CJD SUPPORT NETWORK

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Newsletter

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CREUTZFELDT-JAKOB DISEASE SUPPORT NETWORK NEWSLETTER

Family Support Meeting and AGM

We held our annual Family Support Meeting on Saturday 3 November 2012. This proved to be a very successful day, attracting 50 members of the network. Family members attending included those who had experienced the loss a loved one from all strains of CJD, members who were at risk of Genetic CJD and people who were currently caring for a family member.

We received many *thank-you*s and feedback from people who attended. Below is a sample, which I hope will encourage others to consider attending this year.

Summaries of their talks given at the meeting have been very kindly supplied by Mellissa Hillier, Prof Richard Knight, Derrick Biggs and Simon Mead. See pages 8–13.

'Thank you for another very informative day. It was good to meet up with others and share experiences.' Liz

'Just to say that we found the Family Support Meeting very informative and how helpful it was to meet other people who had experienced similar events.' Sandra and Paul

'We just wanted to drop you a line to express our gratitude for the support and opportunity to discuss issues pertinent to CJD at this year's Family Support Meeting.

We thought all the speakers were excellent; they were not only informative, but so enthusiastic in their work to improve outcomes for those affected by these diseases on several levels. The first meeting we attended was five years ago: it seemed then that anyone diagnosed with CJD could only be offered palliative care. Now we learn that research has come on in leaps and bounds, and there is hope that others, in time, will not have to go through the same ordeal. This work is truly admirable, a comfort to those who have suffered at the hands of CJD in the knowledge that others may not have to endure the barbaric consequences of this disease.' Sandra and Mike

Family Support Meeting 2013

Our annual Family Support Meeting in 2013 will be on **Saturday November 23rd** at the Burlington Hotel, New Street, Birmingham. We do hope you will be able to join us as we have so many families telling us how beneficial they find attending the day. They find it helpful to meet and talk to other families who are caring for, or have lost a loved one through CJD and to be able to ask those unasked questions to CJD professionals.

Please make a note of the day in your diary.

Living with genetic CJD

Stuart Thompson

As an individual living with a positive diagnosis of a rare genetic disease (Inherited Prion Disease or familial CJD) I am always interested in making new connections with networks that may support people/families who find themselves in a difficult and challenging situation – such as living with or being bereaved by CJD or other rare disease.

It was through the Genetic Alliance that I was made aware of an organisation called Rare Diseases UK (www.raredisease.org.uk). I decided to take up individual, free membership.

In April 2013, through the alliance's email mailing list, I was made aware of a very interesting and in-depth *Rare Disease Impact Report*, which can be accessed by using the web address above. A key point of this report is that it was collated from the views of patients, clinicians and the payor. So, the report in effect was gathered from the 'community for the community'.

The report looks at and analyses aspects such as financial implications, diagnosis issues and emotional impact.

Whilst some of the findings are not specifically relevant to our very own CJD issues it is interesting to see the resonance of some of the issues and learning found in this report that we may have as individuals, families or organisations. I personally found it reassuring to know that the diagnosis and on-going support structures I have experienced are well advanced in comparison to some of the experiences shared in this report. I was also reminded that the issues highlighted regarding emotional impact and support are unique and often long term.

I hope our members and readers find the report of interest.

Donations in memory

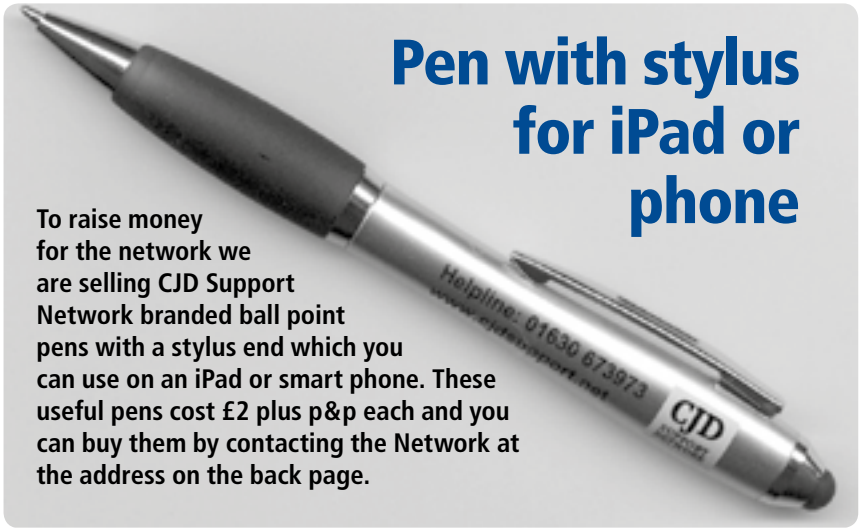
Heartfelt thanks to the families and friends of those below for donations received in their memory between April 2012 and March 2013.

Julian Bailey
Patricia Anne Barnes
Adrian Barras
Alan Bedford
Brenda Bosley
Kenneth Cookson
Ian Jonathan Cox
Stanley Cozens
Robert Craig
Ben Dale
Irene Denne
Margaret Dodd
Linda Fanning
Peter Gatens
Mick Golding
Stuart Hanslow
Peter Hardy
Alan James Hill
David Hitch
Iris Helen Inkpen
Jeanne Mary Hooper
Ann (Jacqueline) Morris
Paul Hope
Michael Lynch
Kathleen Lucey
Margaret Doris Morrell
Susan Jane Potter
Della Scribbins
Carol Smith
Mr Smitherd
Tony Swain
Catherine Whitbourne
Peter Willoughby
Linda Young

We would also like to say a big thank you to all those fundraisers in 2012 who ran marathons and fun runs, climbed mountains, organised coffee mornings, clothes swaps, packed in supermarkets, asked for donations instead of birthday and anniversary presents, gifts from employers – and much more. The range of fundraising ideas has been amazing. Donations in memoriam and raised by fundraisers during April 2012 to March 2013 totalled £37,865.

We are very grateful for this fantastic support for our work and would like to assure you that every penny is used to help families affected by all strains of CJD.

