

# MACHADO JOSEPH DISEASE DOING A BLOOD TEST HELP SHEET



This Help Sheet is designed for primary health professionals who are taking blood from a patient who wishes to know if they have Machado Joseph Disease.

## Background

A patient may be tested for Machado Joseph Disease (MJD) (also called Spino-cerebellar ataxia 3 SCA3) through a genetic test using blood.

The presence of a CAG expansion (also called a Polyglutamine expansion or triple repeat expansion) on chromosome 14q32.1 causes Machado Joseph Disease (MJD).

Each person has two copies of this gene (also called alleles). One inherited from Mum and one inherited from Dad.

If the CAG expansion is between 11 and 44 this is normal. If the CAG expansion is over 52, then the person has MJD and will exhibit symptoms in their lifetime.

If the CAG expansion is between 44 and 52, then the person may never exhibit symptoms, but may pass on the gene to a child and the gene may expand again potentially with an earlier age of onset (this is called an Anticipation effect).

## Results

The results will show two numbers.

One number is for each copy of the chromosome (alleles).

Examples:

18 / 21 – Good news – No MJD

23 / 74 – Bad news – MJD

The reporting time is usually 4-6 weeks.

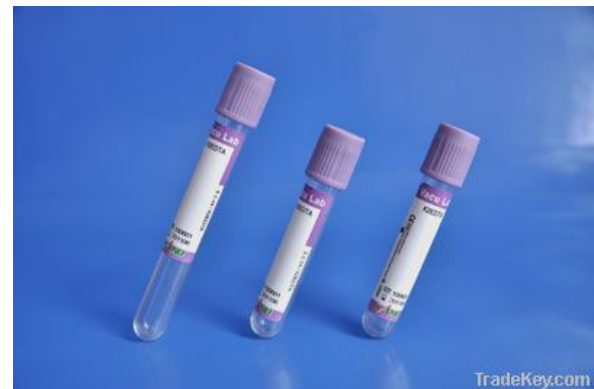
## Collecting the blood

1. Complete the Northern Territory Genetics consent form for molecular DNA testing with the patient (See back of this sheet for a proforma). The laboratory may ask for a copy of it before carrying out the test.

2. Complete the Pathology form used by your clinic. Be sure to state the following:

- Referring Doctor is a Doctor in your clinic
- Note "as discussed with Lindsay Tuer, Genetic Counsellor" in the form.

3. Collection Tubes:



Collect 2 x 5 mls of blood in the **purple** capped EDTA tubes. EDTA (ethylene-diamine-tetra-acetic acid) is a colourless compound used to keep blood samples from clotting before tests are run.

## To regional / community based GPs

- 1) Ensure referral has been sent to the NT Genetics Service at [genetichealth.ths@nt.gov.au](mailto:genetichealth.ths@nt.gov.au)
- 2) Send sample to the Molecular Genetics Laboratory at the Victorian Clinical Genetics Service (sample should go via the Royal Darwin Hospital or the Alice Springs Hospital).