Oculopharyngeal Muscular Dystrophy Registry Questionnaire

The purpose of this form is to collect information from individuals who have oculopharyngeal muscular dystrophy (OPMD) or who are related by blood to someone with OPMD. **Please return this form within 3 weeks if at all possible.** If you have any questions about this form please contact us:

OPMD Registry
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Web site: http://som.unm.edu/programs/opmd

1. Please enter today's date: ____ Month ____ Day ______ Year

2. What is your name and contact information?

NAME: ____
First Middle (Maiden) Last

ADDRESS: ____
Street

____
City State Zip Code

____
Country

EMAIL ADDRESS: ____

TELEPHONE: Primary: (_____) ________________
Area Code Number

Secondary: (_____) ________________
Area Code Number

3. Are you completing this form all by yourself?

○ Yes
○ No, someone is helping me.

If someone is helping you complete this form, please indicate the helper's relationship to you by checking the appropriate circle:

○ Spouse
○ Daughter/Son
○ Brother/Sister
○ Aunt/Uncle
○ Other relative
○ Parent (biologic, adoptive, or step)
○ Grandparent
○ Legal guardian
○ Foster parent
○ Medical caregiver
○ Other non-relative

4. Please provide the name, address, and telephone number of **two** family members or friends we can contact in case you move or change your phone number.

Contact Person 1:

NAME: ____
First Middle Last

RELATIONSHIP TO YOU: ______________________________

ADDRESS: ____
Street

____
City State Zip Code

____
Country

TELEPHONE: (_____) ________________
Area Code Number

Contact Person 2:

NAME: ____
First Middle Last

RELATIONSHIP TO YOU: ______________________________

ADDRESS: ____
Street

____
City State Zip Code

____
Country

TELEPHONE: (_____) ________________
Area Code Number

5. If we need to contact you, how do you prefer to communicate? (**Check only one**)

- ○ Written language
- ○ Spoken language
- ○ Sign language

6. By what method do you prefer to be contacted? (**Check only one**)

- ○ Email
- ○ Telephone
- ○ Postal mail

7. Where did you learn about the OPMD Registry? Check all that apply.

- ○ Internet
- ○ Support group or foundation
- ○ Patient
- ○ Word of mouth
- ○ Publication
- ○ Medical professional
- ○ Media
- ○ Other

<u>Sociodemographic Information</u>

8. What is your date of birth? ______ _____ ________
Month Day Year

9. What is your gender?

- ○ Female
- ○ Male
- ○ Other: Please describe: _______________________________

10. What is your ethnicity?

- ○ Hispanic or Latino
- ○ Not Hispanic or Latino
- ○ Prefer not to say
- ○ Unknown

11. How would you describe your race? Select one or more of the following categories:

- ○ American Indian or Alaskan Native
- ○ Asian
- ○ Black or African American
- ○ Native Hawaiian or other Pacific Islander
- ○ White
- ○ Prefer not to say
- ○ Unknown

12. What is your nationality? (Provide names of the country or countries in which you or your biological family previously lived or had ancestors.)
____

13. In what country were you born? __________________

14. In what state or province were you born? _______________________

15. In what city, town, or village were you born? ________________________

16. What type of health insurance do you currently have? Select all that apply.

- ○ Private health insurance
- ○ Medicare
- ○ Medi-gap
- ○ Medicaid
- ○ Military health care (Tricare/VA, Champ-VA)
- ○ Indian health service
- ○ State-sponsored health plan
- ○ Other government program
- ○ Single service plan (e.g. dental, vision, prescription)
- ○ No coverage
- ○ Don't know

17. What is the highest grade or level of school that you have completed or the highest degree you have received? Fill one circle:

- ○ Eighth grade or less
- ○ More than eighth grade, but did not graduate from high school
- ○ Went to a business, trade, or vocational school instead of high school
- ○ High school graduate
- ○ Completed a GED
- ○ Went to a business, trade, or vocational school after high school
- ○ Went to college, but did not graduate
- ○ Graduated from a college or university
- ○ Professional training beyond a four-year college or university
- ○ I never went to school

18. How many people live in your household (including yourself)? __________

19. What is your current yearly household income in US dollars, before taxes?

- ○ Less than $10,000
- ○ $10,000-$14,999
- ○ $15,000-$19,999
- ○ $20,000-$24,999
- ○ $25,000-$29,999
- ○ $30,000-$34,999
- ○ $35,000-$39,999
- ○ $40,000-$44,999
- ○ $45,000-$49,999
- ○ $50,000-$54,999
- ○ $55,000-$59,999
- ○ $60,000-$74,999
- ○ $75,000-$84,999
- ○ $85,000-$99,999
- ○ $100,000-$149,999
- ○ $150,000-$199,999
- ○ $200,000-$249,999
- ○ $250,000 and above
- ○ Prefer not to answer

