determining risk of developing prion diseases and how long it takes for the disease to develop when someone becomes infected.

There are three genetic types in the UK population known as MM, VV and MV. So far, vCJD has only affected one genetic type of individual: MM. Around 40% of healthy people in the UK are MM, about 50% are MV and around 10% are VV. It is likely that BSE prions will infect people of the VV and MV types also, but they may have much longer incubation periods (the time taken from being infected with prions until the brain disease becomes apparent) and may also develop a pattern of disease which may be different to vCJD. We suspect this again as result of research in laboratory mice where those that had the VV and MV genes had a different type of disease and different types or 'strains' of prions developed.

Kuru was an epidemic prion disease which affected the Fore people living in the Eastern Highlands of Papua New Guinea. It was the practice in these communities before 1960 to consume their dead at mortuary feasts as a mark of respect and mourning and this led to the prion infection passing between people. Kuru has now largely disappeared but from studies over the past decade we have learned that the incubation periods can be over 50 years and that the MM, VV and MV people have different incubation periods, MV individuals having the longest. A similar effect was seen in people accidentally infected with prions when they were treated with growth hormone injections before 1985 (so called iatrogenic CJD).

Some years ago at the National Prion Clinic we looked after a young woman with a severe dementia and loss of balance. Her examination and other tests such as her MRI brain scan suggested that her diagnosis was sporadic CJD. Most patients with sporadic CJD are MM, but the disease also occurs in VV and MV patients. Her genetic type was VV. After she died we investigated the cause of death further by microscopic examination of the brain, and by testing the abnormal prion protein deposits in the brain. Sporadic CJD is usually a disease of late adult life while vCJD has largely occurred in young adults. Because she was young, we obviously wished to consider the possibility that her illness was related to BSE rather than being a sporadic case. The autopsy findings were not typical of sporadic CJD, with a lot of prion protein deposited in the brain. However, these changes were not similar to vCJD either. Also in vCJD, prions are deposited at high levels in certain tissues

outside the brain and spinal cord known as lymphoreticular tissues (lymph nodes, tonsil and spleen or example) but these samples were not available for analysis from this patient. Prions occur in different forms or strains. These can be distinguished by looking at the rogue prion protein (known as PrPSc) from brain tissue. In this patient we found a molecular strain different from any of the types we have seen in sporadic CJD and similar to that which we see in vCJD (which we call type 4 PrPSc). Although quite similar to type 4, we showed that the new type was different because it could be distinguished from the typical vCJD pattern by additional special tests.

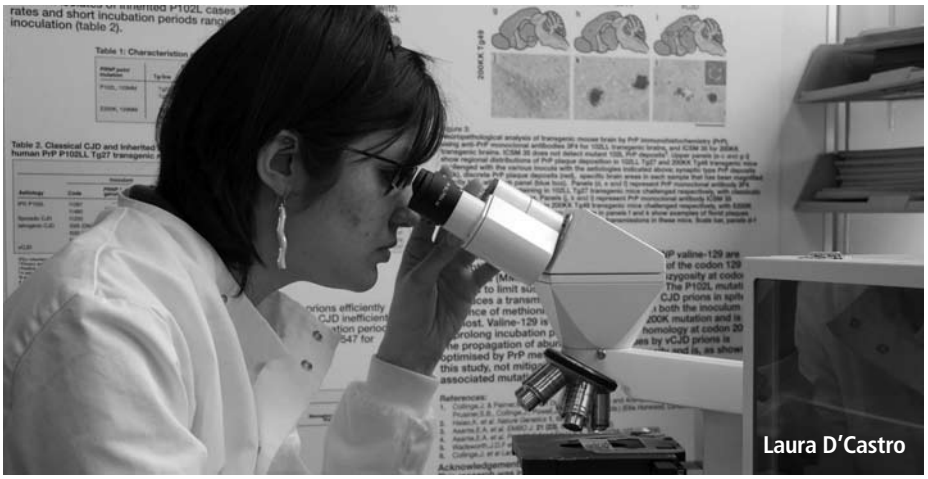
In summary, although this young woman clearly died of a prion disease, we could not conclude she had vCJD, but also we could not be sure this was sporadic CJD either. Although we were not able to make definite conclusions on the basis of a single patient, we felt it important to report this tragic death and these findings in the medical literature so as to bring this to the attention of other doctors who might see similar patients in the future. This unusual finding reminds us of the importance of keeping alert to the possibility that BSE prions will cause disease in individuals with different genetic types, who may develop a disease that may resemble sporadic CJD, or vCJD, or have a new pattern of disease. This work emphasises the importance of continuing to study the rogue prion protein type in patients and we thank all patients and families who have kindly consented to the use of their, or their loved one's, tissues for this medical research.