Pen with stylus for iPad or phone



To raise money for the network we are selling CJD Support Network branded ball point pens with a stylus end which you can use on an iPad or smart phone. These useful pens cost £2 plus p&p each and you can buy them by contacting the Network at the address on the back page.

Collection boxes

You will see that we have enclosed with the newsletter a flat CJD Support Network collection box. If you would like to save any small loose change lying around the house and donate it to the charity you can fold the card and make a collection box. We are always very grateful for members' support.

Stocken Prison Fun Run raises more than £1,000

The Grantham Journal reported on the 18 May that colleagues of a Grantham man who died from CJD raised more than £1,000 in his name at the weekend.

Colleagues of a Grantham man, Ian Cox – who died on April 26 from Iatrogenic CJD at the age of 46 – have raised more than £1,000 for the CJD Support Network. Ian worked as a chef at Stocken Prison in Rutland and the funds were raised for the charity through the prison's annual fun run.

Ian's CJD was caused by human growth hormone injections he received as a boy (see page 13 for more on this).

The Grantham Journal reports: 'Mr Cox's father, Barrie Cox, said: "It was a really nice family day and we had a lot of support. Ian's whole family went along and we were all made to feel very welcome. It was nice to see because he was our son and he was a great lad."'

The Journal goes on to say, 'Three men who worked with Ian in the prison kitchens – Danny Pinner,

Darren Clark and Fred Murad – ran the 10km race as a team wearing special CJD Support Network T-shirts displaying a picture of Ian on their back. Despite having never run such a long distance before, they all made it round in 51min 38sec. More than 40 people took part in the run.'

Email corner

Hi Gill,

I thought I would update you on my most recent fundraiser.

I packed shopping bags at Sainsbury's on Thursday 28 March as I was told that on the day before Good Friday there are lots of shoppers and people are very generous.

I mentioned the plan to a teacher at my old school and she managed to get forty-three willing volunteers in all different years at the school to sign up and do a few hours bag packing – even on a day when they were off school. We filled fourteen tills from 10.30 until 4pm and I stayed on alone until about six.

Counting the money took hours and this morning I finally finished bagging the coppers. The money raised from just that one day at Sainsbury's came to £843.28.

I found it astonishing how many people knew about CJD from the past but thought it had completely disappeared and didn't affect people anymore. And even if they didn't donate even a penny it was still a brilliant way of bringing awareness of the disease to the public in my town.

If there are any more fundraising ideas that you have for me please do let me know and I would love to give them a try. I plan on fundraising yearly for the charity and I hope I can be even more involved in the future.

Kindest regards,
Hollie Sullivan

Calling all runners

The 2013 British 10k London Run is on Sunday 14 July 2013. We are pleased to report that we have secured six places. If you would like to run 10k around the famous sights of London, raising money for the CJD Support Network please contact me on 0630 673993 or gturner@cjdsupport.net

Hello Gill
I wanted to let you know how fantastic the video clip is about the CJD Support Network that's now on the website: www.cjdsupport.net
Christy.

Donations delivered in style

Ian Morris's wife Anne (Jacqueline) Morris died recently of Sporadic CJD. Both Ian and Anne loved motorcycling. Ian wanted to deliver the cheques that had been donated at Anne's funeral to us in person and decided to make the journey on his motorcycle as he felt that Anne would be by his side.

Ian said that Anne, however, had had other ideas. His motorcycle broke down at the beginning of the journey so he had to push it back home. He then decided to do the journey on Anne's passion, *her* motorbike.

We are pleased to report that Ian arrived safely with the cheques. We are very grateful for the donations.

Below is his letter accompanying the donations.

Dear Gillian

Please find enclosed a number of cheques which were donated in memory of my wife Anne (Jacqueline) who passed away on 19 January this year.

It is the wish of myself, Rebecca and Rachel (my daughters) that this money is used by the CJD Support Network to continue their brilliant work helping and supporting the families affected by this horrendous disease.

I am also hoping to be at the support day in Birmingham in November (along with my family), as I found the information we received at last year's meeting invaluable.

I must also offer our thanks for all that you helped us with during the dreadful time of Anne's illness.

Yours sincerely
Ian Morris

Ian Morris

Uncertainties around a screening programme

The CJD helpline received numerous calls from the media (including Russian media!) about the article in the Sunday Times, written by Rowena Mason on Sunday 28 April about Britain being put at risk of CJD through blood transmission. The article told of the concerns of Government that 30,000 people could be carrying the dormant form of vCJD in Britain – double the previous estimates. The infection could then be transferred to many more people from them by blood transfusions.

Mason reported that Frank Dobson, a former health secretary, urged ministers to develop a nationwide screening programme for blood donors to stop future infections of vCJD, 'which has the potential to cause horrendous deaths'.

Prof. Richard Knight from the National CJD Research and Surveillance Unit kindly gave the Network the following comment on the article.

There is evidence that some people exposed to BSE infection through diet in the UK, during a period that may extend from 1980 to 1996, are currently infected but are not clinically affected (ie they are currently perfectly healthy). However, many uncertainties exist, including: the number of people so infected; whether or not they will all become ill at some point (and, if so, when); and whether or not all such infected people can pass infection onto others (and how).

Estimates of the number of infected people have been based on studies of tonsils and appendices removed during routine surgery. A recent study has suggested a higher number than previously thought but there are problems with the interpretation of these estimates and important uncertainties remain.

It is established that variant CJD infection can be transmitted from person to person via blood transfusion, but despite careful study only four instances have been identified so far. In fact, although any individual instance is tragic, this figure is significantly lower than what would have been expected if one makes straightforward interpretations of current infection estimates.

A great deal of work has already and is continuing to be put into developing a blood test for vCJD and, while there are promising outcomes, there is currently no completely validated test that can be used routinely to screen blood donations. Routine screening of materials like blood is a highly complex matter and even with a technically effective test, there are a lot of difficult considerations that need to be made before implementing such a test in everyday practice.

