The CJDSGN provides the below information for *PRNP* (DNA) testing

- **The taking of a blood sample for DNA extraction and long-term storage of DNA and/or future *PRNP* testing**

  (A blood sample, taken while the patient is alive, preserves a sample of the patient's DNA. This allows the patient's family the option to consider *PRNP* testing of their relative at a later stage - no cost)

- ***PRNP* testing for research and surveillance purposes only**

  (Test results will be stored at the Australian National CJD Registry (ANCJDR) and will be available via a genetic service in the future when required by the family – no cost)

- ***PRNP* testing (testing of DNA of a suspected CJD patient) to establish or rule out a genetic cause.**

  (Immediate testing can be requested by the family to rule out or establish a genetic cause for their loved one's illness with suspected CJD particularly if there is a family history of prion disease. This test can only be requested by a neurologist, geriatrician or genetic service and may incur a testing fee)

Please use the 'DNA consent form' and select the preferred option

Sample requirements: At least 5 mls EDTA blood

Send to:
Neurogenetics Unit
Department of Diagnostic Genomics
PathWest Laboratory Medicine WA
Level 2, PP Building
QEII Medical Centre
Hospital Avenue
Nedlands WA 6009
Phone: +61 8 6383 4219
Fax No.: +61 8 9346 4029
Email: Mark.Davis@health.wa.gov.au

The results of *PRNP* testing are not always straightforward, which makes them challenging to interpret and explain. *PRNP* test results provide details of specific pathogenic mutations, benign polymorphisms and codon 129 variants. The implications of a *PRNP* sequence variations (autosomal dominant inheritance pattern and consequences for health care due to infection control ramifications) require specialised guidance for the families involved.

Where possible, the health professional giving the genetic test result should be the same one who provided the pre-test information and counselling. Due to complexity and implications of the genetic *PRNP* results, it is recommended that the request for *PRNP* testing is by a trained specialist (neurologist, geriatrician) or genetic-service.

It is important that the test results are relayed to the family and if a positive result that genetic counselling is encouraged. Regardless of the outcome of the results the family should be offered written confirmation of the *PRNP* result as family members may be required to provide this in the future due to the infection control ramifications that surround prion disease.

For more information please refer to the 'Guidelines for *PRNP* genetic testing'

https://www.cjdsupport.org.au/resources/prnp-genetic-testing/

The 'DNA consent form' can be downloaded from the CJDSGN website and should be signed by both the family member (next of kin) and the doctor and accompany the blood sample.

https://www.cjdsupport.org.au/site/wp-content/uploads/2021/03/DNA-consent-form.pdf

NB: If the family choice DNA extraction and storage only If/when *PRNP* testing is required by the family in the future the CJDSGN can connect the family to the appropriate genetic service so that consent for testing can be completed and a request for testing organised by the genetic service.