

Accessibility of genetic services for Aboriginal and Torres Strait Islander people

Better Indigenous Genetic (BIG) Health
Services partnership project

Hot North Meeting

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Imogen Elsum

Uni Melb

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Northern Territory Genetic Services

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Julie McGaughan

Julie White

Rachel Austin

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Gareth Baynam

Rebecca D'Souza

Project Reference Group and End User Group

Aboriginal Medical Services Alliance Northern Territory, Aboriginal Health Council of Western Australia, Anindilyakwa Land Council, Northern Land Council.

BIG Health Services – Overview



WHAT

Improve the provision of genetic health services to deliver health benefits to Aboriginal and Torres Strait Islanders affected by genetic disease

WHY

- *Unmet need for clinical genetics and genetic counselling in Aboriginal and Torres Strait Islander populations.
- *Under represented in services.
- *Growing potential for genomic medicine and increasing availability of genomic testing.
- *Lack of Aboriginal genomic reference data → delayed diagnosis, treatment and management.
- *Important to ensure delivery of equitable health benefits.

HOW

Partnership between researchers at several universities and genetic service providers that are currently in use delivering genetic health services in NT, WA & Qld

FUNDING

Grants: Lowitja (376,197) and NHMRC (351,240). Partner Contributions: MJDF (60,000) and OPHG (40,000). Total AUD 827,437 (2016 – 2019)

BIG Health Services – Methodology

Exploratory, open approach to explore and identify barriers and facilitators at different stages of the genetic services care pathway.

Qualitative (semi-structured interviews)

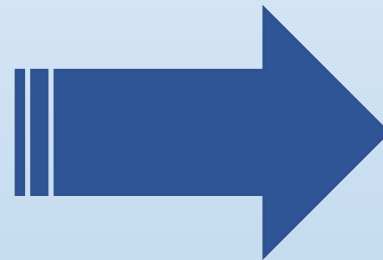
genetic service providers

primary health care providers and workshops

patient journey mapping with Aboriginal and/or Torres Strait Islander users/guardians of genetic services

