



INHERITED PRION DISEASE FACT SHEET

**Frequently asked questions
for people and families
affected by genetic CJD or
other genetic forms of
Prion Disease in Australia.**

This information has been prepared by the
Australian National Creutzfeldt–Jakob Disease Registry (ANCJDR)
for the CJD Support Group Network (CJDSGN)
and updated in January 2009.

Inherited Prion Disease Fact Sheet

The enclosed information is for people who fall into the high infection control risk group for Creutzfeldt–Jakob disease (CJD) and inherited prion diseases; either you have been tested and confirmed as carrying a mutation that may cause you to develop CJD or another form of prion disease, or you have not been tested but remain at risk of inheriting a genetic prion disease. The genetic prion diseases include familial CJD (fCJD), Gerstmann Struassler Scheinker (GSS) or Fatal Familial Insomnia (FFI). The infection control guidelines have been established by the Commonwealth Government and Health Care advisors to ensure the highest levels of safety to minimise the risk of transmission of prion diseases in the health care setting whilst maintaining optimal care for at risk persons.

1. Can I give blood?

No, the Australian Red Cross Blood Service (ARCBS) will decline your donation. If you have a family member who had a negative result to prion protein gene (PRNP) testing, they can be a blood donor. The ARCBS will, however, require a copy of the test results to be recorded on that donor's file. The ARCBS is currently revising the protocol to ensure that future generations of a genetic family member with a negative PRNP test result will also be able to make blood donations.

2. Can I donate organs?

It may be possible for you to be an organ donor, but in some states and territories your donation may be declined. Check with your local organ donation centre for their specific guidelines.

3. Why would I be declined as a tissue donor?

Prion disease is transmissible which means it can be passed from one person to another in particular circumstances including through use of tissues such as cornea grafts. The health care system has established infection control measures to minimise the risk of any transmission of prion diseases between patients.

4. Are there other infection control measures I need to know about?

Yes. The infectious prion protein can be carried in the body for some length of time before the illness shows itself. If you are at risk of inheriting a genetic prion disease mutation, you may carry the disease causing prion protein in your body while you are healthy but have no sign of the illness. Those who are at risk of inheriting or are known to carry a mutation associated with genetic CJD fall into a higher infection control group when having medical care. This means there is a risk of transmitting the disease to another patient when you have invasive health care. Invasive health care involves high infectivity tissue and is defined as some forms of dental surgery, neurosurgery, posterior eye surgery and some fibroscopic endoscopy procedures (for example ventriculosopes) where there is contact with brain tissue.

There are a small number of people recognised who have died from CJD after an exposure to medical instruments previously exposed to patients with CJD.

The number of people who have died of Iatrogenic CJD (Medically acquired) that is believed to have occurred after a recognised medical transmission during a procedure adds up to approximately 1–2% of all prion diseases worldwide.

For more information on invasive health care involving high infectivity tissue where some special precautions may need to be taken refer to the revised version of the National Infection Control Guidelines (December 2007) <http://www.cjdsupport.org.au/docs/chapter31.pdf>

5. Why aren't routine hospital infection control practices and sterilisation techniques enough to manage the risk of CJD infectivity?

Your routine day to day care in a hospital setting will be exactly the same as for other patients. However, when you have invasive procedures a higher level of precaution and sterilisation practice is needed for the infection control management of equipment used in some procedures. Routine sterilising temperatures and solutions are not strong enough to break down the disease-associated prion protein, which means equipment may still carry the prion protein and be potentially infectious after routine sterilisation. Consequently, higher levels of sterilisation are used for procedures involving for tissue classified as higher risk tissues. Unfortunately, some specialised surgical equipment cannot routinely tolerate these more stringent sterilisation techniques and planning will be needed to manage the equipment. Good quality disposable instruments are now available for the many procedures.

