

NATIONAL TOLL FREE HELPLINE 1800 052 466 | www.cjdsupport.org.au

CJD Support Group Network (CJDSGN)

2008 proved to be a very busy year with sadly a lot of contact with new families dealing with a suspected diagnosis of CJD and ongoing issues with access to health care for those at risk of CJD.

The good news once again is that there have been no further cases of CJD associated with treatment of human pituitary hormones.

As many of you are aware the CJD Support Group Network was expanded in 2005 and our contract with the Department of Health and Ageing (DoHA) was extended to include support for families caring for, or coming to terms with the loss of a loved one to CJD, and other 'at risk of CJD' groups.

Although many years have past since many of us were on the human pituitary hormone program there are still issues that we face when accessing health care and the expansion has meant that for recipients needing advice and assistance the network is still very active and well connected and so in a position to offer what ever services are required.

We are very grateful that our funding was expanded to support this work and that funding has also been provided for two important projects

- Education and Awareness Program
- ACCESS project

Funding of the education and awareness program has given us the opportunity to produce our DVD 'Understanding CJD'. The DVD was launch by the Department of Health & Ageing in Canberra in February 2008 and since then has been used as a tool for our education and awareness program.

CONTINUED ON PAGE 2

2009 National CJD Conference

Plans for our 2nd National Conference and Gala dinner in Sydney on 16th May are now in the final stages. This conference is organised for our members, those at risk of CJD and our CJD family members, but will also be of great interest to health care professionals.

With many of us now frequently being asked to fill in screening questionnaires on CJD when we are admitted to hospital for surgery, infection control will be a topic of great interest as will research into sterilisation techniques and treatment options.

There will be plenty of opportunity to ask questions of our expert speakers and during the afternoon session we will hold round table discussions where you will also have the opportunity to ask your individual questions of the experts.

Members are invited to attend for free, and there is also no charge for any friends or members of the family who would like to accompany you. Health care professionals are asked to pay \$75 just to cover the cost of the day and a tax invoice will be issued. This does includes morning and afternoon teas and lunch.

You are all welcome to attend our Gala dinner that evening, and we have kept the cost to a minimum of \$75 which includes a three course meal, drinks and great entertainment.

We look forward to seeing those of you who have registered on the day, no tickets are being issued, your name will be marked off at the door.

If you have not already registered for the conference and/or the dinner and would like to, last minute registration can be done by ringing the toll free number – 1800 052 466.

NATIONAL CONFERENCE Saturday 16th May 2009

SESSIONS INCLUDE • Prion Disease • Research into treatment options • Work of the Australian National CJD Registry • Surveillance in Australia • Work of the National CJD Surveillance Unit in the UK • Prion Research in Australia • Prion Research in the UK • Work of the CJD Support Group Network • Caring for a CJD patient
• Revised infection control guidelines • Genetic counselling for families • Research Project '*Notification of risk of CJD*'

KEY SPEAKERS INCLUDE Professor Colin L Masters MD, Associate Professor Steven Collins, Professor Richard Knight, Professor Simon Hawke, Jennifer Cooke and Margaret Leitch

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2008 CJD National Conference

In May of 2008 the CJD Support Group Network held our first National Conference in Melbourne. This conference was unique in that it brought together individuals at risk and family members to participate in a mutual forum with world renowned experts on prion disease. It was well attended, with people coming from every state and territory in Australia as well as from New Zealand, Italy and the UK.

We invited speakers to provide information on many topics including the status of current research and experimental treatments, the management of CJD, infection control, genetic counselling and patient care. We were very grateful that everyone we asked to speak on the day enthusiastically accepted our invitation. Our members appeared very appreciative of the attendance and informative presentations made by several experts and researchers including Professor Colin Masters, Associate Professor Steven Collins, Dr Victoria Lawson and Dr Andrew Hill. We were also honored that Roberto Borgis, from our sister organization in Italy attended our conference and spoke as a representative of the CJD International Alliance at our gala dinner.

Planning for our 2009 conference in Sydney is well underway and we are again excited that Australia's leading experts and researchers are attending and that Professor Richard Knight from the Edinburgh Surveillance Unit in Edinburgh UK is coming to Australia specifically to present at our conference.

David

CONTINUED FROM PAGE 1 (COVER)

We acknowledge the assistance given in the production of the DVD by Professor Colin Masters, Associate Professor Steven Collins, and Ms Alison Body of the Australian National CJD Registry as well as several infection control experts. Our sincere thanks also go to our family members and recipients of human pituitary hormones (hPH) who agreed to be interviewed and for those who so generously provided footage of their loved one. We also worked very closely with DoHA to make sure that the infection control information was consistent with the revised infection control guidelines published in December 2007.

In the last 12 months we have been invited to present on about 50 occasions involving over 1200 health care professionals in all states and territories. The DVD has also proven to be helpful for new families seeking accurate information and has become part of our information package.

We are also grateful to Bernard Rupasinghe, Australian Dental Association (NSW Branch) Limited for his enthusiasm in assisting us with a national education program for dentists. We are looking at producing Dental Files using edited sections of our DVD that would reach dentists all over Australia to promote awareness of CJD and the revised infection control guidelines.

The ACCESS Project has been funded since July 2008 when Mandy Newton was appointed as Project Manager. Access for families and Carers to Counselling, Education and Support Services, has provided us with a much broader network of contacts and services to assist our members.

We are now in the planning stages for our second annual 'Understanding CJD' National conference and welcome members and health care professionals to attend.

Although our growth has been significant and our network is becoming known and respected for the work we do, with advocacy and support on behalf of our members, we can never be complacent as there is so much more that we need to achieve.

As I write a recipient of human pituitary hormones in the 1970's has had her spinal operation delayed by almost two months while it was decided who would fund an expensive instrument that will need to be quarantined for that patient's use only due to her risk status. As the screening questionnaires are becoming more common at admission to hospitals so then are the problems facing people at risk and family members of a CJD patient.

Our Western Australian families are still facing delays of over 12 months before receiving autopsy results to confirm a definite diagnosis of CJD and delays in diagnostic genetic testing are a common complaint.

Until the day that no-one has the cause to say 'I was treated like a leper', and when no further families have to face a diagnosis of this devastating disease with no hope of a treatment of cure, our work, and the work of like organisations around the world, will continue.

Suzanne

Access Project

I'd like to say a big hello to all the members of the CJDSGN. I have had contact with many of you in the past during the period of time in which I volunteered as the Family Representative for the CJDSGN as a point of contact for family members and individuals who wanted to talk to someone with first hand experience caring for someone with CJD. I lost my Dad to genetic CJD in 2004.

Prior to my involvement, the CJDSGN was funded specifically for individuals who were known to be at risk of CJD because of their participation in a government funded fertility program that was later found to have used CJD contaminated human pituitary hormones. A number of children who were treated for short stature with human growth hormone were also known to be at risk because of their participation in the program. The CJDSGN was established following an intense and long inquiry into the matter to ensure the participants of the program, now at risk of developing CJD, who were understandably enormously distressed, were well supported.

I see the establishment of the Family Arm of the CJDSGN, back in 2005, as an equally important and valuable development in the history of the CJDSGN and I am extremely proud of what has been accomplished since its inception. It gives me great pleasure to say that I am back on board with the CJDSGN in the capacity of Project Manager of the new ACCESS Project, which is a much needed and significant development designed to support family members of CJD patients.

CJD is unique and is the only disease that can be both genetic and transmissible, which can cause a number of difficulties for surviving family members, as many of you may have experienced first-hand. The obstacles faced range from a general lack of information and awareness at a clinical level, delays in diagnosis, insensitive approaches to families regarding autopsy procedures, delays in autopsy results, delays in genetic testing runs, and delayed access to health services. Suzanne Solvyns, who conceived the concept for the project, was inspired to assist families overcome these difficulties and set about designing it, adopting key features, where applicable, from services offered elsewhere, such as those used in the UK.

While the project is only in its very early stages, it will also deal with all aspects of support for families and individuals caring for a CJD patient, from day to day coping, a brokerage to social and health support services, the provision of the services of a medical consultant who has both experience in caring for a CJD patient and palliative care, and outreach visits when practical. Importantly, we will continue to maintain a close working relationship with the Australian National CJD Registry, ensuring the ACCESS project assists their surveillance outcomes. Families will be referred to the Registry, if not already in contact with them, ensuring their access to information regarding the processes for genetic testing and autopsy services are appropriately and accurately delivered.

I intend to touch base with all of our family members on an individual basis in the near future but if there is anything I can do to assist you in the meantime I am happy to help.

