Submission to the Senate Standing Committees on Community Affairs, inquiry on universal access to reproductive healthcare

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Submission Purpose

This submission aims to describe the reproductive and genetic health care issues facing Aboriginal families living with Machado-Joseph Disease in remote Australia and provide recommendations to address these.

Executive summary

Aboriginal families living with the rare, genetic, disabling condition Machado-Joseph disease (MJD) in very remote Australia face a range of obstacles related to the physical and psychological consequences of the disease. Current therapies to assist with the impact of the disease focus on managing the symptoms of those affected. As the lived experience of this cohort matures, access to genetic education, counselling, and reproductive health strategies will form part of their opportunities to mitigate the disease.

Some of the strategies to assist should include:

(i) improving access to contraception and understanding of contraceptive options in remote Aboriginal communities,

(ii) improving access to reproductive health care, especially medically assisted reproduction and termination care in remote regions,

(iii) enhancing Aboriginal Health Worker models which focus on the transfer of cultural knowledge to be used in developing appropriate care and management plans,

(iv) enhancing undergraduate genetic education across the health provider workforce,

(v) producing and promoting appropriate first language reproductive healthcare resources created in collaboration with Aboriginal people from the relevant locations.

Introduction/Background

On 28th September 2022, the Senate referred an inquiry into the universal access to reproductive healthcare to the Senate Community Affairs References Committee for inquiry and report by 31st March 2023.

There is a current consultation listed on the Senate Standing Committees on Community Affairs website, which is open until 11.59 pm AEDT on 15th December 2022. The MJD Foundation appreciates the opportunity to provide a submission. This submission is written in response to the Committee Terms of Reference.

The MJD Foundation consents to this submission being published on the inquiry website and shared publicly online.

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MJD Foundation

Since its inception in 2008, the MJD Foundation (MJDF) has been working in partnership with Aboriginal Australians, their families and communities living with Machado-Joseph Disease (MJD) in a growing number of remote and very remote communities and urban centres across the Northern Territory (NT) and in far northern Queensland (QLD).

The MJDF has substantial and unique experience in Aboriginal very remote non-government disability service provision. We design and deliver programs in response to our clients' expressed needs, despite very high costs and frequently meet gaps in government capacity.

Machado-Joseph Disease

Machado-Joseph disease (MJD) is a genetic neurodegenerative disease experienced worldwide. It is caused by a pathogenic CAG expansion which produces an abnormal protein compromising muscle and nerve function and is the most common of the very rare spinocerebellar ataxias. The disease results in devastating long-term disability and has a protracted history in north Australia, with reports dating from the 1960s in Yirrkala and the Groote Eylandt communities. Families are now living with MJD in remote communities across the Northern Territory and in far north Queensland, where local prevalence rates are among the highest known in the world.

Across these locations, 118 Aboriginal people are experiencing clinical symptoms and a further ~792 people are 'at risk[1]' of having inherited the disease.

To date, there has been no detailed analysis of the health, social and economic implications for individuals and communities of the high prevalence of the disease among Australian Aboriginal families. Unsurprisingly, however, there is a long record of observations of considerable distress for families experiencing MJD. Their experience of the Aboriginal phenomenon of 'shame' and stigmatisation is consistent with reports of the disease in populations elsewhere. Public health costs are also significant, in line with the long duration and profound disability of the disease.

Disease management strategies and considerations

Despite significant research efforts, MJD remains lethal and untreatable, with National Disability Insurance Scheme (NDIS) plans coordinated by the MJDF only able to assist with therapies, community access, personal care and adaptive equipment to manage symptoms. Genetic features of the disease, such as late/adult onset and 'anticipation' (a tendency for onset and severity to increase generationally), compound distress and have meant that genetic education and counselling to family members who are 'at risk' are priorities for families. As government genetic care services are sparse, urban and anglo-centric, the MJDF has worked closely with local communities to evolve place-based culturally sensitive programs, once funded by the Federal Government, but in recent times utilising philanthropic funding.

The families' experiences, the severity of the disease and the lack of current effective treatment options highlight the importance of considering other mechanisms to address the spread of the disease for Australian Aboriginal families.

The use of medically assisted reproductive technologies for autosomal dominant late-onset diseases such as Huntington's disease and familial amyloid polyneuropathy (FAP) is well established internationally. Senior Aboriginal women from the communities on Groote Eylandt have recently commenced investigating the cultural appropriateness of managing the disease from a genetic

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[1] An "At risk" individual has a 50% chance of having inherited the faulty gene from an affected parent.

transmission perspective. However, there is no published evidence about the availability and uptake of medically assisted reproductive health care for this cohort.

Assisted reproductive technology options *theoretically* available for people with MJD who do not wish to pass on the CAG expansion include: (i) IVF and the use of donor gametes (eggs or sperm), (ii) IVF with donated embryos, (iii) PD - the testing of a naturally occurring pregnancy at 10-13 weeks, followed by termination of pregnancy if necessary and (iv) PGD which refers to the genotyping of embryos, testing for expanded CAG before transfer during IVF by intracytoplasmic sperm injection.

Prenatal diagnosis (PD) and preimplantation genetic diagnosis (PGD) technologies are the core procedures that allow prospective parents with hereditary diseases to make family planning choices without using genetic material from other people. Understanding, evaluating the acceptability of, and access to these procedures is essential to allow Aboriginal people living with MJD make choices that are congruent with their priorities and worldview. The barriers to this, however, are substantial and are described following.