DISCLOSURE

6. What should I tell my doctor?

Your General Practitioner (GP) or local doctor should be advised about your genetic status and risk of inherited prion disease. Your GP will often be able to assist you in navigating the health care system. The higher infection control risk means extra care and planning is required for some invasive medical procedures. The CJD infection control measures and your special needs may not be routinely understood by all health care practitioners but your doctor is well positioned to guide you with appropriate referrals when invasive procedures are needed. They may also require education about inherited prion disease and the infection control guidelines to be able to assist you. The Australian National CJD Registry (ANCJDR) is available to provide further information. The CJD Support Group Network (CJDSGN) is happy to provide your GP with a copy of the DVD '*Understanding CJD*'.

7. What if I need to go to the dentist?

Your dentist does not need to be advised of your risk status for any routine dental care. However, it may be advisable to inform your dentist in case you may require invasive dental surgery in the future. Routine dental procedures include cleaning, routine checkups, fillings, extractions and root canal therapy.

For high risk tissue procedures, your dentist would need to alter the sterilisation and management of dental equipment used. For high risk dental procedures a referral to the dental hospital in your state or territory is likely to be necessary, to ensure correct equipment infection control management.

8. If a medical practitioner declines to treat me, what can I do?

The doctor may not understand CJD or the infection control guidelines, but regardless, any private health care practitioner is able to decline to treat a patient. It is recommended that you find a GP or a specialist who is willing and able to work with you, and if you need further assistance contact the CJDSGN who liaise closely with National and State health authorities, or the ANCJDR.

9. If my doctor is referring me to a specialist, can they disclose that I may have the CJD gene or do I need to tell them? In general, which specialists need to be informed?

Yes, for some treatments or investigations your doctor will need to disclose your genetic result or risk status to ensure appropriate infection control measures are taken. You can ask them not to disclose, but depending on the reason for referral this information may need to be passed on to assist other doctors evaluate your illness, or to interpret investigation results, or in the undertaking of invasive procedures. It is appropriate to disclose your risk group status if you are having invasive procedures.

10. Are there situations in health care when disclosure is not needed?

Yes. Some situations do not need disclosure of your genetic status or high infection control risk group. Talk with your GP as you organise your health care, to ensure you both understand which situations warrant disclosure. An example of when disclosure is unnecessary could include having a chest x-ray, a CT scan, a blood test or a pap smear. In contrast, disclosure of your risk of CJD would be appropriate if you were having neurological symptoms and referred to a neurologist for evaluation and further investigations.

11. What if I need an operation?

You will be able to obtain your operation. Alternative techniques and equipment may be needed to ensure some procedures will proceed. With advanced knowledge of your risk status planning can be done so that you are not disadvantaged.

12. What if a procedure is needed in an emergency?

In an emergency, if there is no next of kin to disclose your genetic status information and you are unable to (and do not wear a medi-alert), your treatment and surgery will proceed regardless. It seems appropriate to disclose your genetic result ASAP after the procedure, to allow the doctors and health care staff to manage the equipment used as soon as they can. The aim of disclosure is to minimise any risk of transmission to patients that are treated after you and may have the same equipment used during their surgery or procedure.

13. If I am involved in an accident, what should I or my family disclose?

We recommend you or your family disclose your genetic status information if invasive procedures are needed. The handling and sampling of blood is managed in the same way for all patients, along with the management of airways for breathing, intravenous fluids, etc. Your care can be managed by the same approach used for all patients, with routine infection control measures used to minimise the risk of more common infectious diseases like hepatitis or HIV. The basic approach used is called “universal precautions”. This means in an accident or emergency situation, basic health care interventions used for all patients will protect health care staff and other patients. However if invasive procedures are needed, particularly surgery on higher risk tissues for example, disclosure of your genetic status is recommended to an attending doctor so further infection control measures can take place.

14. Who should I disclose this information to?

This is your private information, to be disclosed only when you choose to or when necessary. You can keep this information private, but there are some circumstances when disclosure is needed.