Mandy Newton
Project Manager CJDSGN

Survey for Families of CJD Patients

We are currently sending out a survey to each of our CJD families the results of which will help all families affected by CJD in Australia.

Only one survey is being sent to each family however, we would like to encourage the family member receiving the survey to discuss the responses to our questions with other family members or to pass the survey onto anyone in the family who may be in a better position to respond.

CJD is a rare disease, always fatal, very difficult to diagnose and is unique in that it is both genetic and transmissible. We are well aware of the problems that many families have encountered due to a lack of knowledge and awareness in the community, particularly amongst health care workers caring for your loved ones, and the ongoing issues many of you continue to face when accessing timely health care and genetic testing.

Many concerns have been raised with us either on the telephone or at our conference in Melbourne in May 2008. The CJD Support Group Network is committed to an advocacy role and determined to continue to work towards changes and improvements for families wherever possible.

By promoting awareness of CJD, working towards enhancing the knowledge of medical and social service professionals with our education and awareness program 'Understanding CJD' and networking with services that can provide support and assistance for families as part of our 'ACCESS' project, we hope to make a difference.

The assistance from as many families as possible with this survey is vital in enabling us to further identify problem areas. The only identifying information needed for our work will be the national state in which you reside, as this information is important in determining any problems unique to your particular state.

Once completed, the survey results will be published together with a background paper.

When published, this unidentifiable data will form part of a research project on patients and families with CJD and people with a genetic risk of CJD. The Cochrane Consumers and Communication Review Group, Australian Institute for Primary Care, La Trobe University in Melbourne, in collaboration with the CJDSGN, are seeking funding to conduct this research. The project will look at the research within Australia and internationally and will identify strategies to improve the experiences of people affected in different ways by CJD. This work will be the first of its kind internationally.

The assistance of each family is very important to us and to families who will unfortunately be affected by this devastating disease in the future.

Suzanne Solvyns

Catherine's Story

Dean's Lecture Series
Faculty of Medicine, Dentistry and Health Sciences
The University of Melbourne

DEBATES IN HUMAN GENETICS
THE BRAVE NEW WORLD OF GENETIC TESTING

Convener: Professor Richard Smallwood

DEBATE 2 – HEREDITARY DEMENTIAS: SHOULD TESTING BE ENCOURAGED? SHOULD TESTING OF CHILDREN BE ALLOWED?

A Patient's Account – Ms Catherine Grasso

1. Family History

My family is afflicted with Creutzfeldt-Jakob disease (CJD). Some 20 years ago an uncle died at age 51 from what was assumed to be sporadic CJD. His death, however, was followed years later by the death of another uncle and then, a year later, by the death of an aunt. They were both aged 67. A DNA sample from my aunt was subsequently sent to America, where a specific gene mutation for CJD was identified.

At this time my father, having been in general practice for 39 years, was toying with the notion of retirement. He was extremely concerned about the risk of inheritance for his children and grandchildren, and consequently underwent genetic testing in October 1996. We were shocked to learn that he possessed the mutation, and were devastated when, two months later, he manifested the initial physical symptoms of the disease. His deterioration was swift. In February 1997 he could no longer work, and, after a truly agonizing ordeal, he died in May 1997 shortly after his 67th birthday. Thus, of my father and his seven siblings, half have succumbed to CJD.

Since my paternal grandparents both lived to the age of 93 with no hint of the disease, our genetic mutation is said to have incomplete penetrance.

2. Genetic Testing

2.1 Why test?

The majority of people offered testing for fatal, adult-onset diseases choose not to be tested. I chose to know rather than spend a lifetime wondering anyway. After all, if I hadn't inherited the mutation, there would be no cause for concern. If, on the other hand, I had inherited the mutation, some contingency planning would be desirable.

Firstly, retirement age is a major consideration in such planning. If there is the possibility of an early death, I wish to organize my finances accordingly, and retire sooner rather than later.

Secondly, I wish to construct a living will. My father knew death was imminent, but we did not discuss it. It was simply too painful. Consequently, there will always be an element of uncertainty, even guilt, about the decisions we made for him in his final weeks. A living will would spare my family and friends such discomfort.

Finally, I wish to choose a hospice where staff are familiar with the disease and would hopefully be sympathetic to my needs.

Reproductive choices have no place in these plans, since I'm an independent 'career woman' having neither spouse nor children and no plans to change the status quo.

2.2 Testing Procedure

I was not apprehensive about genetic testing, as I had accompanied my parents to Dad's initial consultation and was familiar with the procedure. I had helped Dad research the

disease, and knew that my risk of inheriting the mutation was 50/50. I also knew if I did have the mutation, there would be a substantial probability - but not absolute - that I would later develop CJD.

Having read about the significance of dreams and believing in the 'wisdom of the body', I decided to question my body and dream the test result. Although a little far-fetched, I thought it was worth a try, as I had a 50% chance of getting it right! I subsequently had a dream which led me to believe that I did possess the mutation.

Thus, I presented for testing, which included extensive discussion of my probable reactions to either a positive or negative result. This seemed superfluous as my aim was simply to get the result. I didn't see the need for further counselling, and said as much. My opinion was respected, and a blood sample was subsequently taken that day.

2.3 On Getting the Result

I was a little anxious about getting the test result, but eager to know whether my dream was correct. The counsellor confirmed that I had, indeed, inherited the mutation. She also said clients often intuit the result.

I experienced a wonderful sense of euphoria. It seemed as though my life suddenly clicked into place, and all was as it should be. In retrospect, this was perhaps an over-reaction to days of persistent low-level anxiety. However, I was thrilled that my dream was realised, and felt I could trust my body, whatever happened. My inheritance forged a strong bond between Dad and me that, like love, transcends even death. The textbooks say that being at risk can make you feel 'special'. I've always known I'm special, but this confirms just how special I am - possibly one in 5-10 million!

2.4 Effects on Family

Of my 4 brothers, one has chosen not to be tested. Of the 3 who have undertaken testing, one brother has tested positive and one negative. The other, concerned about privacy and the stigma of disease, has declined the test result. He has subsequently become a committed vegetarian, and is convinced a healthy diet will reduce the risk of CJD. Although I think he's living in a fool's paradise, I can appreciate his need to maintain hope and some semblance of control in the face of uncertainty.

3. Follow-Up

It has now been two years since I was tested, and, at age 40, little has changed. My retirement fund is non-existent, but I am more discriminating in my spending. I have discussed my final wishes with family and friends, but there is nothing in writing. I don't obsess about CJD, but am possibly more attuned to my physical and mental condition. If, for example, I experience episodes of unexplained dizziness or increased forgetfulness, I fleetingly wonder whether this is the beginning of CJD. Common sense, however quickly prevails.

4. Final Words

In *Cannibals, Cows & the CJD Catastrophe* Cooke asserts

".....An individual stands more chance of winning lotto than dying of familial CJD".

Perhaps she's right. For years now I've bought Tattsлото tickets. Although I sometimes fantasize about how I'll spend my winnings, I'm not in the habit of waiting with bated breath for my numbers to come up. I've got better things to do.

We thank the Faculty of Medicine, Dentistry and Health Science at the University of Melbourne and Ms Catherine Grasso for permitting us to reproduce this article.

The Infection Control Guidelines for the Prevention of Transmission of Infectious Diseases in the Health Care Setting (ICG) Classical CJD Chapter 31

The ICG is a publicly available document that includes guidance on infection control strategy, environmental cleaning and protection of health care workers in health care facilities. The ICG cover all kinds of transmissible diseases including viruses (eg. HIV, Hepatitis C, Rotavirus), bacteria (eg. Staphylococcus, Tuberculosis) and other diseases (eg. cCJD, head lice). The ICG are designed to be used by all health care workers, such as hospital and clinic infection control staff, nurses, clinicians and dentists.

Chapter 31 of the ICG relates to classical CJD (cCJD), including Sporadic CJD, Inherited CJD and Iatrogenic CJD, but excluding variant CJD (vCJD).

During 2007, the CJD Chapter of the ICG was reviewed by an expert panel and revised to ensure a more concise format. The procedures and processes that infection control staff have to go through to evaluate a patient and determine the best way to protect them and subsequent patients has been simplified into a dichotomy for classification of transmission risk based on the tissues exposed during a procedure and the risk status of the patient. The precautions that have to be taken when a patient at risk of cCJD is identified have also been simplified to minimise the risk of transmission of cCJD and minimise occupational hazards for health care workers. The result is a more user-friendly ICG.

The entire ICG is currently undergoing further review by the National Health and Medical Research Council on behalf of the Australian Commission for Quality and Safety in Healthcare.

