



MACHADO JOSEPH DISEASE GENETIC COUNSELLING SESSION 1

MJD Foundation and Associate Genetic Counsellor Lindsay Tuer
Version 2 – September 2016

Pre-test consent session

***please prioritise first language support by AHW/ACW**

Recognition of the development of this resource goes to the late Assoc Prof John MacMillan

1. Talk about family history - go through pedigree
2. Talk about perception of Machado Joseph Disease
 - symptoms in affected parent / aunts / uncles / siblings
 - symptoms in self: balance, double vision, swallowing problems, formal medical assessments
 - feelings about self
3. Talk about risks
 - self / siblings / children / grandchildren
 - review genetics of MJD / how it is inherited from parent
 - (Note: should have been covered by MJD Foundation Education Session)
4. Explain protocol for test
 - consent form
 - time line for result (~8 weeks)
 - mutually agreed upon follow up plan: where / when / how of result disclosure session 2 (ideally face to face)
5. Talk about motivation for predictive testing and timing of testing
 - good / bad scenarios
 - effect of results (good or bad results) on partner, children, siblings etc
 - family planning
6. Talk about support
 - family dynamics
 - who knows?
 - who will be told?
7. Ask about consent of result to be made known to MJD Foundation yes / no (written consent form)
8. Talk about options in research participation
8. Able to discuss further with health care provider whilst awaiting result.

Do you need more information?

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MACHADO JOSEPH DISEASE GENETIC COUNSELLING SESSION 2



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Version 2 – September 2016

Results session

* please prioritise first language support by AHW/CW

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1. Results
 - a. good or bad
number of repeats
age of onset - not able to be predicted
prognosis / management / treatment / hope for future
 - b. Emotions: shock, incapacity, guilt, relief etc
 - c. Other's experiences
 - young ones - unmarried
 - middle aged, with or without children
 - older ones - adult kids
 - d. Affected person - status / health / condition
 - e. Family communication: *Who* to tell – *how* to tell results to others
2. Time alone with partner / support person
3. Effect on:
 - the family / dynamics
 - normal parent / partner
 - siblings
 - children
 - parents
4. Referral to Neurology (if positive result, or if negative result in the presence of symptoms)
5. Referrals: MJD Foundation / Local care worker / Psychologist
6. Follow up:
 - Health care provider (~2 weeks)
 - Visiting MJD Foundation Medical Education provider (~3 months)

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