



This submission is aimed to draw attention to the needs of a very specific group of people with a profoundly disabling genetic disease living in large family groups in the most remote and inaccessible regions of Australia's Northern Territory.

The document is submitted by the **MJD Foundation Inc**, a charity established in 2008 to improve the quality of life of Indigenous Australian **Machado Joseph Disease** sufferers and their families in Arnhem Land and beyond. Please see www.mjd.org.au for more information about the MJD Foundation.

This document describes:

Why this group are in need of additional support and help

- Nature of Disease
- Prevalence and Geographical spread
- Location and Associated Infrastructure issues
- General disadvantage and disability
- Lack of Allied Health Therapeutic Services
- Unique carer issues

Issues relevant to 'Improving the system'

- Addressing knowledge and advocacy limitations
- Social and Physical infrastructure
- Recognition of systemic problems
- Integration and coordination of services
- Engagement and Compliance

Nature of Disease

Machado Joseph Disease is a genetic disorder following an autosomal dominant inheritance pattern. It therefore fulfils the disability criteria for this submission by being *present before birth*.

MJD is in a 'family' of neurodegenerative diseases that includes Huntington's disease and 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 control and very significant permanent physical disability.

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. In addition the mutation is typically expanded when it is passed to the next generation (known as an 'anticipation effect') this means that symptoms of the disease appear around 8-10 years earlier and are more severe.

There is no known cure for MJD. Progression to dependence occurs over 5 to 10 years and most people are wheelchair bound and fully dependent for activities of daily living within 10-15 years of the first symptoms emerging. Mean life expectancy from diagnosis is 20 years – meaning that people live with a very high disability burden for a long time.

The disease therefore ultimately results in a *profound core activity limitation*, but for many years also presents as *severe core activity limitation*.

Prevalence and Geographical spread

MJD is a rare neurological disorder – an inherited ataxia within the large spino-cerebellar ataxia group (SCA). The prevalence around the world is 0.63: 100 000. It is however, one of the most well known of the SCA disorders, probably as a result of large, intact family groups, in the Azores in Portugal, and Brazil in South America.

While statistics for the Australian cohort with MJD is unverified, the MJD foundation has credible data indicating that the rates of MJD within the NT Australian Aboriginal population are almost certainly the highest in the world, **exceeding the worldwide prevalence more than 100 times.**¹

Following is an indicative comparative graph of the known world population prevalence per 100 000.

Worldwide Prevalence	0.63: 100 000
India	0.15 :100 000
Germany	1.70: 100 000
Portugal	3.10:100 000
Japan	1.50:100 000
Brazil	3.00:100 000
North East England	8.00:100 000
Azores	43.00:100 000
NT Aboriginal	<u>66.00:100 000</u>

Source Assoc Prof John MacMillan (Genetics Qld).

Australia currently has no specific program funding or legislative protection for those with MJD.

In other countries where the disease exceeds normative levels there has been explicit acknowledgment of the disease as

¹ MJD Foundation has 40 gene positive or clinically symptomatic clients on its books at the current time. Of an NT Indigenous population of 60 000, there is therefore a conservative incidence of 66:100 000.

a public health issue² and legislative measures implemented to address the cost and disadvantage the disease creates.³

Current indications are that the prevalence situation in Australia is likely to exceed that of these jurisdictions and it is likely that similar measures will be required to address the growing issue in the NT and the MJDF will be actively advocating for this to occur.

Current Statistics and Spread

Community	MJD clients	At Risk
Groote Eylandt families (Angurugu, Umbakumba, Milyakburra, Numbulwar, Darwin)	23	130
Elcho Island	10	115
Gove/Yirrkala/Birany	6	100
Ngukurr	5	97
Oenpelli (Oenpelli, Darwin)	3	20
Central Australia	3	20
	45	482



Location and Associated Infrastructure issues

The communities depicted above are classified 'remote', access and service delivery are complicated by considerations such as sparse widely spread populations and extreme weather conditions rendering some communities inaccessible by road for much of the year.

² The **Portuguese Government** Report from the 5th International Workshop on MJD: "Machado-Joseph disease is a disease originated in the Azores, where it has registered the biggest number of known cases – 1 in every 2,402 – which is, for the Region, a **public health problem**, stated the Regional Director of Health"
<http://74.125.155.132/search?q=cache:mQOmS8E1w8wJ:www.azores.gov.pt/Portal/en/entidades/srs/noticias/Machado%2BJoseph.htm%3Fmode%3Dentity+Machado+Joseph+Disease+and+legislation&cd=3&hl=en&ct=clnk&gl=au>

³ "the prolonged incapability which generates psychosocial implications, it became necessary to create special supporting legislative measures in the health and social areas, addressed to Machado-Joseph patients living in the Archipelago, underlined the Director. The Regional Director mentioned, in particular, to diploma that provides a set of social, economic and clinical measures, specifying that beneficiaries may, through the offices of the Integrated Network for Citizen Support (RIAC), make the request for a special protection due to disability by reason of Machado-Joseph disease, without any associated costs"

Infrastructure and service delivery including housing and primary and community health care has been so poor in the past that a review of outcomes culminated in the 'intervention' and massive reallocation of augmentative services, personnel and funding.

The costs involved in providing services to remote communities are estimated to be up to 80% higher than in urban locations and include a doubling of the costs of raw materials due to freight costs, charter flights being the norm as RPT services are scarce or unable to accommodate those with disabilities, and recruitment and retention staffing costs inclusive of relocation, remote allowances and multiple turnover.