(i) New Life Insurance Policies:

You are obliged to notify your Life Insurer if purchasing a new Insurance Policy after you learn about your genetic status or a family history. If you choose not to disclose at purchase, a later claim will be rendered void. If you have already purchased life insurance, check your policy, you may need to advise them of you change but it cannot alter the policies and conditions under which you purchased .

(ii) Health Care Insurance:

Health Care Insurers do not need to be informed of your genetic status, but you may need to disclose regarding a family history when purchasing health insurance.

(iii) Health Care Providers:

Your local doctor and dentist should be advised so they can act as a resource person and assist you organise your health care needs.

(iv) Employer:

There is no reason to inform your employer, and in some cases may result in misunderstanding of the information and adverse consequences.

(v) Friends and family:

We recommend informing your partner and selected, trusted friends. Discuss openly with them that this is confidential and trusted information. They can assist you with support and care, but ask that they not disclose this information as you wish to choose who you disclose to. Some people choose to share this information, others choose not to.

WHAT IF I HAVE CHILDREN?

15. Are my children at risk of inheriting this disease also?

If you are a confirmed carrier of a mutation, each child you conceive has a 1 in 2 (or 50%) chance of inheriting the gene from you, just as you had at your conception. If you have not had genetic testing and are at risk of inheriting a mutation from one of your parents, your children also have a chance of inheritance and fall into the higher risk group status for infection control management in the health care setting.

16. Should I tell my child's doctor/dentist etc?

Yes, your child's dentist and GP need to be informed about the genetic illness in your family. As genetic forms of CJD are so uncommon, they will likely need further information to understand the disease in your family, the infection control risk groups and the Australian infection control guidelines. The Australian National CJD Registry (ANCJDR) can be used for any issues they need further assistance with.

17. What if my child needs their tonsils out?

A tonsillectomy is an invasive procedure; however, based on the tissue type and referring to the Australian Infection Control Guidelines, no special precautions are needed for this procedure.

18. Do I need to tell my child's school?

This is your choice; however, there seems no reason to inform your child's school unless the child's parents or Next of Kin were not available for emergency situations. A blood spill on the school ground or broken arms do not pose any problem to other children or staff, and in these situations only universal precautions are needed. If surgery or an invasive procedure were needed, it is recommended the attending doctor is advised.

19. When can my child be genetically tested?

Generally, in an illness that does not develop until middle or late adult years, the accepted approach is to defer testing for children until they can make the decision themselves. This is life altering information for some people and all children have the right to make this decision for themselves' once they reach 18 years of age.

20. Can my child give blood?

No, the family history of CJD or a genetic prion disease they share with you means they will be declined as a blood donor unless they have undergone predictive genetic testing and their carrier status clarified as negative.

GENETIC TESTING

21. Who can be tested for a mutation on the CJD gene?

When CJD has been diagnosed, testing for genetic CJD or inherited forms of prion disease is available at the ANCJDR, at The University of Melbourne.

Genetic testing could also be available to relatives who are not ill themselves, but this is normally offered to people when a family member diagnosed with CJD is found to have genetic disease after testing. If you are looking for more information about PRNP testing in a relative diagnosed with CJD, or for yourself, please contact the ANCJDR directly for further information.

22. How can I be tested for the CJD gene?

When there is a family history of two relatives diagnosed with CJD, or if your relative has been diagnosed with genetic CJD, genetic counselling and testing is available at centres in each state. You will need a GP referral to your closest genetic counselling service, and to book an appointment. The counselling service will discuss the key CJD related issues with you, and if you decide to proceed further, organise for testing. A small amount of blood would be sampled from two separate visits to the pathology service. From these two samples your genetic material (DNA) would be extracted and the prion protein gene can be tested. There are no fees for the PRNP testing (performed by the ANCJDR). Tests are batched and performed several times a year and results will be given to you by the genetic counselling service.