If you are undergoing a procedure, your healthcare provider can download a copy of the cCJD Chapter of the ICG free of charge from the Department of Health and Ageing website (www.health.gov.au). The CJDSGN or the State or Territory Health Department can also give further advice on infection control in your situation.

Article supplied by the: Communicable Disease & Health Risk Policy Section Health Protection Policy Branch Office of Health Protection Department of Health and Ageing

Note: There is a direct link to the revised infection control guidelines on our website

<http://www.cjdsupport.org.au/docs/chapter31.pdf> or for a hard copy contact Suzanne on 1800 052 466 or s.solvyns@cjdsupport.org.au

Medical in Confidence Letter

The medical in confidence letter issued to recipients of human pituitary hormones on a request basis was updated by the Department of Health and Ageing in January 2008 to comply with the revised infection control guidelines.

If you have previously requested a copy and did not receive the new version, or would like to request a copy please ring 1800 802 306. Screening questionnaires are becoming more common during hospital admission so this letter could

assist you.

This letter contained details of your risk status and is designed to be presented to health care professionals if you are undergoing a procedure. It explains some information on CJD, the current infection control guidelines and confirms that standard precautions should apply for your treatment except when high infectivity tissue is involved.

- Brain
- Dura Mater
- Pituitary gland
- Spinal cord
- Posterior eye (including retina, vitreous humour and optic nerve)
- Cranial and dorsal root ganglia; and
- Olfactory epithelium

Medical in Confidence Letter for Genetic Family Members

We are pleased to advise that we have met with the Chief Medical Officer of Western Australian, Dr Simon Towler and have discussed the availability of such a letter for family members with a genetic history in Western Australia. Dr Towler has given us a commitment that this will go ahead and once the process is completed we are hoping that the other state health departments will agree to provide a similar letter to genetic family members in their state. This letter will also be on a request basis only.

Oral Health Alliance

The CJDSGN is a member of the NSW Oral Health Alliance, which represents a number of NSW human service non-government organizations (NGO's), and provides a forum for discussion of oral health issues and the coordination of activities to improve access to dental services for their members. Convened by the Council of Social Services of NSW (NCOSS), the Oral Health Alliance recently conducted important research and produced a pivotal report to the state government, detailing key issues and areas of concern faced by the NGO's in question, and made ten key recommendations.

The CJDSGN was asked to provide a case study for the report and I was pleased to be able to provide a very real account of my own experience of discrimination and appointment related difficulties due to my CJD high risk factor. The recommendations published in the report concerning equitable and timely treatment address the need for appropriate information and materials being made to public dental facilities.

The data produced for the report is also highly suggestive that the NSW public dental sector is significantly poorly resourced resulting in ongoing psychosocial difficulties for individuals, such as those at risk of developing CJD. The Alliance has called for extensive investment of funding into NSW public dental services, which is believed will diminish waiting times and improve access for those affected.

Mandy Newton

High Infectivity Explanations

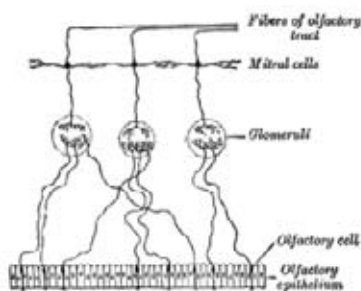
Members have asked for an explanation of the high infectivity tissues mentioned in the infection control guidelines to be included in our newsletter.

The following information is courtesy of Wikipedia <http://en.wikipedia.org> and we take no responsibility for the accuracy of this information. For more accurate advice we suggest discussing this with your doctor.

OLFACTORY EPITHELIUM



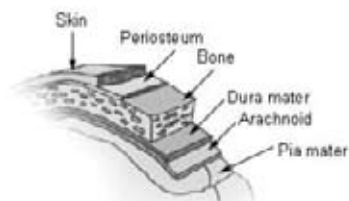
Section of the olfactory mucous membrane.



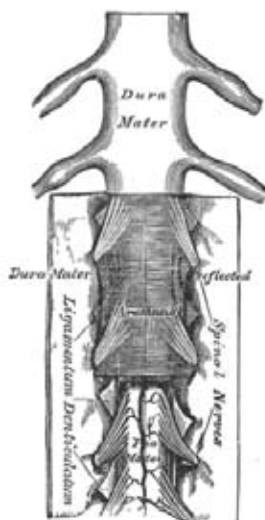
Plan of olfactory neurons.

The olfactory epithelium is a specialized epithelial tissue inside the nasal cavity that is involved in smell. In humans, it measures about 1 inch wide by 2 inches long (about 2 cm by 5 cm) and lies on the roof of the nasal cavity about 3 inches (about 7 cm) above and behind the nostrils. The olfactory epithelium is the part of the olfactory system directly responsible for detecting odors.

DURA MATER

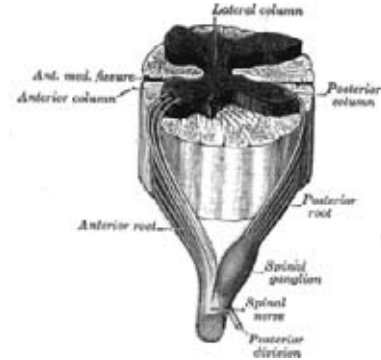


Meninges of the CNS



The dura mater (from the Latin “hard mother”), or pachymeninx, is the tough and inflexible outermost of the three layers of the meninges surrounding the brain and spinal cord. (The other two meningeal layers are the pia mater and the arachnoid mater.) The dura mater is not as tightly fitting around the spinal cord, extending past the spinal cord (at the second lumbar vertebra) to about the second sacral vertebra.

DORSAL ROOT GANGLION



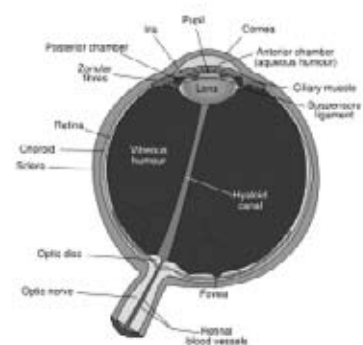
A spinal nerve with its anterior and posterior roots. The dorsal root ganglion is the “spinal ganglion”, following the posterior/dorsal root.

In anatomy and neurology, the dorsal root ganglion (or spinal ganglion) is a nodule on a dorsal root that contains cell bodies of neurons in afferent spinal nerves.

CRANIAL NERVES

Cranial nerves are nerves that emerge directly from the brain in contrast to spinal nerves which emerge from segments of the spinal cord. Although thirteen cranial nerves in humans fit this description, twelve are conventionally recognized. The nerves from the third onward arise from the brain stem. Except for the tenth and the eleventh nerve, they primarily serve the motor and sensory systems of the head and neck region. However, unlike peripheral nerves which are separated to achieve segmental innervation, cranial nerves are divided to serve one or a few specific functions in wider anatomical territories.

VITREOUS HUMOUR



Schematic diagram of the human eye.

The vitreous humour is the clear gel that fills the space between the lens and the retina of the eyeball of humans and other vertebrates. It is often referred to as the vitreous body or simply “the vitreous”.

RESEARCH IN AUSTRALIA

Professor Simon Hawke – Research Prion disease research

University of Sydney Medical Foundation Fellow (Jessie and Isabel Alberti Program Grant)

Professor Simon Hawke is a University of Sydney medical graduate whose prion research conducted at the Imperial College, London and published in Nature demonstrated for the first time that prion replication could be suppressed in vivo by immune manipulation.

After leaving Oxford, Simon was working at Imperial College, London and at the MRC Prion Unit at UCL. With successive MRC funding, he developed a panel of monoclonal antibodies to various conformations of the prion protein and showed that antibodies with prion-binding potential were the most efficient at suppressing prion replication in cell culture models. His group then went on to show in ground breaking experiments, that it is possible to turn off or suppress prion replication outside the brain and to prevent disease in living animals by targeting normal cellular prion protein with monoclonal antibodies (Nature: 471,80-83, 2003). In collaboration with Professor John Collinge he also developed a test for BSE that has since been commercialised by Roche Diagnostics.

Australia currently has relatively few research workers with expertise in the field of prion disease, calling in question our ability to satisfactorily deal with potential epidemics, such that have been faced in Europe from variant CJD. In August 2006, Professor Hawke and his team moved into purpose-built containment laboratories at the new Brain and Mind Research Institute at the University of Sydney. His current research is focused on using gene therapy to treat CJD, and he will discuss how this research strategy is developing at the CJD Support Group Network's National CJD Conference next week in Sydney.