CJD figures

Recent figures from the National CJD Research and Surveillance Unit in Edinburgh

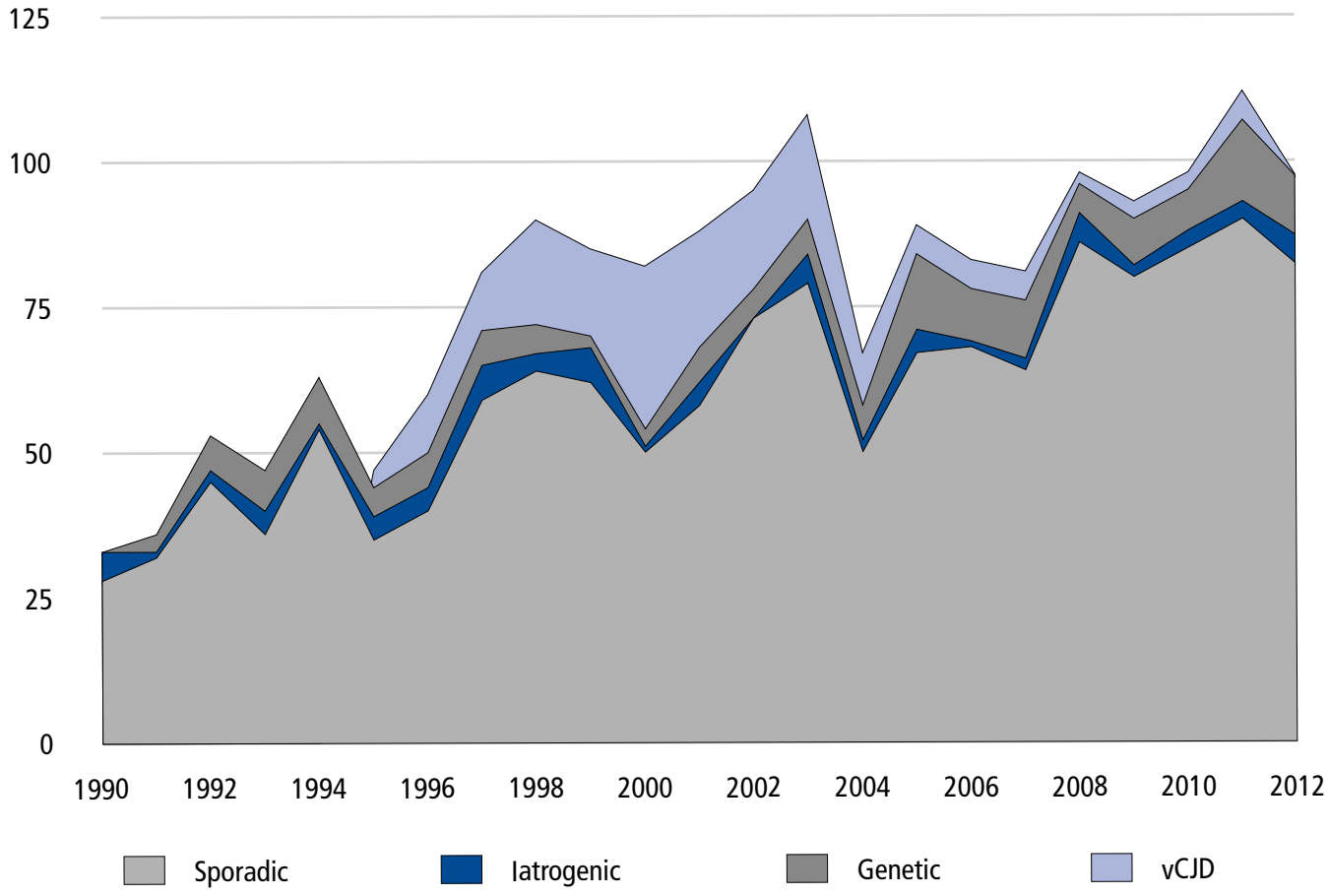
Recent numbers of deaths of definite and probable cases in the UK

Year	Sporadic	Iatrogenic	Genetic‡	vCJD	Total
2009	80	2	8	3	93
2010	85	3	7	3	98
2011	90	3	14	5	112
2012	83	5	10	0	98
2013*	16	0	1	0	17

*As at 6 May 2013 ‡Includes all genetic prion disease, including GSS

Definite or probable vCJD cases (dead and alive) in UK to date	176

Recorded deaths for the years 1990–2012



Genetic CJD Workshop

Melissa Hillier, Assistant Director, Genetic Alliance UK

In the morning workshop patients discussed their experiences of living with genetic CJD. In the afternoon we broke into two groups to discuss what had worked well and areas that needed improving. Below is a short summary of the discussions.

Positive experiences

Those patients that had been referred quickly to the Prion Clinic all experienced high quality services and information in a professional manner. All those who had attended the clinic found it useful and helpful.

Some participants hadn't been referred smoothly to the Prion Clinic and their experiences were less positive up to that point.

People had been offered the opportunity to take part in research and this was welcomed. They also valued learning more about the condition from the researchers and clinicians.

It was noted that in some cases the information provided was quite complex and difficult to digest and that further opportunities to discuss issues would have been valued.

Participants who had received support from a trained counsellor all found this to be very useful and needed. It enabled them to explore their feelings and in part come to terms with the knowledge of having genetic CJD in the family.

Peer-to-peer support was excellent and patients and family members really valued this. The experience of talking to others affected by genetic CJD was invaluable.

Attending CJD Support Network conferences was felt to be really important and a good opportunity to keep up to date with research, meet with other families and generally enable people to be aware of developments in the prion disease field.

Areas that could be improved

Many people felt that the psycho-social issues were not dealt with well. Or rather, they were not discussed enough. The longer term effects of knowing that you have genetic CJD in the family meant that people sometimes wanted and needed to re-visit issues and felt that a one-off counselling session was not appropriate.

Discussing the issues of inherited conditions with children was another area were families wanted more support.

Participants noted that they often wanted to 'dip in and out' of information on genetic CJD but that they didn't always know where to turn for further support and guidance.

Discussing the issues with other family members – how to break the news, how to discuss the impact of the condition and the issues around whether or not to be tested – were all areas where people felt they would like more support, be that peer-to-peer, or professional support such as counselling.

Learning about what types of counselling is available and who would be the most appropriate to see would be useful. Many found that local counselling services weren't well equipped to deal with the issues surrounding genetic conditions and genetic testing.

Possible actions

Peer-to-peer support to be developed

Maybe a sub-group of CJD Support Network where families could communicate with each other – a website is being developed currently and this may be very helpful and allow for online communication too.

