

# WHAT IS MACHADO- JOSEPH DISEASE? HELP SHEET



This Help Sheet explains some basic information about Machado Joseph Disease (MJD).

## **Machado-Joseph Disease (MJD)**

Machado-Joseph Disease (MJD) is a hereditary (genetic) neurodegenerative condition. It is in a 'family' of neurodegenerative diseases called Spinocerebellar Ataxias (SCAs). MJD is also sometimes called SCA3.

## **How is it caused?**

MJD occurs because of a fault in a chromosome that results in the production of an abnormal protein. This protein causes nerve cells to die prematurely in a part of the brain called the cerebellum. The damage to the cerebellum initially causes muscular weakness and progresses over time to a total lack of voluntary muscle control and very significant permanent physical disability.

## **How is it passed down in families?**

MJD is an inherited, autosomal dominant disorder, meaning that each child of a person who carries the defective gene has a 50% chance of developing the disease. The mutation is typically expanded (worsened) when it is passed to the next generation (known as an 'anticipation effect'). Symptoms of the disease sometimes appear around 8 to 10 years earlier for each generation and can be more severe.

## **Is there a cure?**

There is no known cure for MJD. Progression to dependence occurs over 5 to 10 years after the onset of symptoms, and most people are dependent on wheelchair use for their mobility and are entirely dependent on others for all activities of daily living within 10 to 15 years of the first symptoms emerging.

## **MJD in Arnhem Land and beyond**

The effects of MJD have been known to the Aboriginal people of this region for at least four generations, although the first confirmed case was not diagnosed until 1995. Research that was released in 2012 used DNA Haplotyping and linked the genetic strain of MJD found in Australia to that found in Asia. Although it is impossible to predict the number of Aboriginal Australians who will develop MJD, there are currently over 792 Aboriginal Australians thought to be 'at-risk' of developing the disease across the top end. (At-risk individuals are individuals alive today, who are direct descendants of those with the disease. If a parent has MJD, their child has a 50% chance of being born with MJD and therefore developing the symptoms at some stage in their life).

## **MJD impact on social and emotional wellbeing**

The gradual deterioration of nerves and muscles affects the capacity of someone with MJD to take part in many aspects of their daily life. Deterioration in strength, walking and balance, talking and being understood affects an individual's ability to look after themselves, maintain their homes and family, go to work, take part in community events, visit family members, etc. As the disease progresses, people become increasingly dependent on others. Adjusting to progressive loss of independence, while knowing that the disease only gets worse, inevitably affects a person's social and emotional wellbeing.

## **Supportive strategies**

The MJD Foundation encourages people with MJD to 'Stay Stronger for Longer' and works with individuals and families to work out what the person needs in order to 'stay stronger for longer'. Staying stronger for longer will be influenced by:

- healthy living choices
- an adequate and nutritious diet
- appropriate exercise
- adequate rest
- recreation and stimulation
- strong and supportive relationships
- maintaining a person's role within their family and community
- being able to take part in family and community events and ceremonies
- programs and projects that foster empowerment, validation and strength.

**THE MJD FOUNDATION** works in partnership with Aboriginal Australians, their families and communities affected by MJD. MJDF provides:

- equipment to assist people to be independent
- access to community and 'on country' to participate in community and cultural activities
- increased transport options
- kin connection trips that enable people to 'go home' several times a year to keep connected to family and community (when they can no longer live in their community)
- MJD Therapy Programs.