Machado-Joseph Disease PHC Remote Guideline

<table>
<thead>
<tr>
<th>Target Audience</th>
<th>All Clinical Employees</th>
</tr>
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<tbody>
<tr>
<td>Jurisdiction</td>
<td>Primary Health Care Remote CAHS; Primary Health Care Remote TEHS</td>
</tr>
<tr>
<td>Jurisdiction Exclusions</td>
<td>N/A</td>
</tr>
<tr>
<td>Document Owner</td>
<td>Kerrie Simpson</td>
</tr>
<tr>
<td></td>
<td>Atlas Development Officer Primary Health Care Remote CAHS</td>
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<tr>
<td>Approval Authority</td>
<td>Refer to Policy Guideline Centre</td>
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<tr>
<td></td>
<td>NT Quality and Safety Manager Primary Health Care</td>
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<tr>
<td>Author</td>
<td>PHC Quality and Safety Team; Machado Joseph Disease Working Group</td>
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The attributes in the above table will be auto-filled from the PGC System. Do not update in this document.

Purpose

To provide Primary Health Care remote staff with a guideline on the management and care pathways provided to clients with Machado-Joseph Disease.

Guideline

1. General Information

Machado Joseph Disease (MJD) is found in some Northern Territory Aboriginal families with the largest number of people with MJD living in East Arnhem (EA). MJD occurs worldwide with the EA genetic origins from Asia.

MJD is an autosomal dominant neurodegenerative disease affecting the cerebellum, basal ganglia, dorsal column, upper and lower neurones and peripheral nerves. The nerve cells are prematurely destroyed resulting in multisystem degeneration. The disease is hereditary; offspring of affected parent(s) have a 50% chance of being affected; has an anticipation effect of an early onset in the next generation and more rapid progression. The symptoms can be variable within families.

There is no cure for MJD. Some treatments and medications may alleviate some of the symptoms, prevent complications, improve quality of life and assist those with the disease and their carers to live in the community. As MJD is familial disease, it is important to consider whether the carer is at risk of developing the symptoms or has MJD. Many clients diagnosed with MJD care for loved ones who also have the disease in more advanced stages.

For detailed information see the MJD Care Guidelines which provide a description of the disease process, early interventions, ongoing management and details regarding available resources, services and community support.

Note: A hard copy of the MJD Care Guidelines will be available in health centres providing care for clients with MJD. It is important that Health Practitioners new to caring for clients with MJD contact the MJD Foundation for advice, information and support.
2. Definitions

Progression of MJD can be assessed and managed in three stages, mild, moderate and severe:

- **Mild stage**: (duration approximately 10 years) little or no assistance required for mobility.
- **Moderate**: (duration approximately 5 years) requires a walking aid.
- **Severe**: (duration approximately 5 to 7 years) wheelchair dependent.

**Electronic Health Record (EHR)**: two EHR systems are used within Primary Health Care Remote, namely:
- PCIS - Primary Care Information System
- EACS - East Arnhem Communicare System

**My eHealth Record (MeHR)**: a secure, electronic record of an individual's medical history stored and shared in a network of connected systems so that vital health information can be securely exchanged between different health care providers such as Medical Practitioners, specialists, pharmacists and hospitals.

3. Responsibilities

3.1 **Health Centre Clinical Staff**

- Ensure all clients with MJD are placed on an **MJD Care Plan**
- Provide ongoing health care to clients according to the MJD Care Plan and other relevant health checks
- Ensure all clients with MJD are placed on the Electronic Health Record (EHR) recall system and **My eHealth Record**
- Be aware of and liaise with community resources in supporting clients with MJD / carers / families
- Refer clients and their families to the **Disability Services** and the **Machado Joseph Disease Foundation** as appropriate

3.2 **Medical Practitioner**

- Utilise the MJD **Standard Treatment Protocol**
- Complete the clinical examination component of the **MJD Care Plan**
- Support and participate in providing care according to the MJD Care Plan
- Collaborate with health centre clinical staff, clients and their families to develop and monitor clients on MJD Care Plans as required
- Assist with follow up and referral as necessary according to MJD Care Guidelines

3.3 **Office of Disability (OoD) Services** (Allied Health: Speech Pathology, Occupational Therapy, Physiotherapy, and Disability Coordinator)

- Undertake an initial Allied Health screen if mild to moderate stage of the disease and refer as appropriate
- Provide yearly follow up with clients in mild stage
- Undertake regular comprehensive assessment in the moderate to severe stage and complete **individual support plan** and place a copy with the client's EHR
- Coordinate support and educate in providing care according to the **MJD Care Plan**
- Assess and prescribe equipment, aids and home modification
- Liaise with health centre staff and community resources in supporting clients with MJD / carer / families
- Liaise and work with the **MJD Foundation** to provide additional disability support for clients / carers / families
3.4 **Machado Joseph Disease Foundations (MJDF)** - Nursing, Social Work, Occupational Therapy, Physiotherapy and Education and Resource Manager
   - Work closely with individuals, families and the health centre to provide additional support
   - Identify additional education and support needs for families, and community
   - Provide education and support to health and community organisations
   - Provide supplementary equipment and community services
   - Conduct targeted research activities

3.5 **Primary Health Care Outreach Team** - Preventable Chronic Conditions Educator, Public Health Nutritionist, Health Promotion and Evaluation Officer, Aboriginal Health Promotion Officer and Aboriginal Men's Health Worker
   - Supporting induction of new staff on the use of templates
   - Work closely with the health centre to provide additional support such as community education, staff training and case management
   - Liaise with other government and non-government organisations and local agencies

4. **Procedure**

The MJD Standard Treatment Manual Protocol and MJD Care Plan provide guidance for Primary Health Care (PHC) Remote staff to assess and manage MJD.