HAVING A BABY

23. How can the gene be transmitted from one person to another?

The body's genetic information is carried in each cell of our bodies, with half of all a person's genetic information (DNA) contained in each of their egg (for women) and sperm (for men). At conception the egg and sperm join, thus giving each child their full DNA complement. When a parent carries a prion protein mutation, there is a 1 in 2 chance of passing the mutation onto a child. The mutation can only be transmitted at conception.

24. What chance do I have of passing on the CJD gene?

With each conception the chance of passing the mutation to your child is 1 in 2, or 50%. This is for each child conceived, and does not increase or reduce the more children you have.

25. What should I tell my obstetrician and midwife about my risk?

If you are a female at risk of carrying a prion disease mutation, or have been confirmed as carrier of a mutation for genetic prion disease, there is no medical reason your obstetrician or midwife needs advice about your genetic status for care of either your pregnancy or delivery of your baby.

26. Is pre-natal testing available for my baby?

Yes. if you are thinking of pre-natal testing for familial CJD, information about the risk of inheritance in your baby (the parent's information) needs to be given to your obstetrician in the earliest stages of pregnancy. Routinely pre-natal genetic testing of any sort needs to be performed very early in a pregnancy, accompanied by discussions with a genetic counselling service.

27. Can my baby be screened for the CJD gene before birth?

Yes. A genetic test for a foetus could be available if organised through a pre-natal genetic counselling service. Contact your doctor as soon as you plan for a pregnancy or discover you are pregnant to allow organising time. During pregnancy there is a narrow window of time for organising appointments and tests.

COMMON QUESTIONS

28. Is there any risk of transmitting prion disease to my partner sexually?

No, genetic prion diseases are not sexually transmitted.

29. Are there any early signs for genetic CJD?

There are no definite early signs that will be shared by all people starting to develop CJD. Early signs are often missed because the range of first signs is so varied. This is partly because there are many different mutations causing genetic forms of prion disease, including the most common, familial CJD. Early signs could include behavioral changes, balance problems, short term memory problems or a sleep disturbance.

30. Is there a test that can tell me if the CJD gene has become active?

No. We know from experimental work that the disease-associated prion protein is produced in the body some time before any symptoms of the illness can be seen. It is not known when the abnormal protein begins accumulating in the brain.

31. Is there any way of testing if a person has CJD before they die?

There are various tests which can support the suspicion of CJD, but none are considered completely diagnostic because the same results can be seen in a range of other neurological illnesses. The recommended tests include electroencephalograph (EEG), spinal fluid (CSF) testing and magnetic resonance imaging (MRI) of the brain. Brain biopsy is not recommended as the tissue sampled is too small and may not show the CJD changes seen on a full examination of the brain after death. A blood test (DNA) can determine if the patient is a carrier of a mutation for genetic prion disease, if this is not already known.

32. How close are we to a cure?

It is impossible to predict when a treatment or cure is likely to emerge for prion disease, but because it is such a rare and unusual group of diseases there is considerable interest and many research groups throughout the world are conducting laboratory based research including four research teams in Australia.

Trials on treatment options are beginning to emerge around the world.

33. Is there any connection between genetic prion diseases and mad cow disease?

No. Genetic forms of CJD start in one person spontaneously, developing from an error in the replication of genetic material (DNA). The resulting mutation is passed down through the families from parent to child in the genetic material, just as hair or eye colour is passed on. Mad cow disease occurs in cattle and its correct name is bovine spongiform encephalopathy (BSE). BSE has been passed to humans after the consumption of meat products contaminated with BSE. This type of prion disease is called variant CJD (vCJD) in humans. Variant CJD has not been detected in Australia to date.

FOR MORE INFORMATION CALL:

The CJD Support Group Network 1800 052466

The Australian National CJD Registry: 03 8344 1949.

To download the revised National Infection Control Guidelines (Dec 2007)
<http://www.cjdsupport.org.au/docs/chapter31.pdf>

The DVD '*Understanding CJD*' is available free of charge to members of the CJD Support Group Network, their GP's and dentists.

The CJDSGN acknowledges funding provided by the Department of Health & Ageing for the production of this fact sheet.