PRNP (DNA) testing options for patients with suspected CJD

Testing and/or storage options for future *PRNP* testing

Sample requirements:
At least 5 mls EDTA blood

Storage and/or testing options – please indicate below by ticking one option only:

Option 1

☐ DNA extraction and storage for future *PRNP* testing.
As genetic prion disease is rare most families wait for a 'brain only autopsy' confirmation of CJD before being referred to a genetic service to organise for *PRNP* testing on stored DNA from the CJD patient (no cost)

Option 2

☐ *PRNP* testing (testing of DNA of suspected CJD patient) to establish or rule out a genetic cause.
NB: This test can be requested by a neurologist, geriatrician or genetic service but may incur a testing fee. PathWest will also require a referral document from the clinician's pathology provider to proceed with *PRNP* testing.

Signed consent form and blood sample to be sent 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

☐ I consent to the taking of a blood sample for storage and/or testing options as indicated above

☐ I also consent for *PRNP* results now or in the future to be provided to the Australian National CJD Registry (ANCJDR), for research and surveillance purposes in Australia, and to the Neuropathology team in my state if required to assist with diagnostic results.

Name of Patient _____

Date of Birth _____

Name of Next of Kin _____

Relationship to suspected CJD patient _____

Signed _____

Date _____

Name of requesting doctor _____

Facility_____

Signed _____

Date _____

For more information please refer to the 'Guidelines for *PRNP* genetic testing'
www.cjdsupport.org.au/resources/prnp-genetic-testing/

It is recommended to store extracted DNA from a blood sample for every suspected CJD patient in order to secure the option of future genetic testing for the patient's family once autopsy confirmation of CJD is reported.

Diagnostic testing during the workup of a patient (Option 2) is recommended if there is a known family history or suspicion of a family history of prion disease.
A *PRNP* testing request can only be signed by a neurologist, geriatrician or geneticist.