Use of social media

Most people who attended the workshop were fairly internet savvy and thought this could be a good way to communicate with other people affected by genetic CJD – again the creation of the new Facebook page could address some of these issues

Counselling for long term conditions

Does this exist? We discussed looking and working with other patient support groups such as the Huntington's Disease Association to find out what kind of support they provide, or that they know about, that may also be appropriate for those affected by genetic CJD.

Information for children

We discussed how difficult it is to discuss genetic CJD with children and how patients and family members were unsure as to how/when/where this should be done. Development of information specifically aimed at children. The facilitator mentioned that other charities are developing such information and that contact details could be forwarded.

Adult Social Care

Derrick Biggs, Head of Provider Services at Cambridgeshire County Council and lead officer for Association of Directors of Social Services on matters pertaining to CJD

The purpose of this talk was to present information to assist people with CJD and their family carers in accessing services from local authorities

Adult Social Care is the responsibility of local authorities in England and Wales. Often local authorities will also work in partnership with health authorities. However, authorities do work differently so this summary may be implemented differently by your authority on some aspects.

Allocation of resources is according to need and not a diagnosis. People with support needs are encouraged in the first instance to contact their local adult social care services.

The first aspect is for an assessment to be carried out. It is a responsibility for local authorities to offer and carry out an assessment.

Following an assessment that somebody has eligible needs and meets the criteria for care then a care plan will be devised. There are no specific timescales but people are encouraged to try to work within a four week period to carry out an assessment.

Recent major changes

In recent years there have been major changes in the way that Adult Social Care is delivered.

First of all, local authorities are moving towards personal budgets whereby an amount of money is determined in order to meet people's support needs. Once decided the personal budget can be managed by Adult Social Care, but if people want to have the money themselves they can take it under a direct payment and buy services themselves. People can also opt to have a mixture of taking some of the cash to buy services themselves and having some services provided for them by the local authority or agencies acting on their behalf.

In terms of charging there still is in place an exemption from paying domiciliary care services for people with CJD. However, this does not cover residential care. People entering residential care will still be required to pay a fee unless it's under Continuation of Health Care funding.

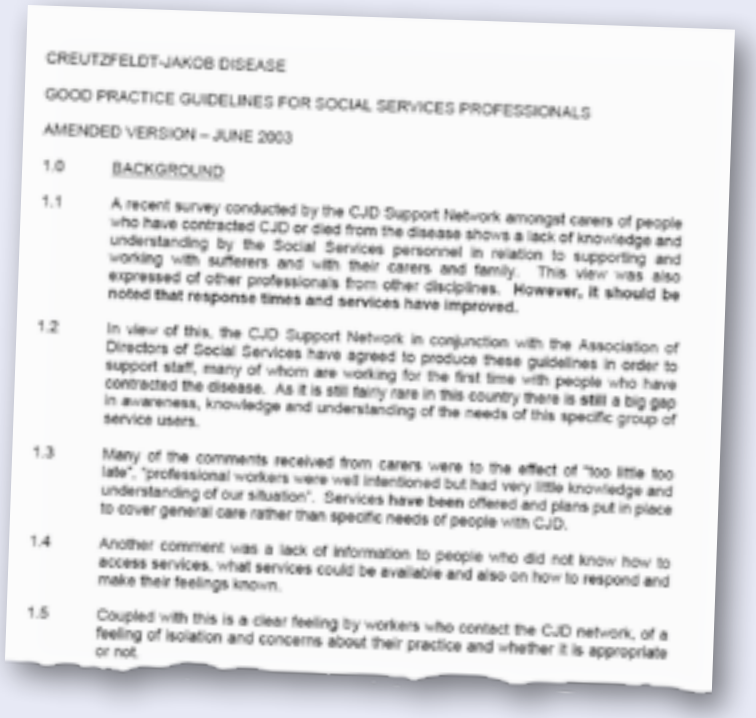
The good practice guidelines for social work professionals were written a number of years ago. They are still relevant and can be found on the web at www.cjdsupport.net (or see the link below).

The reasons for writing good practice guidelines were the difficulties many people experienced in accessing appropriate care and professionals also were feeling very isolated as CJD remains a fairly rare condition.

Social care professionals are asked to be aware of people's sudden change in needs, timescales and the need to monitor care packages on a frequent basis.

In terms of carers there is specific legislation and carers have a right to an assessment in their own right as carers. Carers can also receive services themselves, e.g. grants if they need to purchase something as a result of their function as a carer.

Currently there is new legalisation being proposed by the government, which is being consulted on. This will strengthen carers' rights. Another proposal is portable care plans; when people move areas they should be able to take the care plans with them.



CREUTZFELDT-JAKOB DISEASE

GOOD PRACTICE GUIDELINES FOR SOCIAL SERVICES PROFESSIONALS

AMENDED VERSION – JUNE 2003

1.0 BACKGROUND

1.1 A recent survey conducted by the CJD Support Network amongst carers of people who have contracted CJD or died from the disease shows a lack of knowledge and understanding by the Social Services personnel in relation to supporting and working with sufferers and with their carers and family. This view was also expressed of other professionals from other disciplines. However, it should be noted that response times and services have improved.

1.2 In view of this, the CJD Support Network in conjunction with the Association of Directors of Social Services have agreed to produce these guidelines in order to support staff, many of whom are working for the first time with people who have contracted the disease. As it is still fairly rare in this country there is still a big gap in awareness, knowledge and understanding of the needs of this specific group of service users.

1.3 Many of the comments received from carers were to the effect of "too little too late", "professional workers were well intentioned but had very little knowledge and understanding of our situation". Services have been offered and plans put in place to cover general care rather than specific needs of people with CJD.

1.4 Another comment was a lack of information to people who did not know how to access services, what services could be available and also on how to respond and make their feelings known.

1.5 Coupled with this is a clear feeling by workers who contact the CJD network, of a feeling of isolation and concerns about their practice and whether it is appropriate or not.