Hill Laboratory – University of Melbourne

Andrew Hill is Associate Professor and Principal Research Fellow in the Department of Biochemistry and Molecular Biology at the University of Melbourne. After graduating from Victoria University in Wellington, New Zealand, he began working on Prion diseases in 1992, firstly as a research assistant and subsequently while studying for a PhD at Imperial College, London. His PhD focused on demonstrating the similarities in the prion strain responsible for BSE and vCJD and developed a classification system for human prion disease cases based on properties of the abnormal prion protein. As a post-doctoral fellow in the MRC Prion Unit, Andrew developed the tonsil biopsy test for diagnosing vCJD using the sensitive methods developed during his PhD studies. Further work from Andrew around this time also included demonstrating the presence of subclinical forms of prion disease in animal models. In 2000, Andrew came to Australia as a Wellcome Trust Prize Travelling Research Fellow and worked in the laboratory of Professor Colin Masters to gain experience in working with other neurodegenerative disorders such as Alzheimer's and Parkinson's diseases.

After a 12 month sabbatical back in the MRC Prion Unit in 2002, Andrew returned to Australia as an NHMRC Career Development Awardee and established an independent laboratory at the University of Melbourne. Here Andrew recruited a research team which now contains 13 members and primarily works on prion diseases, with associated work continuing in Alzheimer's and Parkinson's diseases. He is currently training a number of PhD students in prion research and in 2006 was awarded a Victorian Young Tall Poppy Award which has taken him to schools in Victoria and New South Wales to present to year 10-12 students,

and has had several students visit the lab for work experience. Andrew's laboratory is based in the Bio21 Molecular Science and Biotechnology Institute which opened in 2005 and his team works closely with the Collins and Lawson laboratories as well as the Australian CJD Registry which are also based at the University of Melbourne. Since returning to Melbourne, Andrew's research team has developed several novel systems for studying prion infection in cell cultures which has reduced the need for animal experiments and allowed us to speed up the research, being able to perform more experiments. Using these systems we are looking at how abnormal prion proteins travel from cell to cell and infect neighbouring cells. This is the basis for identifying potential therapeutics that can interfere with this process. We are also working on novel diagnostic approaches for prion infection and also study the basic biology of the prion protein to try and understand its role in the disease process in more detail.

Communicating with and Supporting People at Risk of CJD

Sophie Hill and Rebecca Ryan, with other staff from the Cochrane Consumers and Communication Review Group and the Australian Institute for Primary Care at La Trobe University in Melbourne, have been involved in research on how best to notify, communicate with and support people at risk of CJD where the risk has been acquired through medical treatment.

This research was commissioned by the Department of Human Services, Victoria. We're very pleased to have now completed a comprehensive review of the research on the experiences, needs and views of people at CJD risk. This review aimed to identify what people at risk need and would like, and how to meet these needs, by looking at research from Australia and internationally.

The review included over 40 pieces of research and literature. When assembled, the research identified many issues for people at risk – such as harms of being notified, difficulties accessing medical care, or problems arising from a poorly informed healthcare workforce. As part of the review, we have developed a model, based on the research evidence, outlining ways that notification, communication and support for people at risk could be improved – for example, by providing support to people at risk over time, or by educating healthcare workers, the public and the media about CJD. We hope that this model might help to improve future notification and support strategies for people at risk. A summary of this report will be available through the CJD SGN in the future.

We're also completing a Cochrane systematic review, which is a formalised summary of the best available evidence on ways to communicate with people at CJD risk and support them. The protocol (plan of the review) has recently been published on The Cochrane Library - available at www.thecochranelibrary ('Notification and support for people exposed to the risk of Creutzfeldt-Jakob disease (CJD) through medical treatment (iatrogenically)'), with access free within Australia. When the review is complete it will also be published on the Cochrane Library and so will be available to interested people nationally and internationally.

So far our research has focused on the needs and experiences of people at risk of CJD. There are also many issues for patients and families who have CJD; and those families with genetic CJD risk, yet the best ways to support them, provide information, ensure professionals are well informed and so on are not yet known. We are hoping to continue this research looking at ways to improve communication and support for these other groups of people affected by CJD and are in the process of seeking funding to continue this work.

For further information or enquiries about this research please contact Rebecca Ryan r.ryan@latrobe.edu.au.

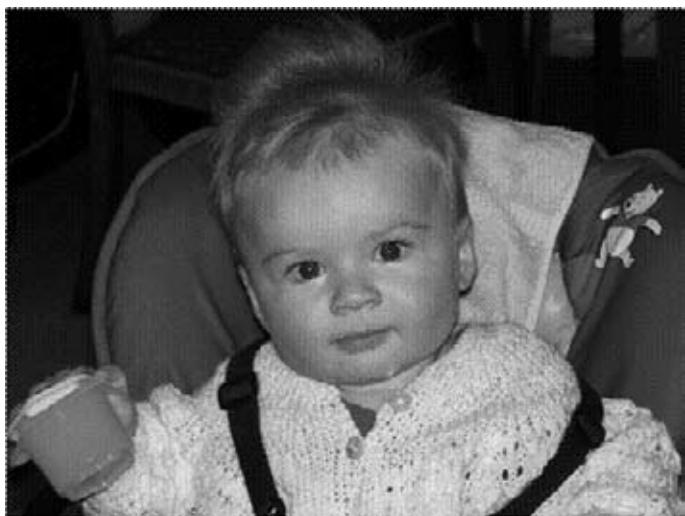
Notification of Risk in NZ

In 2007 the use of possibly contaminated instruments made the headlines in New Zealand and around the World. 43 people had been informed that the instruments used in their neurosurgery may have previously been used on a patient who was seriously ill and suspected of having CJD. These patients were informed that they were now considered to be at 'low but increased risk' of developing CJD. Several of the 43 patients were babies and children and in those cases it was their parents who received this news. This news was totally devastating for all the patients but the level of reaction and concern varied. The families with children involved were particularly fearful about the future of their children.

As there is no CJD Support Network in place in New Zealand the hospital had the responsibility of taking on that role. I met with hospital personnel and for them this was also a very stressful and upsetting time. Many of the families were understandable very angry and the hospital staff had to deal with that, provide information and protect a family whose loved one was dying from this devastating disease associated with a dura mater graft when only a child. How that family dealt with their subsequent loss and the amount of media attention is beyond comprehension.

The CJD International Support Alliance is committed to assisting newly formed networks around the world to grow and develop so if a network can be established in the future in New Zealand the Alliance's will be happy to provide guidance and share information.

Suzanne



I live in New Zealand and my dreams came true when my first baby was born but it was not long before my world was shattered with news I am still coming to grips with.

I would like to share my story and my feelings about how I felt when I found out that my precious baby was at increased risk of contracting Creutzfeldt-Jakob Disease (CJD).

In April 2007 I received a phone call that changed my life. I will never forget that day or what I was doing at that moment when I was told that my baby may be 'at increased risk of developing CJD' due to exposure to possibly contaminated surgical instruments. We were told that some of the instruments used on him during that operation had previously been used on a woman who three months later was to die of CJD, confirmed as iatrogenic or medically acquired CJD. She had received a dura mater graft as a child 23 years before

and after her surgery she did not recover and subsequently died.

The instruments had been through one basic sterilization procedure and there was no guarantee that all contaminated matter could have been removed.

I had never heard of CJD, I was totally naïve that such a revolting and devastating disease even existed.

That phone call turned my happy world into one of confusion, frustration and anger.

Why my baby, why us?

I researched for hours in an attempt to understand what I was being told. I felt total despair for my little baby wondering what might happen to him and what our future held as a family.

I cannot believe that this is such a hidden disease - no one I know had ever heard of it. Yes, I had heard of Mad Cow Disease but that was as far as it went. I had no idea what a prion was.

I then had contact with the International CJD Support Alliance (CJDISA), in particular Suzanne Solvyns, director of the CJD Support Group Network in Australia (CJDSGN) on the alliances' behalf and finally received information, support and assistance.

Suzanne Solvyns attended as a representative of the CJDISA and the CJDSGN at the first CJD group meeting in New Zealand and provided a wealth of information on how things were done in Australia and overseas.

My dream is to see a similar organisation established and available in New Zealand. If we could secure some funding I know that Suzanne Solvyns would assist me with that dream. There is no support for people at risk or families facing a diagnosis of CJD in New Zealand.

I would like to sincerely thank Suzanne - she has been a tower when I have been very scared!!!

I understand that scientists are working very hard to find a treatment or cure and I so hope they can as it is very worrisome when your child experiences symptoms that could be the early stages of CJD and there is no early screening test available to rule that out. If we were to face in the future a suspected diagnosis of CJD then we know there is no hope as there is no cure.