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Commentary under the Inquiry's terms of reference

Barriers to achieving priorities under the National Women's Health Strategy for 'universal access to sexual and reproductive health information, treatment and services that offer options to women to empower choice and control in decision-making about their bodies', with particular reference to:

a. cost and accessibility of contraceptives, including:

  i. Pharmaceutical Benefit Scheme (PBS) coverage and Therapeutic Goods Administration (TGA) approval processes for contraceptives,

  ii. awareness and availability of long-acting reversible contraceptive and male contraceptive options, and

  iii. options to improve access to contraceptives, including over the counter access, longer prescriptions, and pharmacist interventions;

People affected by MJD can elect not to have children to avoid passing the disease on. Should this be a valid option for them, understanding the benefits and compromises of a range of easily accessible contraceptive options and access to personal, individually targeted advice needs to be prioritised.

Most remote communities have no place-based, direct-to-customer pharmacy services within several hundred kilometres, reducing the opportunities for a discrete consultation. Instead, availability and advice for contraception is provided through local primary health care services, which are inevitably open to the scrutiny of others in the community and, anecdotally, prioritise female-centred, long-term contraceptive options. Broadening this would enhance the contraceptive opportunities and potential uptake, especially for young people at risk of MJD.

b. cost and accessibility of reproductive healthcare, including pregnancy care and termination services across Australia, particularly in regional and remote areas;

Minimal place-based reproductive care options are available in remote Aboriginal communities- beyond standard antenatal care until the weeks before birth. All birthing for remote communities is mandated to occur in regional hospitals. The combination of a lack of resources, hesitancy to engage with services and traditional preferences can result in a relatively late presentation of pregnancy. This raises a range of issues relevant to reproductive care for people at genetic risk, including the timely availability of prenatal diagnostic procedures, which might, in some circumstances, result in the need for termination services. Termination services, especially surgical terminations, are also routinely provided only in regional centres. Medically assisted technology for infertility or prevention of genetic disease is rarely accessed and almost unknown in local remote Aboriginal communities.

The MJD cohort in remote communities has no working knowledge of these procedures at this point in time. Access to regional hospitals for any health care need involves significant travel, cost, dislocation from support systems and the delivery of health care bedded in Anglo/western perspectives and language. All of these are barriers to effective engagement and the comfort and well-being of the patient. Consulting with the communities in question will be integral to developing more tailored and effective models of reproductive care.

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c. workforce development options for increasing access to reproductive healthcare services, including GP training, credentialing and models of care led by nurses and allied health professionals;

The medical workforce in remote Australia, particularly in Aboriginal communities, is infamously itinerant, with short-term tenure the norm and much of the medical care provided by agency staffing sent from urban centres. The capacity for individuals working within this model to respond to unique local cultural perspectives and priorities requires bolstering. Enhancing this could be achieved by utilising the techniques that develop capacity and ownership within local staff, such as Aboriginal health workers.

Genetic health literacy is demonstrably poor across the Australian healthcare sector, with minimal content at the undergraduate level provided for medical, nursing and allied health students. In locations where genetic disease is known to be present, providing targeted education opportunities for these students and staff will improve knowledge and information exchange in both targeted and general interactions with patients and increase the capacity for people at risk of genetic disease to make genuinely informed choices.

d. best practice approaches to sexual and reproductive healthcare, including trauma-informed and culturally appropriate service delivery;

Cultural protocols dictate much of the interactions between people in remote Aboriginal communities. These include gendered interactions in some circumstances and may be underpinned by important moiety-related patterns of appropriate behaviour. These protocols may extend to the composition of support persons - which may encompass people beyond the nuclear family and deviation from notions of individual autonomy within decision-making.

Any healthcare interaction, but especially those related to intimate/sexual/reproductive matters, needs to be informed by these critical protocols. Providing reproductive support and advice to families dealing with genetic diseases requires local expertise and first language support. This support should be achieved through collaborative partnerships with local community agencies and oriented to sharing medical and cultural knowledge in a dynamic exchange. Engagement without these critical supports may breach protocols and is not only likely to fail, but also deeply disrespectful.

e. sexual and reproductive health literacy;

Many of the families experiencing MJD speak English as a second, third or fourth language. Despite this, the bulk of all health resources remains in printed English. Access to interpreters is often challenging, especially in regional hospitals. Consideration should be given to producing and promoting appropriate first-language resources involving Aboriginal people from the relevant locations.

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f. experiences of people with a disability accessing sexual and reproductive healthcare;

MJD causes significant permanent disability, however, for this Australian Aboriginal cohort, the majority of those currently affected developed symptoms in adulthood, having lived until then as abled-bodied people. Frequently their reproductive years are spent with minimal symptoms of MJD, although this may change in coming decades as younger people experience symptoms earlier (due to ‘anticipation’).

Despite this and the minimal impact of the disease on physical access to reproductive health care to date, there are barriers to accessing reproductive/genetic health care related to the impact within families and subsequent emotional, social and psychological distress. As for people experiencing Huntington's disease, there may be disengagement with genetic or reproductive health care services, and people may not want to know or engage with their genetic risk. In these circumstances access to psychological support and the education of medical professionals will be necessary. This right not to know is valid and vital, although seemingly contra to scientific/medical opinion.

Recommendations

Universal access to reproductive healthcare is essential. The MJD Foundation supports this important Inquiry, with the following recommendations:

- improving access to contraception and understanding of contraceptive options in remote Aboriginal communities,
- improving access to reproductive health care, especially termination and medically assisted reproduction in remote regions,
- enhancing Aboriginal Health Worker models, which focus on the dynamic transfer of cultural and medical knowledge to be used in developing appropriate care and management plans,
- enhancing undergraduate genetic education across the health provider workforce,
- producing and promoting appropriate first language resources involving Aboriginal people from the relevant locations.