Good practice guidelines for social work professionals are available from www.prion.ucl.ac.uk/clinic-services/healthcare-professionals/professional-guidelines/ and from www.cjdsupport.net

Introduction to prion diseases

Prof Richard Knight, Consultant Neurologist at the National CJD Research and Surveillance Unit in Edinburgh

This talk gave a general introduction to prion disease suitable for anyone with no previous knowledge. It was accompanied by many pictures which cannot be reproduced here.

Background Biology

A basic understanding of the brain, proteins and genes is helpful.

The brain, like all body organs, is made up of cells. Brain cells include the electrically active neurones that communicate with each other at junctions called synapses. The complex functions of the human brain (which include movement, sensation, emotions and thinking) are made possible by the number of neurones (around 100 billion) and especially by the vast number of synapses (around 100 trillion). Some functions of the brain are localised to specific anatomical areas; for example, vision is organised at the back part of the brain. If disease disrupts the brain, then, depending on the areas affected, certain functions will be impaired.

Proteins are vital structural and functional components of the body and there are around 50,000 different human proteins, which are made following instructions from genes. Genes consist of a series of codes with each code unit relating to a chemical structure called an amino acid. The initial protein is therefore a string of amino acids but this string is folded into complex shapes and the shape of a protein is key to its particular function.

Gene codes can contain variations or errors, resulting in a change in the protein for which it is responsible. These changes can therefore affect the protein's function and thus lead to disease- these are termed pathogenic (disease-causing) mutations in the gene. There are also variations in the gene code that do not adversely affect protein function (although they can have other effects); these are found distributed in the normal population and are termed polymorphisms.

What are Prion Diseases?

These are progressive, presently untreatable, brain diseases that affect animals (eg scrapie in sheep) or humans and are, at the molecular biology level, related to an abnormal folding of a particular protein, the prion protein.

The normal protein (whose function is uncertain) is designated PrP^{C} and, in disease, it takes on an abnormal conformation designated PrP^{Sc}. When the abnormal protein is analysed in the laboratory, it is found to exist in two main types that have been labelled Type I and Type II.

As a protein's shape is important in its function, it is understandable that this change could result in malfunction and disease although the precise mechanism of the brain malfunction in prion disease is not completely clear. In humans, *PRNP* (on chromosome 20) is the gene responsible for the prion protein. Pathogenic mutations in this gene are responsible for genetic prion diseases.

There is a polymorphism in the gene (at code position 129) that, in itself, does not cause disease but which can modify the risk of getting a prion disease or affect the clinical picture of the disease if an individual is unfortunate enough to get it.

The polymorphism is such that any person is either 129MM, 129MV or 129VV (the M and V referring to the particular amino acid code and the letter doubling reflecting the fact that our cells contain two copies of genes – one from each of our parents). In the UK, about 50% of the normal population are MV, about 11% VV and the remaining around 39% are MM.

Prion diseases are also called Transmissible Spongiform Encephalopathies and the commonest form of human prion disease is CJD (Creutzfeldt Jakob Disease).

Prion Diseases have been classified into three main types according to their general causation: idiopathic (occurring sporadically, of unknown cause), genetic and acquired.

Idiopathic Human Prion Disease

Essentially, this is what is termed *sporadic CJD* (sCJD). It principally affects the middle aged and elderly. *PRNP* 129 MM individuals are more susceptible and MV individuals less susceptible to sCJD but the cause is unknown.

The typical clinical picture is that of a rapidly progressive brain disease with multiple problems but dominated by dementia (loss of cognitive function), ataxia (incoordination) and myoclonus (muscular jerking). The average survival from first symptom is from around only 4 months.

There are variations in the clinical picture. 20-25% of patients follow a less typical course; for example, slower progression with a longer survival, unusually young age at onset and specific isolated symptoms (such as isolated ataxia or visual symptoms) prior to the development of more general brain problems. These clinical variations are reflected in variations in the neuropathological findings and, to some degree, are associated with the individual's 129 polymorphism and the PrP^{Sc} protein type. As a result, it is current practice to broadly classify sCJD into subtypes such as MMI, MVII, VVI etc.

Acquired Human Prion Disease

These are diseases that are acquired as infections.

Iatrogenic CJD (iCJD) results from accidental transmission of other forms of CJD via medical or surgical treatments or procedures; the commonest instances relating to the use of cadaveric derived human growth hormone or human dura mater (a fibrous membrane that surrounds the brain and spinal cord). These to date have resulted in 226 and 228 CJD cases respectively worldwide. The clinical features of iCJD are generally similar to those of sCJD, apart from human growth hormone cases who generally present with a progressive ataxia.

Variant CJD (vCJD) arose from the contamination of human food with BSE from cattle. Dietary protection measures were instituted in 1989 and 1996. To date, in the UK, vCJD has not been seen in anyone born after 1989. Secondary, human to human, transmission of vCJD has occurred via blood transfusion and the use of blood products.

All cases of definite or probable vCJD (as defined by formal diagnostic criteria) to date have been in 129 MM individuals, but there is a report of an MV individual who is likely to have had vCJD and non-MM individuals are not thought to be immune from the illness. The age at onset in affected individuals is significantly lower than for sCJD; the average being 28 years. The illness also presents differently from sCJD, typically with psychiatric or behavioural problems and a slower progression with a survival time averaging 14 months.

Genetic Human Prion Disease

Genetic prion disease results from pathogenic mutations in *PRNP* (the prion protein gene); many different mutations have been identified that are passed on from parent to child in what is termed an Autosomal Dominant pattern; this means that any offspring of a mutation carrier parent has a 50% chance of inheriting it. The clinical picture of gPD varies, partly according to the particular mutation responsible. In some instances, it can closely resemble that of sCJD and, as a family history may be absent, genetic testing is required for absolute exclusion of a genetic cause.

Diagnosis

At present, an absolutely definite diagnosis of prion disease requires neuropathology and this is usually undertaken at post mortem; in rare instances, a brain biopsy in life is considered. However, a very confident clinical diagnosis can often be made, based on three elements: appropriate clinical features, the exclusion of other possible diagnoses and supportive investigations.

The EEG (electroencephalogram-recording of the brain's electrical activity), brain MRI (magnetic resonance imaging) and the CSF (cerebrospinal fluid) 14-3-3 protein test are all useful in sCJD.