I, like many of you wish I had never heard the words Cruetzfeldt-Jakob Disease and I long to hear the words 'CJD cure discovered'. To any scientists reading this my plea is to please keep up the good work and as Suzanne said in her speech, as part of the CJDISA presentation at NeuroPrion 2008 in Edinburgh, "you are our future"!!!

My thanks also go to Dr Paul Brown (USA), Dr Michael Geschwind (USA) and Associate Professor Steven Collins (Australia) who have been supportive and offered assistance and personally taken time to help by answering so many of my questions.

A very worried Mummy

GLOBAL ASSOCIATIONS WORKING TO ASSIST ALL AFFECTED BY PRION DISEASE

CJD Foundation USA Family Conference 2008 – Washington DC

In July 2008 Suzanne Solvyns and David Ralston were invited to attend the CJD Foundation Family Conference in Washington DC. On Sunday morning we did a presentation on the work of the CJD Support Group Network in Australia and were pleased to report on the expansion of support for families as well as those at risk of CJD. We talked about our education program and plans for our very new ACCESS project. We were joined by Gillian Turner, the CJD Support Network, Minae Askaawa and other representatives of the CJD Support Network in Japan and Roberto Borgis and Raffaella Robello from the newly formed Italian group. Associazione Italiana Encefalopatie da Prioni (A.I.En.P), who also presented about their work.

Attending the CJD Foundation Family Conference and seeing the achievement of Florence Kranitz and her hard working team in the USA gave us the confidence to hold our own national conference for the first time in May 2008 in Melbourne. This was the first time that our members had the opportunity to hear from our experts We have developed an amazing number of friends, who have a common goal to support and help families in various way affected by CJD, as well as a network of contacts for assistance and advice.



FROM THE PRESIDENT'S DESK

By Florence Kranitz
President of the CJD Foundation USA

The sixth Annual CJD Foundation Family Conference and CJD 2008 was held on July 11–14 in Washington DC and according to the feedback we've received, this was our most successful conference yet!

Any worries we had about our families being overwhelmed by the science or the larger attendance disappeared the first afternoon during the scientific meeting (held for scientists to present their work to each other). We wound up with standing room only and most of the attendees were family members.

After the meeting, I asked a few family members what they took away from the experience, the answers, though phrased differently, were basically that although they didn't understand most of the science presented they felt that the isolation they lived with when CJD entered their lives disappeared when they entered the meeting room. It was, they said, an emotional experience to be surrounded by brilliant scientists who had devoted their careers to this terrible disease that had dramatically changed their lives, and yet few in their world had ever heard of.



For me, the conference memories have taken on a wonderful kaleidoscope-like effect which includes listening to fascinating research reports presented by brilliant scientists, hearing about new discoveries which may provide hope for those to come, and of being exhausted at the end of Saturday's sessions only to fully revive and delight, with everyone, at the wonderful surprise performance by Dr Valerie Sim an MD, PhD researcher from the NIH Rocky Mountain Lab. Dr Sim, also an accomplished concert violinist, played her 'fiddle' after our banquet. She delighted us all with her running commentary and her dexterity and skill which included playing while lying on her back and holding the violin under her body and never missing a note!!

Sunday's presentations, which include those of our CJD International Support Organisations, were extremely special in a different and more personal way.

The presenters are all friends I have developed through our mutual interest in speaking out to the world at large with one voice about CJD issues of concerns.

(This article has been reproduced with the permission of the CJD Foundation USA)

If you are interested in attending the 2009 CJD Foundation Conference in July, in Washington DC go to the website www.cjdfoundation.org

NeuroPrion

NeuroPrion was formed in 2003, and when the CJDISA came into being in 2006, it attracted the attention of NeuroPrion. That year Florence Kranitz was invited to speak in Torino Italy on behalf of the CJD Foundation USA and the newly formed CJDISA. Since then the CJDISA has been invited to speak in Edinburgh 2007, Madrid 2008 and in Greece in September 2009. NeuroPrion has generously dedicated a section of their website to the CJDISA and very supportive of the work we all do representing you our members. In 2008 NeuroPrion sent two representative to film the presentations of the CJD Foundation USA Family conference.

For more information on NeuroPrion visit:
www.neuroprion.com and to view any of the above presentation.

Note: The following article has been reproduced from the CJD Support Network newsletter 2008 with their permission and the permission of Dr Jens Schell.

NeuroPrion European Network of Excellence Science at the Service of Society

Dr. Jens Schell
NoE NeuroPrion – Scientific Manager Commissariat à l’Energie Atomique
www.neuroprion.com

NeuroPrion is the European network of excellence dedicated to research on prion diseases. It involves 52 public research organisations in 20 European countries and federates over 120 individual research groups corresponding to more than 90% of the leading researchers in Europe. NeuroPrion constitutes a first approach towards a new European Research Area open to collaboration with prion researchers from all over the world to protect human and animal health against prions and related diseases.

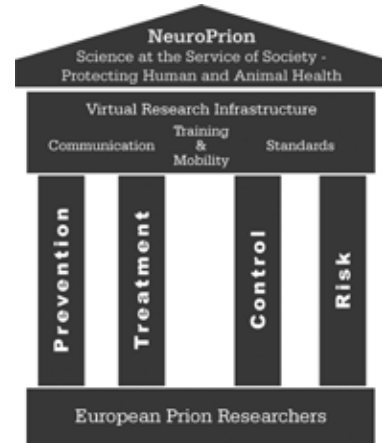
Since the appearance of the first cases of ‘mad cow disease’ in the 1980s, prion diseases have become a major problem for society both in Europe and worldwide, with important health and economic consequences. Prion research has already led to major advances in the understanding of these diseases, enabling European and national decision makers to properly manage the crisis and to reduce the negative impact on society. The efficient cooperation between science and regulatory authorities has resulted in better consumer protection and traceability of animal products. Thus, in parallel to a decrease in the total number of cattle and human cases, the public perception of the risk related to prions has also declined. Today, however, many questions remain. Fundamental research is crucial to understanding the processes of infection, replication and pathogenesis of these lethal diseases, which infect humans and animals with transmissible agents that are resistant to almost all classical decontamination procedures.

The recent discovery of previously unrecognised atypical cases of scrapie in sheep and atypical BSE in cattle in Europe and the US has raised concerns that previously unidentified strains of prions may pose new risks for public health. Furthermore, the compelling evidence that prion diseases

are transmissible through blood transfusion, indicates that research on prion disease must continue to progress in order to ensure the safety of public health.

Today, thanks to NeuroPrion, the combined know-how of the main research teams constitutes a unique core of expertise putting science at the service of society in order to prevent new prion crises in the future.

NeuroPrion – more than a flexible structure and more than research



Prion research is at the heart of the NeuroPrion consortium. However, research and research policy are changing in the global context and researchers must adapt to new developments. NeuroPrion has identified areas where greater coordination of research would be beneficial and others where novel, applied research is needed. Based on these assessments, NeuroPrion has implemented a joint programme of activity, which is built on the following four research pillars:

PREVENTION:

Decontamination
Diagnosis
Differentiation of strains

TREATMENT:

Development of New Drugs
Definition of New Targets

CONTROL:

Coordinated human Surveillance
Epidemiology/Surveillance animal TSEs
Tissues Bank (human and animal)

RISK:

Communication & Management
Assessment & Factors

NeuroPrion – Communication and exchange of information is the key to progress

Therefore, NeuroPrion’s communication includes not only to scientists, but is also directed towards authorities, stakeholders, industry and citizens. The annual prion conferences have become the major forum for knowledge dissemination within the field and cover an increasing range of scientific areas. Ever since Prion2006 in Turin, Italy, NeuroPrion has established a strong link to organisations dedicated to support CJD-patients and their families. It was the start of an Italian support group, which was officially founded in 2007 and joined the international CJD Alliance during the Prion2007 conference in Edinburgh. Florence Kranitz and Suzanne Solvyns gave a moving speech during the plenary session about the importance of working together globally as CJD patient and family representatives.

Furthermore, together with Inga Zerr from the German national CJD reference centre and Hans Kretzschmar from the Ludwig-Maxillian University NeuroPrion has organised an information day in 2007 for families of CJD patients. The specialists presented briefly the current situation in prion research and perspectives for future treatments, but the

main purpose was to allow exchanges between people who all have experienced or are experiencing the same situation. Babara Ludwig has presented passages of her book "Zum Weinen ist die Zeit zu schade" in which she describes how CJD has modified her and her husband's life in a sensitive but also in a true way. Gillian Turner from the UK CJD-Support Network presented the work and the importance of CJD support group for patient families by patient families. May be it was a first step towards a German group as well.