General disadvantage and disability

As the data indicates, this places those with MJD at a double disadvantage, not only are they living with one of the most limiting forms of disability known, but they are doing so in a region where *general disadvantage is combined with disability*

The Federal Government is already aware of the unacceptably high rates of Diabetes; Cardiovascular disease; chronic respiratory disease; chronic renal (kidney) disease; and Cancer within the NT Indigenous population.

People with MJD are amongst the most vulnerable of this population, they are simultaneously statistically very likely to have a co-morbidity found within the list above, by virtue of their demographic, and the process of the disease itself contributes to some of these clinical outcomes. They also live in some of the most remote and inaccessible regions of Australia.

Mortality analysis of the best known MJD population in the world (Azores, Portugal) found that the most common cause of death for people with MJD was **respiratory disease**, often as a result of the concomitant complications of MJD -bronchopneumonia, often due to aspiration caused by dysphasia.

Other causes of death known to be associated with MJD, such as decubital ulcer or catheter-related sepsis, are also heavily impacted upon by chronic diseases such as **chronic renal** and **cardiovascular disease**.⁴

It is therefore logical that Australian Indigenous morbidity statistics for this population would reflect an over-representation of the identified chronic diseases within those who have MJD.

Lack of Allied Health Therapeutic Services

Despite the uncontroversial benefits of the provision of regular Physiotherapy, Occupational and Speech therapy services in the prevention and treatment of the disabilities that occur as a

⁴ Lima et al *Arch Neurol*. 1998; 55: 1341-1344

result of MJD and of respiratory, cardiovascular and renal disease there is NO access to regular on site therapy in ANY of the locations in which MJD occurs in the NT.

Existing therapeutic service delivery by the NT DH&F is rudimentary and operated on the basis of fly in fly out advisory capacity (around once per 6 weeks per community) in a cross discipline format (i.e. one discipline covers all three for a region).

This disproportionately disadvantages those with MJD in the region. It is highly probable that access to daily implementation of physiotherapy and hydrotherapy programs, speech pathology vocalisation strengthening, as well as the maintenance and monitoring of swallowing issues would prevent or slow down the development of known risk factors to the development of fatal respiratory disease and other deformities and functional restrictions in those with MJD.

Unique Carer Stress

Carer stress for those with MJD is magnified by genetic nature of the disease and cultural considerations. Due to the anticipation effect demonstrated by the disease there can be up to 3 generations effected within the same family.

Close family relationships and kinship taboos also mean that family carers are the only acceptable carers for personal care. These same people are often at direct risk of developing, or may be experiencing the early symptoms of the disease which include sleeplessness and lethargy. On top of this, they fear it occurring in their children, all while caring for severely disabled older family members.

These are carers with all the recognised burdens of care and then some. The support options available outside the family in these communities are negligible consequently they are at significantly increased risks of depression and family breakdown.

Improving the system

Addressing knowledge and advocacy limitations

The clients who are the subject of this submission have experienced poor delivery of services, over several lifetimes. They live in remote locations and have culturally embedded ways of dealing with morbidity and disability that are not necessarily in concert with mainstream western medical service delivery models.

It will be important to address this by ensuring that this client group is provided with information sufficient to ensure that they are aware of the range of services and rights available to people in less remote locations.

Opportunities to engage with advocacy and complaint processed and services will also be important. Single service provider models leave clients vulnerable to loss of service if not carefully monitored – ideally this would be conducted by independent organisations.

Social and Physical infrastructure

In order to adequately provide care it is essential to properly cost service on costs. In situations where service delivery costs swallow direct client provision funding, the outcome is poor service delivery and commensurately poor health and disability care outcomes.

This is clearly the case in the context of the bulk of remote health and disability service delivery. Where the costs are demonstrable and essential to the maintenance of a stable well resourced personnel they should be added to the funding models and not incorporated within them.

Recognise systemic problems

Situations exist in this region where clients have had to wait up to 6 months for the provision of a wheelchair and the repair, maintenance and replacement options for other essential equipment are negligible.

While there are undoubtedly many barriers to providing services and equipment, recognition of them and the setting of best practice standards which trigger review if unmet would vastly improve the outcomes for those with disability living remotely in general and people with MJD specifically.

An example of a model which may improve service delivery can be found at Appendix A (TIME scheme submission).

Integration and coordination of services

Data collection methods, eligibility criteria and allocation of resources per client/need utilised by government and non government service providers need to be consistent and transparent and capable of use in integrated service delivery planning.

At present this does not happen, with multiple territory and commonwealth funded government and non government operated programs operating simultaneously - creating a veritable maze for consumers and providers to negotiate.

Mandatory collaboration and interaction regarding client priorities, needs issues to be addressed and service delivery plans to would greatly alleviate this and the need for clients to provide the same information repeatedly.

Engagement and Compliance

Difficult as it undoubtedly may be, accounting for specific local issues when determining service provision is an essential part of providing accessible well planned services.

In this context it is especially important to recognise the underlying social and economic conditions which are the precursors to poor remote indigenous community health. The only credible way to understand these issues and change outcomes is to work closely with local community organisations, taking seriously language and education differences and building time allowances to deal with these into programs.

Currently the lack of time spent in communities particularly by remote therapists is extremely detrimental not only to the clients experiencing the service provided, but also to the personnel employed (invariably for short periods of time).

Alongside this, inter-sectoral collaboration designed to maximise local skill development and credentials should be at the forefront of program design. Teaching local people to provide services and to work alongside appropriately credentialed professionals should be essential to properly addressing local workforce issues.

Recommendations

Embedded in this document are several generic and specific recommendations. The MJDF are committed to working toward improvement in life quality for those with MJD and are actively involved in research, advocacy, education, respite and augmentation of services as described in the annual report and newsletters which can be found at www.mjd.org.au.

Further consultation is invited and welcomed.