In vCJD, the brain MRI is very helpful but the 14-3-3 test is less useful than in sCJD and the EEG generally unhelpful in supporting the diagnosis.

However, all of these tests are not totally specific for prion disease and show abnormalities that are not directly linked to the fundamental prion protein abnormality. Recent developments are promising with a blood test for vCJD and a CSF test (the RT-QuIC test) for sCJD that are currently being evaluated – both depending on the specific presence of abnormal prion protein. A tonsil biopsy can also be very helpful in suspected vCJD which depends on the specific finding of abnormal prion protein (PrP^{Sc}) in the tonsillar tissue.

Genetic disease is testable via mutation screening on tissues including blood. Iatrogenic CJD is diagnosed on the basis of previous exposure to a known risk factor.

Progress towards clinical trials in rapidly progressive prion diseases

Simon Mead, neurologist at the National Prion Clinic

As many readers know from personal experience, human prion diseases, such as Creutzfeldt-Jakob disease (CJD), are devastating and rapidly progressive conditions. At the NHS National Prion Clinic (NPC) our doctor and specialist nurse teams are dedicated to rapid assessment and follow-up of patients, providing information about the disease for doctors and patients, promoting early diagnosis, management of symptoms and care, and support for families.

Unfortunately however, this is not enough, as we are lacking any treatments to slow or halt the underlying disease. Aside from clinical care, the top research priorities of scientists and doctors at the Medical Research Council (MRC) Prion Unit and the NPC are to develop drug treatments for prion diseases and understand the best way to test these drugs in patients by clinical trials.

The following is a summary of my talk on this subject at the Annual General Meeting of the CJD Support Network in November 2012.

Although much progress has been made there are still several hurdles before we can begin a clinical trial. Do visit our website this year for updates www.nationalprionclinic.org. Or contact our office directly on 0203 448 4037 and our staff will be happy to direct you to the best member of the team to deal with your query.

Prion disease

The commonest form of human prion disease is sporadic CJD, which has an average life expectancy of only four months. These diseases are caused by a change in shape of a normal protein found on the surface of cells in the body known as prion protein. The prion protein changes shape into an abnormal or rogue form which sets off a chain reaction. In this reaction the rogue form of the prion protein (or 'prion') encourages more of the normal form to change shape into the abnormal form. Prions build up in the brain and eventually this leads to death of brain cells.

In some respects, prions are like germs such as bacteria or viruses; they can be spread from person to person by medical procedures, blood transfusions, and organ transplants, or can be spread from animals to humans in infected food. However unlike bacteria or viruses they cannot spread through coughing, sneezing or normal intimate contact between couples.

Antibodies

Antibodies are body proteins that our immune systems make to fight off infections from germs. Antibodies work because they bind very tightly to one of the proteins on the surface of the germ, and prevent it from multiplying. Over the last fifteen years the MRC Prion Unit has been studying special antibodies in the laboratory.

We have made a large number of antibodies which tightly bind the normal form of the prion protein and

prevent it from changing shape to the rogue form or prion. We have selected the most effective antibodies based on how much antibody is needed to treat mice infected with prions, how much antibody is needed to cure infected cells grown in the laboratory and how tightly antibodies bind cells infected with prions.

Mouse antibodies

In some mice infected with prions, treatment twice per week with the antibody can cure the infection when started at a very early stage.

The antibodies we used in testing are mouse antibodies. We cannot give these to patients as the human body would reject these antibodies because they contain mouse protein. Therefore we have converted the mouse antibody to a human form that should not be rejected by patients.

Testing on patients

We have also found a way to make enough of this antibody (now called PRN100) so that it can be used in patients.

Initially we need to test whether PRN100 might do more harm than good when given as single doses, and find the dose that results in the concentrations we want to achieve in the blood. Later we aim to test whether this dose actually works against the disease itself.

Randomised trials

Testing new drugs such as PRN100 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 of seeing if a new drug offers overall benefit to patients. Understandably, with such fatal 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.

Monitoring patients

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, in the National Prion Monitoring Cohort Study we have been building up a detailed picture of the progression of the different forms of the diseases and how useful various types of assessments are to monitor patients. Based on assessments of over 500 participants in the Cohort and interviews with carers and other family members we have developed a way to monitor patients called the MRC Prion Disease Rating Scale or MRC Scale. This scale records everyday activities like eating, speaking, walking, going to the bathroom and thinking skills like memory.

We now know that using the MRC Scale will mean we need to treat fewer patients in order to know if the drug is working or not than we would have if we simply looked at life expectancy. Not only will this speed up clinical trials, it also means that the results of the trial will likely be more relevant to patient and carer experiences of the disease.

Earlier diagnosis

Learning how to make the diagnosis much earlier is particularly important now as we 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. There is no point in recruiting patients to clinical trials in the final stages of the disease, when irreversible brain damage has occurred, because we will not able to work out if the drug has helped.

MRI scan interpretation

We remain frustrated that MRI brain scans are not being interpreted correctly around the country as this very effective diagnostic test could lead to earlier referral. Communication and education is important to get the message out about how to read brain scans correctly in such a rare condition. We have been doing this by developing our website, writing papers for medical journals and recently we have been working on an iPhone App ▪

Our story

Julie and Chris

We first noticed small changes in my wife Mary; she was a bit upset about my daughter Julie being pregnant and worried about a forthcoming hip operation for myself. She always said 'she did not feel right' and started misplacing things, glasses, purse, and other things, later she started to remove paintings and pictures off the wall saying they were hers and not mine. We went to the doctor who diagnosed it as stress and proscribed medication, eventually as things where not getting any better we changed doctors who agreed with the original doctor and thought it was stress.

Things at home were not getting any better. Mary seemed to lose sense of direction. She used to visit Julie (my daughter) who lived on the other side of the road but started walking in a different direction. Mary stopped making cups of tea and coffee and then meals, then washing and ironing, although when friends asked her what she had being doing she replied 'washing and ironing I have never stopped.' This was a very difficult time as I was struggling with my hip waiting for the operation and trying to manage the house

Eventually we went back to the doctor and explained the situation, who then referred Mary to the hospital for further tests. Again this was Mary, completely out of character, she used to run her hands through the doctors hair and touch his legs it sounds funny now but at the time was strange. The doctors organised a MRI scan, lumber puncture, electroencephalogram and dementia testing. Mary never questioned the tests, we never told her the truth, we just told her they where investigating her leg (which she was having trouble with) After approximately three months the doctors diagnosed sporadic CJD which I had never heard of. He explained the disease and informed us that no one had survived over 18 months and then informed us that she already had it for 12 months. We were devastated.