By combining the efforts of science, industry, policy makers and society, NeuroPrion contributes to the protection of public and animal health, to the avoidance of future prion crises, and to putting Science at the service of Society

The CJD International Support Alliance (CJDISA)

The CJDISA was formed by a group of grassroots non profit organisations who share a commitment to prion disease victims, their families, and those at risk of prion disease.

Since the first meeting held in Washington DC in July 2006 we have been pleased to welcome Roberto Borgis, Raffaella Robello and Angela Chilli as representative of the newly formed Associazione Italiana Encefalopatie da Prioni ONLUS (A.I.En.P.), Italy.

CJDISA was founded to fill the gap that exists on an international level and to assure excellence in the service to all individuals and families affected by prion disease.

The member organisations are dedicating to working together in meeting the educational, social, emotional, spiritual and practical needs of those we represent, and while we continue to operate independently we all benefit from the sharing and assistance we gain by belonging to an international framework.

The CJDISA was founded on the belief that by raising awareness, educating health care professionals and the public at large we can help to;

- remove the stigma surrounding CJD and prion disease,
- Promote Research activities around early detection, prevention, treatment opportunities, improved quality of life and ultimately a cure
- Increase proper utilisation of resources
- Promote continued access to care for those in active disease or those at risk.
- Assist by ensuring safe blood and food supply.



CJD International Support Alliance Meeting in Washington DC July 2008

Member Organisations:

Foundation – USA

CJD Insight – USA

CJD Support Group Network – Australia

CJD Support Network – UK

CJD Alliance – UK

CJD Support Network – Japan

A.I.En.P – Italy

Blood Donations

Recipients of human pituitary hormones and other people notified as being in an 'at risk' group are not accepted as blood donors in Australia. The children of the group, providing they fit other eligible criteria, can donate blood.

For families who have a loved one with suspected CJD or have lost a loved one to CJD, you will be permanently deferred from being a blood donor unless CJD is ruled out by an autopsy. This deferral only applied to first and second degree family members. This covers parents, children, brothers and sisters as well as cousins, aunts and uncles, niece and nephews and grandchildren who are blood relatives of the CJD patient. If your relationship is 3rd degree or more and you are deferred from donating the advice we have received from the Australian Red Cross Blood Service is to ask to speak to a doctor at the facility and you are attending to clarify your relationship.

If you are a family member with a known history of genetic CJD but you have undergone predictive testing with a negative result yourself then you are eligible to donate blood but will need to supply the ARCBS with a copy of your test results for your file in order to avoid future confusion.

Prion disease is known to be transmitted from variant CJD patients to blood recipients but there is no evidence that it can be transmitted by classical CJD. The ARCBS deferral policy is as a precaution to protect public safety.

Our Electronic Mailing list

This is available to our members and also for any health care professionals who would like to be added to our 'Interested Party' list.

If you are not already and would be happy to receive information and newsletter from us electronically please email; contactus@cjdsupport.org.au with your request.

Please note: Your name and email address will not be displayed or seen by any other recipients of information.

All About My Mother



Actress Carol Willesee was a sassy 58-year-old when her body and mind started to mysteriously fall apart. And few in the medical system seemed to care. Here, Amy Willesee recounts what happened next.

It is just over a year since two ambulance officers wheeled my mother, Carol, into the clatter and dash of a hospital emergency department at 9 o'clock on a Saturday night. She was sleepy and confused, dehydrated and having trouble speaking, but she was happy, she mumbled, because she was about to have a baby. "Make sure Dad [media legend Mike Willesee] knows he has to look after you while

I'm having the baby," Mum instructed. "Okay," agreed Lucy, my 29-year-old sister, as she slipped Mum's thin arms into a hospital gown.

Incoherent, delusional, no longer able to walk unassisted and in need of constant supervision, Mum was now unrecognisable as the strong, sassy 58-year-old of just five months earlier. And the scariest thing was, nobody could tell us why.

When my two sisters and I were little girls, we thought Mum was a princess. We'd watch her at her dressing table, laid out with its antique silver hairbrush and pretty perfume bottles, brushing her long golden hair. She was beautiful.

As it turned out, she wasn't a princess at all. She had too much spunk for that. She danced on tabletops, laughed with an almighty snort and was happiest either in her King Gees driving a tractor or having dinner with her girls. The best meals at Mum's were always running late because they were so full of dancing and silliness. The night I introduced her to my future husband, she interrupted dinner to lead us in a handstand contest.

She took great juicy bites of life and chewed like crazy. In her late 30s, having seen my youngest sister off to preschool, she embarked on an acting career, bagging a Sydney Theatre Critics' Circle Award for best newcomer before going on to fulfil her dream of playing Blanche in *A Streetcar Named Desire* at the Opera House in 2000. Later, with no farming experience, she took on a vineyard in the Hunter Valley.

By 2006, she was living on a gorgeous old dairy farm in Berry, two hours' drive south of Sydney, running a small herd of Murray Grey cattle. Then, one crisp June morning, she woke to find her legs tingling – a feeling like champagne running through her veins. For eight days the bizarre sensation persisted, along with an aching in her legs and back. She made an appointment with her GP, who prescribed something to relax her muscles. But her symptoms

started to multiply.

The Panadeine Forte stopped working. Her feet felt inexplicably cold. She couldn't sleep, couldn't stand bright light, couldn't remember things. She began a round of doctors' appointments, but nobody could figure out what was wrong.

By her 59th birthday in early August – just two months later – she couldn't walk in a straight line and had difficulty sitting due to the pain. Her eyesight was deteriorating and, when she swallowed, it felt as though she had golf balls in her oesophagus. Pretty much all she wanted to do was lie down and close her eyes. Of greater concern than the physical symptoms to us at the time was that she seemed like a different person. We couldn't say quite what it was, but something in Mum had changed. She was submissive and fearful, disconnected. She didn't seem interested in us. Didn't want to hold my new baby.

She had brain scans, spine scans, blood tests galore and everything was normal. Only she wasn't normal. Acquaintances who saw her in the street were stunned by her appearance: one thought she'd had a stroke, another a car accident. A tremor that enveloped her whole body would come and go. But it was in October, four months after her legs started tingling, that I began to get really scared.

Her rapidly deteriorating condition had forced her to move up to her apartment in Sydney, where Lucy and I could visit her every day. (Our other sister Jo, 27, was working in Ethiopia.) She had a live-in nurse. She had a wheelchair. A wheelchair. Holy crap. How had it come to this? Then I phoned her one day and asked how she was. "Good," she replied, not sounding at all good. "Jo was just here." By now, she had become difficult to understand, her thoughts jumbled, her words slurred. I thought I must have misheard.

"Jo was just there?" I checked.

"Yes, Jo was here. She's just left."

I felt sick. Jo was still in Africa.

Mum had had an episode of depression 10 years earlier, so there was enough of a history for the specialists to concur: in the absence of a physical disease, her symptoms were psychosomatic. They called it conversion disorder. It seemed bizarre that such severe symptoms could be triggered by an emotional event, but we were assured it happens. People can develop paralysis, lose their sight, their motor functions, even suffer seizures, and it's attributed to psychological factors.

So we asked Mum if anything was upsetting her, was there any trauma that could have triggered this. She kept insisting she was fine. She'd been happy before it started. She was distraught now, of course: no one would believe how sick she was. She talked about checking her will. Asked me if I wanted the farm.

"You're not dying, Mum."

She was quiet in the car as we drove her to a private psychiatric hospital, fidgeting pointlessly through her handbag, staring out the window. We hoped we were getting her the right help.

The place felt cold, her room all empty surfaces and bare walls. "We don't normally accept people in wheelchairs," one staff member said, annoyed. "Don't leave me here," Mum begged, struggling to form the words. It was horrendous. But we were acting on the only medical advice we had and we desperately wanted her to get better.

She took it badly, her body shutting down ever more rapidly over the next couple of days. At times, she became almost catatonic, just sitting, staring. When we weren't with her, she'd call, asking us to take her home. Sometimes you could hear her down the line trying to work out how to use the phone or how to talk. She was terrified, hallucinating. People were coming to get her. There were rats and snakes in her bed.

The doctor told us this wasn't usual for patients with conversion disorder. Typically, once treatment was started, they got better very quickly. But Mum was getting worse. His opinion now, he said, was

that Mum's symptoms were not consistent with a psychiatric illness. We needed to keep searching. With no referrals, no guidance, the hospital gave us the weekend to make alternative arrangements for her care.