NHS National Prion Clinic, National Hospital for Neurology and Neurosurgery, London. www.nationalprionclinic.org

The shape of prions

A summary of a report by Laura D'Castro given at our family support meeting in Milton Keynes.

Laura's research is being carried out at the National Prion Unit and is partly funded by the CJD Support Network. Funds have been donated by members who have asked for their donations to be ring fenced for research.


Laura D'Castro

I was very kindly invited to speak to you at the Family Support Meeting of the CJD Support Network held in Milton Keynes in November. In the first part of my talk I provided an overview of prion diseases, and then went on to describe in detail my research which the CJDSN is generously supporting.

Prion diseases are associated with the presence of rogue, mis-shaped versions of naturally occurring prion protein. Once formed in the body, rogue prion proteins convert normal prion protein into a likeness of themselves setting off a chain reaction leading to progressive accumulation of the rouge protein in the brain. This causes the death of nerves resulting in the clinical symptoms of prion disease.

The key aim of my research is to discover the precise shape of infectious rogue prion proteins. We think that this knowledge will be of great benefit to developing new therapies for treating prion diseases.

In my research I will be initially working with an experimental prion strain, which provides a model for human prion diseases. I aim to purify the rogue proteins and examine them using high powered microscopes.

At this stage of our research, our purified samples mainly consist of rogue prion protein together with other minor contaminants. We are presently investigating what these contaminants are and how they may be involved with the progression of disease.

Our microscopic investigations have now started and we are able to see infectious rouge prion proteins forming bundles and individual rods. The aim now is to take detailed pictures of the individual rods and build up a three-dimensional image of the rouge prion protein using computers. We still have much more work to do, but we feel that we have made excellent progress in this first year.

I would like to thank you once again for your generous scholarship which is helping me to carry out this work and I look forward to meeting you again next year and reporting on our progress.

Variant Creutzfeldt - Jakob disease (vCJD) and patients with bleeding disorders

Information and advice given by the Health Protection Agency

A person with haemophilia has been found to have evidence of infection with the agent (abnormal prion protein) that causes variant Creutzfeldt-Jakob Disease (vCJD) only in his spleen at post mortem. All Haemophilia Centres are contacting their patients throughout the UK to give them this information.

Tests carried out on a haemophilia patient who died last year have shown that he was infected with variant Creutzfeldt-Jakob Disease (vCJD). The patient did not die of vCJD, and never had any symptoms of this disease when he was alive. The patient was in his 70s when he died of a completely unrelated cause. The tests were carried out as part of an ethically approved research study jointly co-ordinated by the UK Haemophilia Centre Doctors Organisation and the National CJD Surveillance Unit.

This haemophilia patient had been treated with several batches of UK-sourced clotting factors, including one batch of factor VIII that was manufactured using plasma from a donor who went on to develop vCJD.

A final view as to how this haemophilia patient became infected with the vCJD abnormal prion protein has yet to be reached and investigations are therefore continuing to establish this.

This is the first time that the vCJD abnormal prion protein has been found in a patient with haemophilia, or any patient treated with plasma products. This patient did not die of vCJD, and the only reason we know he was infected with the vCJD abnormal prion protein is because of the research tests carried out after he had died.

This new information does not change the way any patients with bleeding disorders will be treated.

Patients who have a bleeding disorder or congenital antithrombin III deficiency[1] and who received clotting factors or antithrombin derived from UK-sourced plasma[2] between 1980 and 2001, should have already been told that they have an increased risk of vCJD, and should be following public health advice (see box below).

If patients are unsure about this and would like more information, they should contact their haemophilia centre.

Other patients with bleeding disorders who do not have an increased risk of vCJD, do not need to take any action. Patients should contact their haemophilia centre if they are unsure about their vCJD 'at risk' status.

This case does not change the public health 'at risk' status of any patients with bleeding disorders.

Patient information leaflets and other information are available on the Health Protection Agency website http://www.hpa.org.uk/vcjdplasmaproducts

How to reduce the risk of spreading vCJD to other people

If you have been identified as being at increased risk of vCJD, you can reduce the risk of spreading vCJD to other people by following this advice.