Things went downhill rapidly from then. She would not get dressed in a morning or ready for bed at night. We had to inform social services for carer help in the morning and evening. Mary went through an aggressive stage accusing me of hiding things, stealing money and having a girlfriend living in the house, she also gave the carers a hard time, refusing to get dressed or undressed. In this aggressive stage we found it easer to either agree with her or walk away rather than cause an argument. It was a very hard learning process.

I think Mary knew something was wrong. She started shaking at first in the hand and then the whole body, especially at night. This shaking could last up to an hour. Shortly after that she lost direction not knowing where the bedroom was or the toilet and at times did not believe we lived here. She also thought that everything on the television was real and often used to talk to it. I had to be very careful about what programs she could watch as she would get very upset especially at anything violent even although it was fiction. It would be no good explaining it was not real as she would not believe me Eventually she started to loose mobility and became incontinent she had a couple of falls where I had to call an ambulance out, it got to a stage where Julie and I had to make the difficult decision to put Mary in a nursing home

The progression of the disease was rapid she started having hallucinations, I think these were good and gentle visions as she often used to say 'can you see all the balloons'. When she entered the home she could walk with help, eat unassisted and within three months she lost all these facilities including communication and had to be fed by hand. Toward the end she lost the swallowing reflex. This is such a devastating and horrible disease which robs you of everything. At the best all we could hope when visiting was a smile or a laugh, but more often she was asleep. It was a very emotional time for both of us

Towards the end I don't think she knew who I was although I would like to think she did. Mary went into the home in February and passed away in May.

One wonders why this happens to such nice, kind and gentle people. I miss my wife terribly. She was not only a loving wife but also my best friend. I don't think I could have got through all of this without the help of friends and especially my daughter who was always there when I needed her. My only regrets are that I lost my temper on a couple of occasions. I always apologised to Mary to which she accepted and promptly forgot, although I felt bad for hours after. All this was before she was diagnosed with CJD, I now realise it was the disease and nothing else.

I hope this will help people and especially carers who are going through a similar situation and hope in the future a cure will be found for this devastating disease.

Ian Jonathan Cox

Ian Cox died of Iatrogenic CJD on the 23 April 2012 at the age of 46. Here his parents Janet and Barrie remember him, and how he contracted the disease

Ian was the third eldest of our four boys. We saw more of Ian than his other brothers (who had regular family environments), he was divorced from his wife, having two children, Jacob 13 and Katie 10. He worked as a chef in HM Prison service and was very highly regarded by his fellow chefs, staff and prisoners. His job involved training prisoners to obtain NVQs in catering. From letters we received from prisoners during his illness they spoke of him with affection and with great respect.

We would often text him to come and share an evening meal with us and he would reply 'oh, alright then', as if he was doing us a favour (but was really delighted to come and share mum and dad's fare and not face a lonely evening).

Ian was born on the 7 December 1965. He had a 'hare lip' which after three months was successfully repaired in Nottingham City Hospital. As time passed by we noticed he did not grow as other children in his age group did. Concerned, we took him to our GP who in turn referred him to Sheffield Children's Hospital. He was eventually placed on Dr Gordon's NHS programme to receive Human Growth Hormone (HGH) treatment. In the early days of treatment a district nurse would come three times a week to give him injections. Eventually, Ian injected himself and he gained in height over the 12 years period. He received HGH treatment from 1973 to 1985. The HGH project was terminated in 1985 due to batches being contaminated and Synthetic Growth Hormone was introduced from then on.

As a result of this HGH treatment, Ian contracted Iatrogenic CJD. He died on the 23 April (St. George's Day) 2012 aged 46.

Janet and Barrie have very kindly shared their diary outlining Ian's progression of the illness over a four months period. You can view this on our website at www.cjdsupport.net by clicking on carer's views *or by by telephoning the CJD Support Network office on 01630673993 or emailing gturner@cjdsupport.net.*

My experience

Mark Croll

I thought you may be interested in a recent incident I had when hospital for a back operation.

On the 13th January 2008 my father Robert Croll very sadly passed away from 'suspected' sporadic CJD. It was a very traumatic time not knowing what was wrong with him and the rapid deterioration from a healthy man over a short period of time was distressing for us all to watch. He did not undergo a brain biopsy or an autopsy at the time when he died due to family request, and so we were told it was a 'suspected' sporadic CJD case and we were all relatively happy with that outcome.

However almost five years on I recently had to undergo neurosurgery at Salford Royal Hospital Manchester for a discectomy of a prolapsed disc.

During my pre-op it was noted that my father had died from sporadic CJD and it was picked up that it was 'suspected' due to no brain biopsy or autopsy, nothing conclusive. I was then asked if I had taken a test to see if I was a carrier as there was no evidence to say it was not a 'genetic' type. It was said at the pre-op that they would need to take further advice from their local infection control service and I heard nothing more so thought nothing more of it.

On the day of surgery the question of CJD came back up again. It did not seem to matter when I restated that it was 'suspected' sporadic – the hospital still wanted to limit any contamination risk, which of course I do understand. Even though I was asked to come in at 7:00 am on the day of the surgery I was told not to be alarmed but I would now be last in theatre that day and that the surgeon joked that 'she had drawn the short straw' for my operation. It was also said that I was not to be alarmed but the surgical team would be taking extra precautions for example, would be wearing double gloves, disposal instruments would be used and there was talk of a £250,000 insurance risk if there was damage causing leak of spinal fluid – which I didn't fully understand. I was also moved from a ward of four people to a side room in the ward on my own. However I must stress that the surgery went well and the care was second to none, but it has raised a big question for me and possible future medical issues and importantly concern for my three children.