By the Saturday night, though, she was in such a bad way the nurses called an ambulance and that's how she ended up in a hospital in outer Sydney. Maybe this would be for the best, we reasoned. She was in a public hospital now; someone would have to investigate her condition properly.

Part of me felt ridiculous typing mum's growing list of symptoms into Google each night after the kids were in bed. But no one else was offering us any explanations. No one else seemed to feel there was any urgency.

One night, I came across a case study from *The American Journal of Psychiatry*. It told the story of a 49-year-old woman, Ms A, who suddenly began experiencing severe lower-back pain that didn't respond to treatment. Soon she developed a body tremor and difficulty walking. All the tests came back normal and she was assured her symptoms would, in time, take care of themselves.

They didn't and, five months later, Ms A was diagnosed with major depression and conversion disorder and sent to a psych facility. Her deterioration was rapid: soon, she was confined to a wheelchair and had double vision, speech problems and difficulty swallowing. At which point she was hospitalised. It sounded exactly like Mum.

It got worse: she began squinting or closing one eye to compensate for her double vision. While standing, assisted, she was unable to place her left heel on the floor. Her legs grew rigid. Ongoing neurological testing revealed nothing; her aggressive treatment for conversion disorder was unsuccessful. Then, one night after dinner, Ms A was found in cardiopulmonary arrest, apparently having choked. Thirty-six hours later, an EEG declared her brain-dead. It was only at autopsy that the mystery of her condition was revealed: her brain tissue was riddled with tiny holes. It gave pathologists their answer: she had been suffering from Creutzfeldt-Jakob disease.

I Googled it. Creutzfeldt-Jakob disease (CJD) is a rare degenerative brain disease, marked by rapidly progressing dementia, co-ordination difficulties and personality changes. It is a "prion" disease, and occurs when usually harmless prion protein in the brain changes shape, setting off a chain reaction by converting other healthy prion proteins into the abnormal shape. This process essentially turns the brain into a sponge. Unlike the more famous variant CJD (mad cow disease), which is a separate disease not yet seen in Australia but thought to be caused by eating contaminated beef products, classic CJD – the one I was looking at – is thought to occur spontaneously and at random. In other words, no one really knows why classic CJD occurs, but if this is what Mum had, it wouldn't be related to her beloved cows.

CJD affects about one person in a million. Symptoms most commonly appear around the age of 65 to 70 – Mum was 59 – and it can be difficult to diagnose. Electroencephalograms (EEGs), MRI scans and lumbar punctures are the best diagnostic tools doctors have, although they're not always conclusive, particularly early in the disease. CJD can only be confirmed with a brain biopsy or at autopsy. There is no known treatment.

The day after the ambulance dash to emergency, I arrived at the hospital with my sister Jo, back from Africa, to find Mum lying motionless in a dull, soulless medical ward. A nurse told us Mum was under the care of a neurologist, Dr X, and a member of his team told me by phone that she'd have an EEG and lumbar puncture the next day. I hoped to meet Dr X and discover what was going on. I was worried they would just run their tests and discharge Mum, like everyone else had done. I wanted to make sure, since she could no longer properly communicate for herself, that I could give him a thorough history. I wanted to ask him what he was looking for, what he could do. What if we were all missing something and irreversible damage was being done? Mostly, I wanted to know that Mum was

a priority to him, that he was going to see this through to the end.

The nurses said he did his rounds in the morning. So I turned up early. I didn't see Dr X, but I saw his intern, either that day or the next. He turned up to check on Mum, young and neat, breezy and perfunctory, jotting down notes.

"What are you looking for?" I asked.

"We just want to exclude some things," he said.

"What things?"

"Prion disease," he said.

"Like CJD?" I prompted.

"Yeah. But it's unlikely." He finished writing up his notes and left.

Lucy, Jo and I took it in turns to be with Mum. Dr X remained elusive. I gave Jo a copy of Ms A's story to read. She rang me from the hospital one day soon after. "Mum's starting to squint. She's been closing one eye." Another time she reported that, during a physio session, Mum had started walking (assisted) on her toes. Her muscles were spasming so badly that she could no longer place her feet flat on the ground. "Everything that happened in that case study is happening to Mum," she said.

And still no doctor. Nor had the promised tests been carried out. "Tomorrow," they kept saying. It turned out to be five days before the lumbar puncture was performed; 11 before she was given the EEG. A week and a half in which Mum withered, in pain and afraid, waiting for tests to rule out a catastrophic brain disease. The lumbar puncture results would take at least two weeks, we were told. The EEG, however, revealed apparent changes in brain function. That might mean something – or it mightn't. If only someone could explain it to us.

In the absence of a human, I took to rifling through Mum's chart. There I saw an entry from the neuro intern: "Patient comfortable." Comfortable? Was he serious?

Nothing about Mum looked comfortable. She was "stiff" and "frightened", as noted by the nurses. "Anxious", "withdrawn", "easily startled". Her muscles were seizing ever tighter. When she held my hand she would squeeze so tightly, involuntarily, that she'd cut off my circulation. Her cheeks were hollow, her eyes haunted. One day I arrived to find her sitting with her right arm held above her head, motionless, like one of the patients from Oliver Sacks's book *Awakenings*. Her muscle contractions were too strong to pull down her arm. Even as her eyes closed for more and more of the time, and she slept more and more, her muscle spasms made her look perpetually tense.

None of this was written in her chart by anyone from the neuro team. And since Dr X was not seeing Mum's condition for himself – according to the chart he saw her just twice in the first three weeks – I don't know how he was keeping abreast of it. Where was he? We couldn't fathom it. We were in a foreign environment, frightened and in shock, watching as, for all we knew, our mum lay dying. It was happening right under the noses of doctors and nurses in a public hospital and, as far as we could see, no one was doing anything. It was left to us to discover her pressure sores, the size of gold balls. Her mouth ulcers and bleeding gums. Most days it didn't even look to us as if Mum's face had been cleaned.

Word trickled down that Dr X wasn't expecting it to be CJD. As far as he was concerned, we were told, she had depression. From what we heard, he wanted to hand her care over to a psychiatric team. We wanted to hear it from him.

Jo asked the registrar to set up a meeting with him. A television producer, she approached it like a job. She researched, looked up words that we didn't understand, wrote a list of questions. She asked our dad [he and Carol's marriage had ended 18 years earlier, but they remained on good terms] to join us. And, come Wednesday, she suited up. But Dr X didn't show up. His young registrar did his best to fill in. Maybe, Jo felt, contrary to his boss, he was leaning towards CJD, but it seemed he couldn't find the words. When Jo pressed him, asking whether we should tell Mum's family overseas that she was dying, he didn't explain that he'd seen others die from

CJD and that it wasn't something you forgot. He only whispered, "I can't answer that."

An attempt to get mum moved to another hospital and another neurologist failed, and by then the test results were due. It was the afternoon of November 27, 2006, when Jo called me at home from the hospital. She was crying so hard I couldn't understand what she was saying.

"Has she got it?" I asked.

"Yep."

"She's got CJD?"

"Yes."

The lumbar puncture results had come back positive. Only the day before, the psych team had been ready to take over from the neurologists. Instead, Mum was handed over to the palliative care team. We never met Dr X.

CJD is universally fatal. Nothing anyone did could have protected Mum from that.

But she had been robbed of her choices and her dignity. The chance to say goodbye. For months she had desperately wanted someone to acknowledge how sick she was but, by now, we didn't even know if she could understand her diagnosis.

With his Santa Claus beard and honest voice, our palliative care specialist was our soft landing. He had a gentle touch when he acknowledged that Mum appeared to be in pain and distress. Why had no one else noticed? He started her on sedatives and morphine, the first time in 25 days she had been given any pain relief stronger than paracetamol. The tension in her body, the distress on her face, finally, mercifully, melted away. Her beauty returned.

Mum was transferred to Sacred Heart Hospice in the inner city, where Lucy, Jo and I sat with her for her last four days, among the gardenias, hydrangeas and lavender that filled her room. Dancing and silliness interspersed with the tears. Until it was time to say goodbye.

In many ways it was like a birth. The births of her children were the happiest moments in Mum's life, especially her youngest, Jo. That was an easy birth, at dawn. Afterwards, her baby at her breast, she ate marmalade on toast and watched the sun come up.

Thank you to the Sterilising Research and Advisory Council of Australia SA Inc

I would like to thank and acknowledge, on behalf of the CJDSGN, the Sterilising Research and Advisory Council of Australia SA Inc (SRACA SA inc.) for their generous donations to research in Australia.