4.1 **Machado Joseph Disease Care Guidelines**

The MJD Care Guidelines (2nd Edition) provide care, management and support information on management and support of MJD symptoms.

4.2 **Initial Assessment**

Office of Disability Services utilise an [Allied Health Screening Tool](intranet) which provides an initial screen and checklist for referrals.

The Allied Health Screening Tool is used in the early to mild stage of the disease. The tool screens ataxia, muscular strength and pain, sleep, vision, emotional wellbeing (PHQ-2) dysarthria, dysphagia, continence (bowel and bladder) weight, and fatigue.

4.3 **Genetic Counselling**

Genetic counselling provided by the Medical Practitioner or others trained in genetic counselling. For further information [MJDF Genetic Counselling Guide](#)

4.4 **Documentation**

Follow MJD Standard Treatment Protocol, Care Plan, Care Guidelines and referral reports and plans such as Disability Services Individual Support Plan.

4.4.1 **Machado Joseph Disease Care Plan**

The MJD Care Plan is developed with the person with MJD / family and carers to provide the best health care and ensure appropriate and timely interventions are put in place.

The [MJD Care Plan](#) should reflect the stage of the disease:

- **Mild stage:** little or no assistance required for mobility
- **Moderate stage:** requires a walking aid
- **Severe stage:** Wheel chair dependent.
4.6 Referral

Clients with MJD may require a range of referral services according to their need. Referral services may include:

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<tr>
<th>Service</th>
<th>Description</th>
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<tbody>
<tr>
<td>Ophthalmology</td>
<td>to assess the clients vision and any changes and support the client and carer understand how visual difficulties impact on function and have the skills to cope with these difficulties</td>
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<tr>
<td>Rehabilitation Medical Specialist</td>
<td>for assessment and follow up of neurological functional needs</td>
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<tr>
<td>Oral Health Services</td>
<td>assessment of oral health status</td>
</tr>
<tr>
<td>Mental Health Services</td>
<td>to provide mental health support and intervention</td>
</tr>
<tr>
<td>Physiotherapy</td>
<td>Assessment of mobility and coordination, equipment, environmental and natural supports for client and carer, and patient and carer education</td>
</tr>
<tr>
<td>Occupational Therapy</td>
<td>Assessment of daily activities, home modification, aids and equipment, environmental and natural supports for client and carer, and patient and carer education</td>
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<tr>
<td>Speech Pathology</td>
<td>Assessment, management and education of swallow and speech</td>
</tr>
<tr>
<td>Dietitian</td>
<td>Assessment, management and education on maintaining nutritional status including fluids</td>
</tr>
<tr>
<td>Radiographer</td>
<td>u/s of bladder volumes pre/post voiding (In some cases MRI has been used for diagnosis size of cerebellum)</td>
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<tr>
<td>Visiting physician</td>
<td>Advice on management of disease complications and management of comorbid conditions</td>
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<tr>
<td>Neurologist</td>
<td>Assessment and review of neurological management</td>
</tr>
<tr>
<td>Community services</td>
<td>In home Support (carer support, MOW, day respite)</td>
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<tr>
<td>MJD Foundation</td>
<td>Provide additional support, identify educational and support needs of families and community, provide education, some equipment, support health and community organisations, provide advocacy, and monitor family pedigrees.</td>
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<tr>
<td>Territory Palliative Care</td>
<td>to support clients with a life limiting illness with the aim of improving the quality of life of clients and their carers</td>
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4.7 Resources and Community Supports

Clients / carers and families have a need for a variety of resources, services and community supports. Details related to equipment; financial; housing; and resources, services and community supports, are provided in the MJD Care Guidelines, Section 3 - Resources and Community Supports.
### Document Quality Assurance

<table>
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<th>Method</th>
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<td><strong>Implementation</strong></td>
<td>Document will be accessible via the Policy Guidelines Centre and Remote Health Atlas</td>
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<tr>
<td><strong>Review</strong></td>
<td>Document is to be reviewed within three years, or as changes in practice occur</td>
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<td><strong>Evaluation</strong></td>
<td>Evaluation will be ongoing and informal, based on feedback.</td>
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### Key Associated Documents

<table>
<thead>
<tr>
<th>Forms</th>
<th><strong>Allied Health Screening Tool</strong> (intranet)</th>
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</table>
| **Key Legislation, By-Laws, Standards, Delegations, Aligned & Supporting Documents** | **Advance Personal Planning**  
**Machado-Joseph Disease Care Plan**  
**Standard Treatment Protocol for Machado Joseph Disease / Spinocerebellar Ataxia Type 3**  
NT Department of Health: **Disability Services** – intranet document library  
**Allied Health Clinical Guidelines**  
**Machado Joseph Disease Foundation**  
**MJDF Genetic Counselling Guide**  
https://www.ataxia.org.uk/Handlers/Download.ashx?IDMF=7f6fd040-6928-4121-a225-9a42d4e80c2d  
www.ninds.nih.gov/disorders/machado_joseph/detail_machado_joseph.htm  
<p>| <strong>References</strong> | <strong>As Above</strong> |</p>
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<th>Evidence level (I-V)</th>
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