

## Survey on genetic diagnosis of myotonic dystrophy type 1

**Question 0: Consent for participation** (if you consent to participate, please check the box below)

- I understand the purpose of the study and consent to complete the survey.

**Question 1: What is your specialty?**

- |   |   |
|---|---|
| <input type="checkbox"/> 1 Neurology                      | <input type="checkbox"/> 2 Internal Medicine (excluding Neurology)          |
| <input type="checkbox"/> 3 Pediatric Neurology            | <input type="checkbox"/> 4 Pediatric (excluding Pediatric Neurology)        |
| <input type="checkbox"/> 5 Rehabilitation                 | <input type="checkbox"/> 6 Obstetrics and Gynecology                        |
| <input type="checkbox"/> 7 Orthopedics                    | <input type="checkbox"/> 8 Neurosurgery                                     |
| <input type="checkbox"/> 9 Dermatology                    | <input type="checkbox"/> 10 Surgery (excluding Orthopedics or Neurosurgery) |
| <input type="checkbox"/> 11 Oncology                      | <input type="checkbox"/> 12 General Genetic Medicine                        |
| <input type="checkbox"/> 13 Others (                    ) |   |

**Question 2: Have you been involved in genetic diagnosis (e.g. genetic testing, genetic counseling) of patients with myotonic dystrophy type 1 (DM1) as a clinical geneticist?**

- 1 Yes, currently.  
⇒Please go to question 3.
- 2 Yes, but only in the past.  
⇒Please go to question 3. (If difficult to answer, please skip to question 12.)
- 3 No  
⇒Please skip to question 12. (If you are able to answer the questions assuming that you work with DM1 patients, please go to question 3.)

**Question 3: Which of the following do you perform in genetic testing for the purpose of obtaining a definitive diagnosis for affected patients with DM1? (choose more than one if applicable)**

- 1 Explain the benefits and potential issues of genetic testing prior to the examination.
- 2 Explain that the information may be relevant to the patients, as well as their family.
- 3 Obtain written consent from the patient or the legal representative.
- 4 Explain that the information obtained is needed if the patients wish to proceed with prenatal diagnosis or preimplantation genetic diagnosis.
- 5 Explain that the information obtained may be used as a criterion for participation in a clinical trial or treatment when a novel treatment is developed.

(\*As of April 2020, there are currently no medications for DM1 that are covered by insurance. However, clinical trials are ongoing in Japan and around the world.)

**Question 4: If a patient is diagnosed with DM1, do you explain to at-risk family members (asymptomatic) about the potential genetic risk?**

- 1  Yes, in principle
- 2  Yes, depending on the situation
- 3  No

**Question 5: What is your opinion about providing a genetic diagnosis to individuals (adults with sufficient capability for judgment) who are genetically at risk but asymptomatic? (select multiple, if applicable)**

- 1  Genetic diagnosis should not be performed for asymptomatic individuals.
- 2  Genetic diagnosis may be performed to help them prepare for developing motor dysfunction.
- 3  Genetic diagnosis may be performed to enable early detection and treatment of multiple organs complications.
- 4  Genetic diagnosis may be performed for female individuals if they are planning to have children.
- 5  Genetic diagnosis may be performed for male individuals if they are planning to have children.
- 6  Genetic diagnosis may be performed to ensure that they have the opportunity to receive any treatments that may become available in the future. (\*As of April 2020, there are currently no medications for DM1 that are covered by insurance. However, clinical trials are ongoing in Japan and around the world.)

**Question 6: What is your opinion on providing a genetic diagnosis to individuals (children without sufficient capability for judgment) who are genetically at risk but asymptomatic? (select multiple, if applicable)**

- 1  Genetic diagnosis should not be performed even if their parents or legal representatives wish to receive it.
- 2  Genetic diagnosis may be performed if their parents or legal representative wish to receive it.
- 3  Genetic diagnosis should be performed only after the individuals become old enough and gain the ability to make the judgement.
- 4  Genetic diagnosis may be performed with approval from the parents or legal representatives if the individuals are not expected to gain the ability to make the judgement.
- 5  Genetic diagnosis may be performed to enable early detection of multiple organs complications and to help them prepare for developing motor dysfunction.
- 6  Genetic diagnosis may be performed to ensure that they have the opportunity to receive any treatments that may become available in the future. (\*As of April 2020, there are

currently no medications for DM1 that are covered by insurance. However, clinical trials are ongoing in Japan and around the world.)