The SRACA SA Inc. has committed donations of \$10,000 to the Carol Willesee Trust Fund established at the University of Melbourne to fund PhD students working in the prion research, as well as \$10,000 to assist the Lawson Laboratory headed by Dr Vicki Lawson at the University of Melbourne. Vicki's laboratory is doing research into sterilisation techniques for prion reduction for surgical instruments.

In August 2008 I was invited to do a presentation on CJD at the AGM of the SRACA SA inc. by Dora Gadeleta, who is president of the SA branch. Dora mentioned that their AGMs were very popular and well attended by the members. Mandy Newton was with me in Adelaide at the time and Dora promised us an interesting but enjoyable evening. We were not disappointed! As the meetings are sponsored by Meile, we arrived at the Miele showroom where we found the SRACA SA Inc. members, some had come from far and wide, chatting and enjoying food samples being prepared by the

Meile representatives. We were made very welcome and with a glass of wine and some testing of the delicious food sampling we had a chance to finally meet Dora and many of the others. Following on from the AGM we gave our presentation and we were delighted with good feedback and interest.

Dora thanked us and gave us an envelope saying it was a little thankyou. It was actually a cheque for \$1000, which we gratefully accepted as a donation to maintain our fundraising account.

As we guarantee that all donations received by the CJDSGN will go directly to research into prion disease we are constantly fundraising by the sale of our CJD bracelets and donations for our DVD, to maintain the cost of running the fundraising account to cover cost such as credit card fees.

On the way back to the hotel Dora asked about the Willesee funding raising event that had been on in Sydney the previous night. She had read Amy's very touching article and was impressed by the effort of the family to raise money for research.

I told her about Dr Vicki Lawson's research work on sterilisation techniques at the University of Melbourne and promised to have Vicki send her information.

It was with great excitement and appreciation that Vicki Lawson, the Willesee family, the CJDSGN and the University of Melbourne received notification that the committee of the SRACA SA Inc. had decided to make their most generous donation.

A special thanks to Dora, Bronwyn and all members of the SRACA SA Inc. Unlike many other diseases more prevalent in the community, funding for CJD research is extremely limited and donations made by our families and their friends and organisations such as this make a big difference in providing research opportunities. Suzanne

Fundraising for Research in Australia

Congratulations to all our families who have helped to contribute to funding for CJD research in Australia.

To date \$174,000 has been donated via our fundraising account directly to prion research in Australia. This is due to the efforts of so many of you since our first fundraising dinner in November 2006 and also aided by the support of the SRACA SA Inc.

At our first National conference in May 2008 in Melbourne we acknowledged Ronnie Donnellan, his partner Loz and his support team for the amazing effort and money raise during his 'Riding of Dad' event. Ronnie rode his bike from Alice Springs to Uluru and back, raising \$11,000 and much awareness of CJD in Alice Springs with his spirit and enthusiasm in the name of his Dad Ronald Donnellan who died of CJD in 2006.

We also acknowledged the Kelly and Ferry families who, in a wonderful united fundraising effort that spread over several states, raised almost \$30,000. The 'Hell's Angel team' consisting of 80 family and friends ran and walked in a half marathon event on the Gold Coast in July 2007 to remembrance of Helen Ferry who died of CJD in 2005.

We mentioned many others lost to CJD whose families and friends have made generous donations in lieu of flowers at their funerals. All of these donations add to the tremendous difference our combined effort is making.

Marlene Hutchins, who lost her husband Phillip to CJD, donated the money from the sale of Phillip's caravan to CJD research as she knew that is what he would have wanted that. She also very generously contributed so much to our DVD with some very personal footage.

We received at the dinner following the conference a special message from Professor Glen Bowes, Associate Dean, Faculty of Medicine Dentistry and Health Science, The University of Melbourne.

Professor Bowes words really reflect the importance of our combined efforts.

"As a health professional and researcher it is always such a privilege to work with and alongside support groups like the CJD Network. We are inspired in our work by the courage and resilience of those with medical conditions, their families and friends. We learn from your experiences and respect greatly the very special cooperation you provide in the conduct of our research programs. We are humbled by the generosity of those who raise and donate funds to support the ongoing research effort. The funds contributed by philanthropy to research are of very special significance. These funds mean that as researchers we can really be creative and take the risks needed to forge new frontiers of discovery. By way of contrast the funds from government and other research funding agencies are usually directed at the "safe bets" and knowledge is extended in small steps. The research funds provided by your donations give our researchers the chance to make the breakthroughs and enable 'big' ideas to be explored. Furthermore, we are able to leverage donated funds in a way that increases the likelihood of success in winning additional funding from nationally competitive granting agencies. My most sincere thanks personally and on behalf of our faculty."

Many families and their friends continue to make very generous donations in memory of those they have lost to CJD and in August 2008 many of us enjoyed a wonderful film preview and fundraising event organised by Amy Lucy and Jo Willesee in memory of their mother Carol who died of CJD in 2006. The film preview and cocktail party raised \$40,000 and each has contributed personally to raise that amount. The 'Carol Willesee Trust fund' of \$100,000 has been established to contribute to and support PhD students working in the field of prion research at the University of Melbourne.

We have already received some very generous donations in support of a PhD student to work with Professor Simon Hawke at the University of Sydney.

One donations came from a caller named 'Simone' who read Amy Willesee's story and has contributed and wants to continue to contribute by donating the interest that she receives from a term deposit. She says she has no personal involvement but was very touched when she read Amy story. Her donations will go to support the work of Professor Simon Hawke.

I have been personally touched and have felt very honoured to have been part of and involved in many of your events.

We would also like to acknowledge other families, including the Tehan family, who have made substantial donations directly to the Australian National CJD Registry to assist the research team in Melbourne.

I was privileged when visiting the University of Melbourne with Amy and Jo Willesee to be taken on a tour of the research laboratories by Dr Vicki Lawson. Many of you ask if your donations are used on prion research only, what exactly

they contribute to and that day we actually saw some of the equipment that your donations have helped to purchase.

Dr Vicki Lawson kindly agreed to write an article detailing exactly how the funds raised through the CJDISA, the Tehan Trust and other family contributions are assisting research in Australia.

Suzanne

Research Teams at the University of Melbourne acknowledge your support

Scientists within the prion research group at the University of Melbourne, headed by Associate Professor Steven Collins, Dr Andrew Hill and Dr Victoria Lawson are grateful for the generous support we receive from the CJD Support Group Network (CJDSGN).

Donations from the CJDSGN and other donations received by the Australian National CJD Registry have been used to purchase equipment for prion (CJD) disease research. Due to the transmissible nature of prion diseases our research is conducted in a contained facility where we have restricted access to high-end instrumentation. Your donations have facilitated the purchase of equipment for our contained facility, greatly relieving these previous restrictions and enhancing our research capacity. These equipment purchases have included

- FLUOstar multifunction plate reader which enables quantitation of light activated dyes,
- LAS-3000 imaging system to enable digital capture and data analysis,
- Fluorescence microscope to enable the observation and digital capture of cellular components that have been labelled with a light activated dye.
- AKTExpress and probe sonicator to enable the production and purification of recombinant proteins.

In addition to enabling the direct purchase of equipment donated funds have also been used to leverage funds through competitive grants. For example our fluorescent microscope was funded with donation money and a grant from the Brain Foundation.

The equipment is fundamental to undertaking new avenues of investigation. For example, using the FLUOstar, LAS-3000 and fluorescence microscope we recently conducted a preliminary project to investigate how individual brains cells may be ying in a CJD affected individual. This data will now provide the preliminary data for a competitive grant bid to the Australian Government and is an example of where your support has not only increased our understanding of CJD but has also helped facilitated funding from external granting bodies.

In addition the new PC2 laboratory occupied by the Collins and Lawson groups has been refurbished by the Department of Pathology. Equipment will be purchased for this lab using donated funds.

Thank you again for your wonderful support.

Victoria Lawson, PhD
CR Roper Senior Research Fellow
The Department of Pathology
The University of Melbourne



Raising Money to Support Our Fundraising Account

Our 2008 CJD bracelet has proven very popular and will shortly be available to purchase on line at www.cjdevents.org



Our heart and star bracelets are available in green (shown), purple (shown), black/grey, red, pink, white, blue.

Cost: \$20 each + \$2 postage & handling

To order: Telephone 1800 052 466 or email [Suzanne s.solvyns@cjdsupport.org.au](mailto:Suzanne.s.solvyns@cjdsupport.org.au)
– credit card facilities available.

The CJDSGN acknowledges the funding provided by the Department of Health and Ageing which enable us to produce and distribute this Newsletter. The CJDSGN asserts that apart from providing funding, the DoHA is not involved in the compilation of this Newsletter and the views expressed herein.