- Don't donate blood. No-one who is at increased risk of vCJD or who has received blood donated in the United Kingdom since 1980 should donate blood
- Don't donate organs or tissues, including bone marrow, sperm, eggs or breast milk
- If you are going to have any medical or surgical procedures, you should tell whoever is treating you beforehand so that they can make special arrangements for the instruments used to treat you
- You are advised to tell your family about your increased risk. Your family can tell the people who are treating you about your risk of vCJD if you need medical or surgical procedures in the future and are unable to tell them yourself.

Background information on the 2004 notification

In 2004 the HPA, the UKHCDO and colleagues notified patients who had received plasma products manufactured using plasma from donors who had developed vCJD.

That notification dealt with plasma donations which had been used to manufacture factor VIII, factor IX, antithrombin, intravenous immunoglobulin G, albumin, intramuscular human normal immunoglobulin and anti-D.

Patients treated with these plasma products were managed according to an assessment of potential vCJD infectivity carried out by the Health Protection Agency with the CJDIP.

The UK Haemophilia Doctors and patient representatives, the CJDIP and UK Health Departments agreed that it was likely that many patients with bleeding disorders would have had a sufficient exposure to these implicated plasma products to put them 'at risk of vCJD for public health purposes'. It was also thought likely that further batches of UK-sourced plasma products would be implicated in the future as more cases of vCJD arose.

Therefore all patients with bleeding disorders who were treated with UK-sourced pooled factor concentrates or antithrombin between 1980 and 2001 are 'at risk of vCJD for public health purposes' and public health actions should be taken. This risk is in addition to the general risk of vCJD that many people in Britain have through eating beef or beef products.

1 Congenital and acquired haemophilia (Haemophilia A and Haemophilia B), Von Willebrand Disease, other congenital bleeding disorders and congenital antithrombin III deficiency.

2 Factor VIII, factor IX, factor VII, factor XI, factor XIII and prothrombin complexes, as well as antithrombin.

Family Support Meeting

A summary of the opening address by Dr Angus Kennedy, Chairman of the CJD Support Network, at the annual Family Support Meeting in November last year

Dr Kennedy warmly welcomed 40 members of the network; families with experience of vCJD, Sporadic and Genetic CJD were present, together with families who had lost family members through secondary transmission and growth hormone injections. We were also very pleased to welcome a number of staff from the National Prion Unit

Dr Kennedy talked of the importance of our helpline and the need for funding to enable our work to continue. All information books and factsheets had been updated in 2008 and he thanked everyone who had helped with this.

He noted how valuable the committee's visits to the National CJD Surveillance Unit and the National Prion Unit had been. He thanked the staff of both units for being so welcoming and helpful.

Dr Kennedy thanked Firefly, in particular Anna Edmonds, for their year's free help with publicity and awareness-raising of the network.

Gillian Turner had attended many meetings throughout the year to ensure that the patient and family needs are at the centre of policymaking. He mentioned in particular a meeting of NeuroPrion in Madrid, which she attended along with CJD groups from Australia, Japan and Italy. The groups gave a joint presentation about their work to over 800 researchers of prion disease from all over the world. NeuroPrion said that it is important that patient stakeholders have a presence at this meeting as it reminded researchers that there was a patient at the end of their test tubes.

Dr Kennedy also praised the work of Committe members Sarah, Roger, Mike and Judy, who had also attended meetings on behalf of the network during the year.

He thanked fundraisers and sponsors for an excellent year, raising over £15,000. For the last 12 years the network has received Section 64 funding from the Department of Health, but because of changes in the funding criteria we will not qualify again. We need to acquire new funding lines. It was felt that we now need to charge a membership fee to help with costs. He asked those present for their views and it was agreed by them.

He reported that we had at least four large fundraising initiatives in place for next year; Anna Edmunds running in the London Marathon, Team Prion from the National Prion Unit running in the Charity London 10k, Neil Thompson climbing Everest – to mention just a few. He hoped each member would give their support.

Dr Kennedy gave an update on the educational DVD which the network is hoping to produce. Although we were refused funding in 2008 from the Department of Health to make a DVD aimed at GPs, we were encouraged to submit a new funding application for a DVD aimed at infection control teams. The application was submitted in November and we should hear a decision sometime in the New Year.

Dr Kennedy ended his report by thanking Gillian and the Committee for all their hard work and support over the past year.

CJD Support Network Management Committee 2008-2009

Dr Angus Kennedy – Chairman
Consultant Neurologist

Maria Bryne – Secretary
Maria is a mother with three children whose husband Graham died with GSS

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

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.

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

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

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

Gillian Turner – CJD Support Network co-ordinator

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

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, P.O Box 346, Market Drayton, Shropshire TF9 4WN

CJD SUPPORT NETWORK
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