**Question 7: What information do you provide when your clients are given a genetic diagnosis of DM1? (select multiple, if applicable)**

- 1  Explain about potential risk of onset and progression of motor dysfunction.
- 2  Explain about potential risk of multiple organs complications.
- 3  Explain about potential risk for sudden death.
- 4  Recommend regular visits to specialists.
- 5  Explain that DM1 is an autosomal dominant disease, and that they need to undergo genetic consultation before planning for pregnancy.
- 6  Explain the genetic anticipation phenomenon (that the disease tends to become more severe in the next generation).
- 7  Explain the social systems that are applicable, such as measures against intractable diseases and welfare services for the persons with disabilities.
- 8  Explain that there is a patient registry (Remudy).
- 9  Do not provide any specific explanations.

**Question 8: How do you perform prenatal diagnosis for female patients with DM1?**

- 1  Both genetic counseling and prenatal diagnosis are performed at my institution.
- 2  Genetic counseling is performed at my institution, but prenatal diagnosis is outsourced.
- 3  Prenatal diagnosis is performed at my institution, but genetic counseling is outsourced.
- 4  Both genetic counseling and prenatal diagnosis are outsourced.
- 5  Not dealing with prenatal diagnosis.

**Question 9: What do you do when male DM1 patients request prenatal diagnosis?**

- 1  Prenatal diagnosis does not apply to male DM1 patients and should not be performed.
- 2  Prenatal diagnosis should also apply to male DM1 patients.
  - ⇒ Please answer these follow-up questions.
  - 3  Both genetic counseling and prenatal diagnosis are performed at my institution.
  - 4  Genetic counseling is performed at my institution, but prenatal diagnosis is outsourced.
  - 5  Prenatal diagnosis is performed at my institution, but genetic counseling is outsourced.
  - 6  Both genetic counseling and prenatal diagnosis are outsourced.

**Question 10: What is your opinion on preimplantation genetic diagnosis for DM1 patients?**

- 1  Preimplantation genetic diagnosis should not be performed.
- 2  Preimplantation genetic diagnosis should only be applicable to female patients and not

male patients.

- 3  It should be performed regardless of the sex of the patient, if they wish to undergo the procedure.
- 4  Criteria for individual ethics review should be relaxed to facilitate the selection of the procedure.

**Question 11: Please provide the approximate number of DM patients for whom genetic diagnosis was performed in the last year.**

- 1  None
- 2  1-5
- 3  6-10
- 4  11-20
- 5  20-40
- 6  41 or more

**Question 12: If you receive a request for genetic diagnosis of DM1 patients, will you perform the procedure yourself?**

- 1  I will perform the diagnosis.
- 2  I will only perform the diagnosis in consultation with a DM1 specialist.
- 3  I will refer the patient to another physician.

**Question 13: How many years of experience do you have as a physician?**

- 1  6-10 years
- 2  11-15 years
- 3  16-20 years
- 4  21-25 years
- 5  25-30 years
- 6  31-35 years
- 7  36-40 years
- 8  41-45 years
- 9  46 years or more

**Question 14: Where do you work?**

(            ) prefecture

**Question 15: Please choose what best describes your primary institution.**

- 1  University Hospital
- 2  National Hospital Organization
- 3  General Hospital
- 4  Clinic
- 5  Others

**Question 16: Is your primary institution part of the National Liaison Council for Clinical Sections of Medical Genetics?**

- 1  Yes
- 2  No

**Question 17: Please comment if you have any opinions about genetic diagnosis of DM1 (Please describe).**

This is the end of the survey. Thank you for your cooperation.