Quantitative

genetic service provider database audits



**Development and
piloting
resources/strategies**

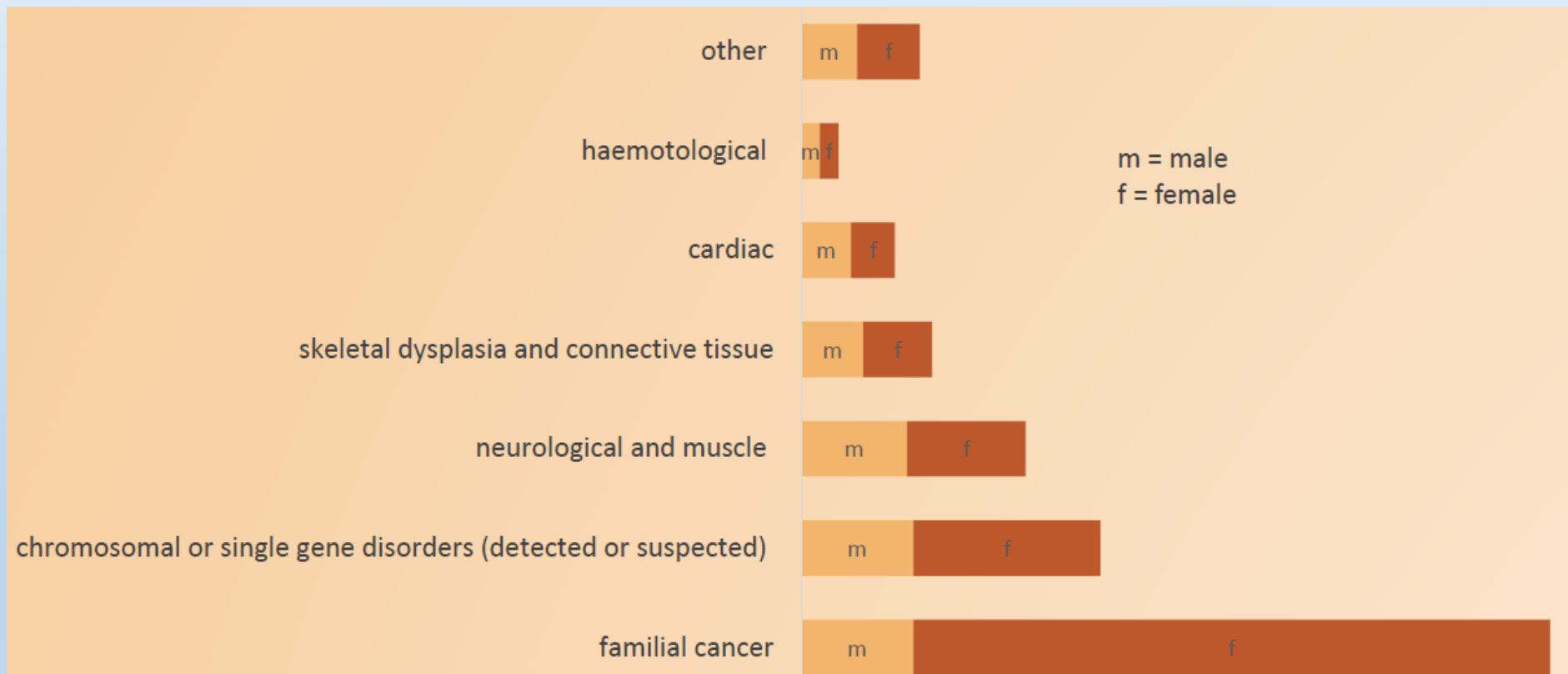
BIG Health Services

THE DIFFERENT MODELS OF CLINICAL GENETIC SERVICES

1. **The Machado-Joseph Disease Foundation (MJDF)** model, a community-driven that provides health and social support services to people affected by the autosomal dominant genetic neurodegenerative condition Machado Joseph Disease.
2. **The Northern Territory Genetics Services (NTGS)** model operates as a specialist outreach service across NT with clinics times per year in Darwin and Alice Springs.
3. **Genetic Services of Western Australia (GSWA)** model provides paediatric, adult, prenatal and familial cancer services in metropolitan and regional WA.
4. **Genetic Health Queensland (GHQ)** model offers a state-wide service providing clinical genetic services (diagnosis, screening, education and counselling).

Northern Territory Genetics Services (NTGS)

- Patchy history of genetic service provision to the NT
- NTGS is staffed by geneticists and a genetic counsellor based VCGS, Melbourne and Darwin-based coordinator
- Small team covering all genetic conditions



2015 referral distribution by primary genetic indication

Northern Territory Genetics Services (NTGS)

- Referrals can be through specialist/tertiary care, general practitioners or self referred
- Vast majority of referrals coming from urban based specialists
- Under representation of Aboriginal and/or Torres Strait Islander (by approx. 50%). **Why?**
- *?Lack of referrals from PHC in Aboriginal communities?*
?Model of service provision not aligned with needs?

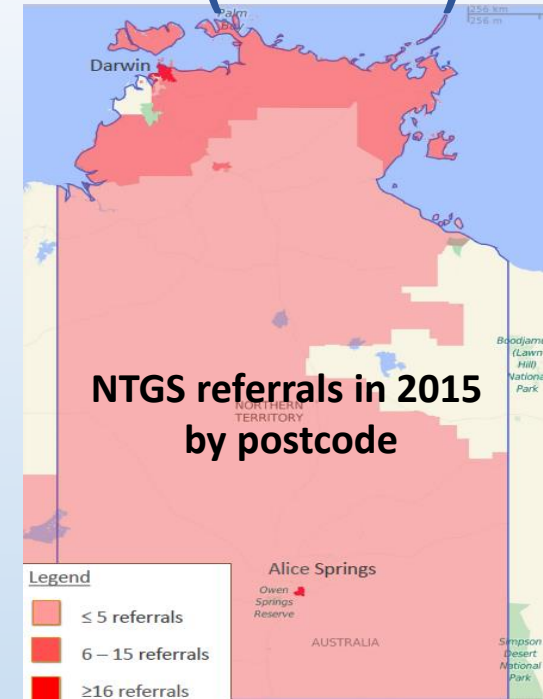


Table 1. Summary of Aboriginal and/or Torres Strait Islander representation in the NT Genetics Services in 2015

	Aboriginal and/or Torres Strait Islander	Non-Indigenous
NT population data [2]	29.8%	70.2%
Total NT genetics referrals	15.1% [45% male, 55% female]	84.9% [28% male, 72% female]
Patients seen in NT clinics	13.9% [9.6% RDH, 37.5% ASH]*	86.1% [90.4% RDH, 62.5% ASH]*