Stumbling across a previous CJD Newsletter (March 2011) I was further alarmed to read of people experiencing further discrimination by some insurance and mortgages companies due to a family connection with CJD and I have recently heard of cases of dentists refusing to treat patients (although not experienced this myself). It makes me angry that in this day in age people are still ignorant of CJD, especially in my case the medical profession and more importantly that they do not understand the impact this attitude has on family who through no fault of their own still continue to live under the shadow of CJD long after their loved one has passed away.

Editor's note

If you have experienced a similar incident and you want to share it please do contact us. By collecting all these examples it makes our campaigning stronger for greater awareness of CJD and the surrounding issues.

Infection prevention and CJD

Andrew Smith, Professor and Consultant Microbiologist, College of Medical, Veterinary and Life Sciences, Glasgow Dental Hospital & School, University of Glasgow

The trauma of a diagnosis of any form of CJD has many effects on family, friends and caregivers. Occasionally, problems can be compounded due to the unconventional nature of the infectious agent causing this disease, in particular the resistance of the prion agent to conventional methods of disinfection and sterilization which can lead some healthcare staff to have heightened concerns over cross-infection to other patients. This may lead to treatment and care delays or postponement due to worries over cross-infection issues.

Several family members have reported these difficulties to the CJD Support Network which have ranged from difficulties in accessing dental treatment to other forms of surgery. The Support Network has access to several healthcare professionals who are able to use their experience to help overcome these difficulties. From my own perspective, I have helped several family members of the network who have experienced difficulties in obtaining dental treatment. One of the reasons for these difficulties is that, thankfully CJD in all its forms is still quite rare and few clinical staff have experience of managing patients and family. In my experience, once clinical staff have had an opportunity to be reassured of appropriate protocols for care of patients without presenting risks of cross-infection, the situation is usually rapidly resolved to the satisfaction of family, carers and healthcare staff.

The guidelines and advice for dealing with CJD and infection prevention issues are overseen by the Advisory Committee on Dangerous Pathogens (ACDP) Transmissable Spongiform Encephalopathy (TSE) risk management sub-group and these guidelines can be found on-line at www.gov.uk/government/publications/guidance-from-the-acdp-tse-risk-management-subgroup-formerly-tse-working-group and are regularly updated. The advice is divided into four parts and several annexes, with topics ranging from pre-operative surgical assessments to information for funeral directors. Probably the most useful section for family and caregivers can be found in the Frequently Asked Questions, and in Part 4 on infection control in section 4.58 dealing with community healthcare.

To summarise the guidance on CJD and infection prevention: the prion infectious agent does not pass from person to person by routine social contact, so most forms of care and treatment do not require any additional precautions over and above that required for any other patient. Practical difficulties can arise when different forms of surgical intervention are required because of the difficulties in removing and inactivating the prion infectious agent from invasive medical devices. In its simplest form any surgical instruments that do not contact the central nervous system or lymphoid tissue do not need any special treatment after used on patients or family relatives. For other surgical procedures the advice now gets a little more intricate with slightly different protocols depending on the type of CJD and definition of 'at-risk' of CJD. These differences being due to the different tissue distribution of the infective prion protein in the different forms of CJD. For vCJD there is more infectivity in the lymphoid tissue outside of the CNS compared to other forms of CJD. The definition of 'at-risk' CJD is evolving as more research and information comes to light on the transmission potential of vCJD. The most up to date information on 'at-risk' status can be found on-line at in Part 4 in the ACDP TSE subgroup guidance mentioned earlier.

An important element of the infection prevention guidelines are that patients and family should never be discriminated against in provision of treatment or care. However, sometimes misunderstandings and resulting frustration can occur. To help overcome some of these issue that patients, family and caregivers experience, the CJD Support Network is able to call on myself to act as an expert advisor to healthcare teams. My day-to-day roles and responsibilities in Glasgow include acting as the lead Microbiology Consultant for medical device decontamination. Together with my infection control colleagues we have a wealth of experience in dealing with CJD issues that we can draw on to help other healthcare workers to resolve any issues that may have arisen.

I can be contacted through the CJD support network helpline and I am happy to provide advice and assistance on any infection prevention queries that may arise.

CJD Support Network

Management Committee 2012

Professor Richard Knight, Chair Richard is a Consultant Neurologist at the National CJD Research and Surveillance Unit in Edinburgh

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

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

Derrick Biggs
Derrick is Head of Provider Services, Adult Social Care, and lead officer for Association of Directors of Social Services in matters pertaining to CJD

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

Dr Andrew Smith
Andrew is a Professor and Consultant Microbiologist at Glasgow Dental Hospital & School, University of Glasgow

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

Stuart Thompson
Stuart's family are affected by Genetic CJD. Stuart has been tested and is gene positive.

Roger Tomkins
Roger's daughter Clare, died of vCJD

Dr Simon Mead
Simon is a neurologist working at the National Prion Clinic

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

Sandra Walshe
Sandra is a Registered General Nurse whose sister in law died of Sporadic CJD

Jean Bailey
Jean is a retired human biology lecturer. Jean's husband, Julian, died recently of sporadic CJD

Gillian Turner
CJD Support Network co-ordinator

Can you help us this year to raise money?

Due to the present economic climate it is very difficult to attract grants, so fundraising by members and their families is even more necessary to maintain the work of the network. If you have any ideas or you would like help to arrange a fundraising activity, please contact Gillian Turner (see below).

The CJD Support Network was established in 1995 by relatives of people who have died with CJD and is now recognised as the leading charity for all forms of CJD. Our aims are:

- To offer support to individuals and families concerned with all forms of CJD.
- To offer support to people who have been told they are at a heightened risk of CJD through blood and surgical instruments
- To provide emotional support for carers and to link families with similar experiences of all forms of CJD..
- To offer small care grants for families in need whilst caring for a family member with CJD.
- To provide accurate, unbiased and up to-date information and advice about all forms of CJD.
- To provide a national helpline on all forms of CJD.
- To promote good quality care for people with all forms of CJD.
- To promote research into all forms of CJD and the dissemination of research findings.
- To develop a public response for all forms of CJD

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 with 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

Contact: Gillian Turner, CJD Support Network, PO Box 346, Market Drayton TF9 4WN
Telephone 01630 673973 · Email gturner@cjdsupport.net · Website www.cjdsupport.net
Charity number 1097173