20. Have symptoms of OPMD affected your employment/ability to work?

- ○ Yes
- ○ No

If yes, how was your employment affected? Check all that apply:

- ○ I could not apply for some jobs
- ○ I lost a job
- ○ Job was modified to accommodate my physical limitations
- ○ I had to go on disability
- ○ I had to take early retirement

Symptoms of OPMD:

21. Do you have eyelid drooping (ptosis), or have you had surgery to correct eyelid drooping?

○ Yes
○ No

If yes, give your age when you first experienced eyelid drooping. Give your best estimate even if you are not sure. ______________years old

22. Do you have difficulty swallowing (dysphagia)?

○ Yes
○ No

If yes, give your age when you first experienced difficulty swallowing. Give your best estimate even if you are not sure. ______________years old

23. Do you have muscle weakness in your legs?

○ Yes
○ No

If yes, give your age when you first experienced muscle weakness in your legs. Give your best estimate even if you are not sure. ______________years old

24. Do you have muscle weakness in your arms?

○ Yes
○ No

If yes, give your age when you first experienced muscle weakness in your arms. Give your best estimate even if you are not sure. ______________years old

25. How long does it take you to eat an average meal? (Check only one answer.)

○ Less than 15 minutes
○ About 15-30 minutes
○ About 30-45 minutes
○ About 45-60 minutes
○ More than 60 minutes
○ Unable to swallow at all

26. Have you been formally diagnosed with OPMD?

- ○ Yes
- ○ No

If yes, what was your your age when you were formally diagnosed with OPMD? Give your best estimate even if you are not sure.______________years old

27. If you answered yes to question 26, please answer the following questions (27a-b, and 28). Otherwise, skip to question 29.

a. How was your diagnosis of OPMD made? Check all that apply.

- ☐ I diagnosed myself based on my symptoms and a history of OPMD in my family
- ☐ My doctor diagnosed me based on my symptoms and family history
- ☐ I had DNA testing (genetic testing, usually on a blood sample)
- ☐ I had a muscle biopsy that showed I have OPMD
- ☐ I do not know how the diagnosis was made
- ☐ Other (please describe): ______________________

b. If a doctor diagnosed you with OPMD, please describe the type of doctor. Check all that apply.

- ☐ Primary care physician
- ☐ Neurologist
- ☐ Specialist in a neuromuscular clinic or Muscular Dystrophy Clinic
- ☐ Ophthalmologist (eye doctor)
- ☐ Gastroenterologist (digestive system doctor)
- ☐ Otolaryngologist (ear, nose, and throat doctor)
- ☐ Other (please describe): ______________________

28. Have you had any of the following procedures to treat OPMD? (**If yes**, give the **YEAR or YEARS** of the procedure in the space provided. Give your best estimate of the year even if you are not sure.)

	Yes	**No**	If yes, number of times	If yes, year (or years if you had multiple procedures)
Eyelid surgery to treat eyelid drooping				
Esophageal dilatation (stretching of the throat muscles by passage of a tube into the throat)				
Botulinum toxin (Botox) injection into the throat muscles				
Cricopharyngeal myotomy (surgery to cut the throat muscles)				

29. Do you use any type of medical or assistive device to help you move around?

- ○ Yes
- ○ No

If yes, check all that apply:

- ○ Cane or walking stick
- ○ Crutches
- ○ Walker
- ○ Scooter
- ○ Wheelchair

30. Do you now take any food or liquid through a feeding tube?

- ○ Yes
- ○ No

If yes, give the date that the feeding tube was placed:

_____ Month _____ Day _______ Year

31. Have you ever received any of the following? Check all that apply.

- ○ Swallow therapy
- ○ Speech therapy
- ○ Physical therapy
- ○ Genetic counseling
- ○ Other type of therapy Describe: ____________________

Previous medical testing:

32. Have you had a muscle biopsy (this is a surgical procedure in which an incision is made in the skin and a small piece of muscle tissue is removed and later examined with a microscope)?

- ○ Yes
- ○ No

Where was the muscle biopsy done (give name of medical center and city/state)?____

What year was the muscle biopsy done? _____________

33. Have you had DNA testing (genetic testing) for OPMD?

- ○ Yes
- ○ No

Which doctor ordered the DNA test (give name of doctor, clinic, and city/state where you had the test done)? ____
____

What year was the DNA testing done? _______________

If you know the result, please describe here: ____________________

Please include a copy of your DNA test result, if you have it.