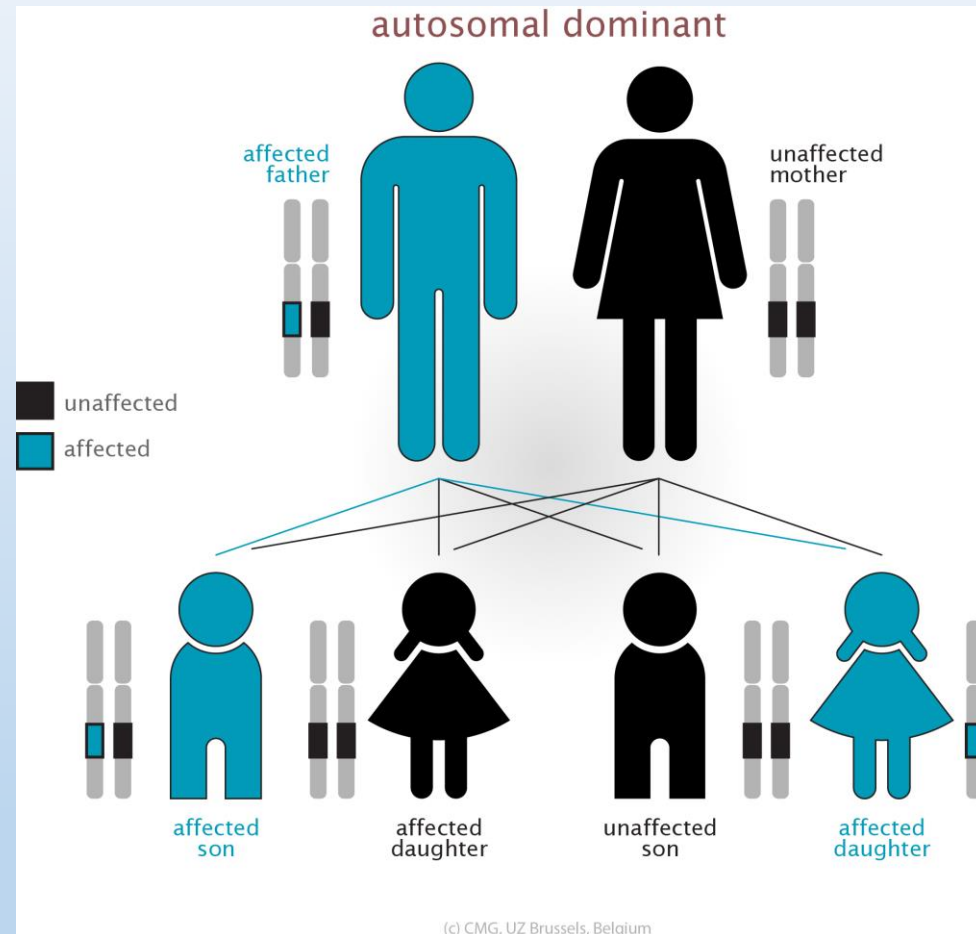
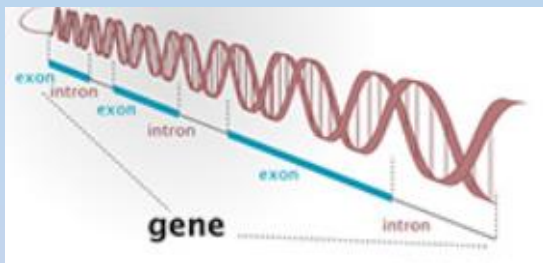
Note: Indigenous status was determined by the NT Department of Health Records.

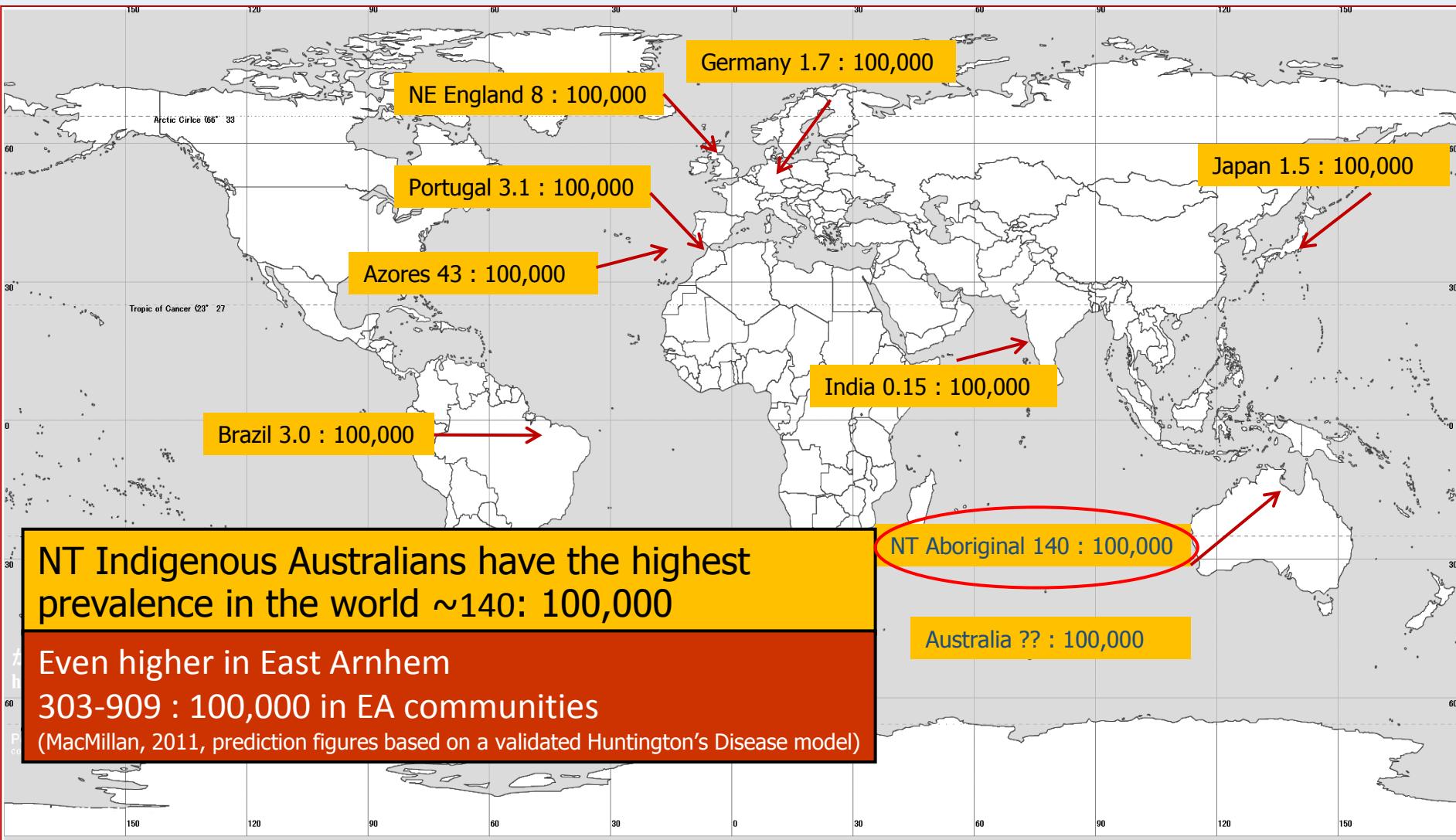
* Royal Darwin Hospital (RDH), Alice Springs Hospital (ASH)

Tuer et al, NTGS, 2016

Machado Joseph Disease (MJD)

- MJD (Spinocerebellar Ataxia 3) is an autosomal dominant genetic disease that exhibits anticipation
- Neurodegenerative; impacts on mobility, speech, swallowing, continence, sleeping and vision. No loss of intellectual capacity
- Disease progression from onset to total dependence ranges from 5 – 15 years, influenced by age of onset and number of CAG repeats (in expanded SCA3 gene)





MJD Prevalence - Aboriginal Australians

Updated: Oct 2017



Location	Symptomatic or +ve test	Monitored ^	At risk *
Groote Eylandt	12	13	186
Yirrkala	3	14	112
Elcho Island	11	12	121
Ngukurr	5	10	95
Central Australia	5	3	97
Numbulwar	1	2	4
Oenpelli	3	1	8
Nth Qld	5	1	23
S. Australia	1	0	0
Total	46	56	646
MJD	101		

^ Monitored –people who've had symptoms reported and are being monitored by the MJDF

* At risk – people alive today who have a parent/grandparent with MJD. Children with a parent with MJD have at least a 50% chance of inheriting the defective gene.

Education



Advocacy



MJDF Activities

Equipment



Research



Improved Services



Barriers - Lessons learned

- Lack of awareness and information especially in primary care around what services available. NTGS not in service directory or referral software (PCIS). See specialist before being referred.
- What is standard in Western model of genetic services often not doesn't fit with priorities and culture of Aboriginal communities.
- Needs to be tailored to what community wants and what fits. Will take more time and resources to get right but will result in improved health outcomes.

Examples from communities that MJDF service re talking about genetics :

- Collective decision making is important. Differs from Western emphasis on confidentiality and privacy
- Informal and familiar settings are key. Clinical settings are not always conducive to engagement.
- Gender roles (women's and men's business). Having choice of female/male geneticist/genetic counsellor in a discussion around genetics and family history.
- Trust – genetics can involve intimate topics. Relationships and trust needed.
- Timing – when people will be ready to talk will be highly variable and personal.

Thank you

A couple of questions for you.... welcome feedback and thoughts on:

- *How do you see the role of PHC in the genetic services care pathway, bearing in mind the increasing role personalized medicine is playing in health care?*
- *What do you think are priorities to facilitate increased access to clinical genetic services for Aboriginal and Torres Strait Islander people?*

Imogen.Elsun@unimelb.edu.au