Family history:

34. Are any of your biological family members (those related to you by blood) affected with OPMD?

- ○ Yes
- ○ No

If yes, indicate which relatives have OPMD or had OPMD (check all that apply)

- ○ Mother
- ○ Father
- ○ Brother
- ○ Sister
- ○ Maternal grandmother
- ○ Maternal grandfather
- ○ Paternal grandmother
- ○ Paternal grandfather
- ○ Niece
- ○ Nephew
- ○ Granddaughter
- ○ Grandson
- ○ Son
- ○ Daughter
- ○ Maternal uncle
- ○ Maternal aunt
- ○ Paternal uncle
- ○ Paternal aunt
- ○ Paternal cousin
- ○ Maternal cousin
- ○ Half-brother
- ○ Half-sister

35. For women only: Have you ever been pregnant?

- ○ Yes
- ○ No

If yes, please state the number of times you have been pregnant (count the number of pregnancies even if you did not have the baby): _________

If yes, how many children did you give birth to? (count all live births): _________

General health:

36. What is your weight? ______ pounds

37. What is your height? ____feet ____inches

38. Have you ever had aspiration pneumonia?

- ○ Yes
- ○ No

If yes, how many times? ______

39. In general, would you say your health is . . .

- ○ Excellent
- ○ Very good
- ○ Good
- ○ Fair
- ○ Poor

40. Does your health now limit you in doing vigorous activities?

- ○ Not at all
- ○ Very little
- ○ Somewhat
- ○ Quite a lot
- ○ Cannot do

41. How much does pain interfere with your enjoyment of life?

- ○ Not at all
- ○ A little bit
- ○ Somewhat
- ○ Quite a bit
- ○ Very much

42. How often do you feel tired?

- ○ Never
- ○ Rarely
- ○ Sometimes
- ○ Often
- ○ Always

43. I feel depressed . . .

- ○ Never
- ○ Rarely
- ○ Sometimes
- ○ Often
- ○ Always

<u>Please answer these questions even if you have not been diagnosed with a swallowing problem.</u> (For example, you may select “not true” or “never” if you do not experience the problem). **Circle one number on each line**.

IMPORTANT NOTE: We understand that you may have a number of physical problems. Sometimes it is hard to separate these from swallowing difficulties, but we hope that you can do your best to concentrate ***only*** on your ***swallowing problem***.

(Source: SWAL-QOL, McHorney et al.)

44. Below are some general statements that people with ***swallowing problems*** might mention. In the last month, **<u>how true</u>** have the following statements been for you?

(circle one number on each line)

	Very much true	Quite a bit true	Somewhat true	A little true	Not at all true
Dealing with my swallowing problem is very difficult.	1	2	3	4	5
My swallowing problem is a major distraction in my life.	1	2	3	4	5

45. Below are aspects of day-to-day eating that people with ***swallowing problems*** sometimes talk about. In the last month, **<u>how true</u>** have the following statements been for you?

(circle one number on each line)

	Very much true	Quite a bit true	Somewhat true	A little true	Not at all true
Most days, I don’t care if I eat or not.	1	2	3	4	5
It takes me longer to eat than other people.	1	2	3	4	5
I’m rarely hungry anymore.	1	2	3	4	5
It takes me forever to eat a meal.	1	2	3	4	5
I don’t enjoy eating anymore.	1	2	3	4	5

46. Below are some physical problems that people with ***swallowing problems*** sometimes experience. In the last month, **how often** have you experienced each problem as a result of your swallowing problem?

(circle one number on each line)

	Almost always	Often	Sometimes	Hardly ever	Never
Coughing	1	2	3	4	5
Choking when you eat food	1	2	3	4	5
Choking when you take liquids	1	2	3	4	5
Having thick saliva or phlegm	1	2	3	4	5
Gagging	1	2	3	4	5
Drooling	1	2	3	4	5
Problems chewing	1	2	3	4	5
Having excess saliva or phlegm	1	2	3	4	5
Having to clear your throat	1	2	3	4	5
Food sticking in your throat	1	2	3	4	5
Food sticking in your mouth	1	2	3	4	5
Food or liquid dribbling out of your mouth	1	2	3	4	5
Food or liquid coming out your nose	1	2	3	4	5
Coughing food or liquid out of your mouth when it gets stuck	1	2	3	4	5

47. Next, please answer a few questions about how your ***swallowing problem*** has affected your diet and eating in the last month.

(circle one number on each line)

	Strongly agree	Agree	Uncertain	Disagree	Strongly disagree
Figuring out what I can and can't eat is a problem for me.	1	2	3	4	5
It is difficult to find foods that I both like and can eat.	1	2	3	4	5

48. In the last month, **how often** have the following statements about communication applied to you because of your ***swallowing problem***?

(circle one number on each line)

	All of the time	Most of the time	Some of the time	A little of the time	None of the time
People have a hard time understanding me.	1	2	3	4	5
It's been difficult for me to speak clearly.	1	2	3	4	5

49. Below are some concerns that people with ***swallowing problems*** sometimes mention. In the last month, **how often** have you experienced each feeling?

(circle one number on each line)

	Almost always	Often	Sometimes	Hardly ever	Never
I fear I may start choking when I eat food.	1	2	3	4	5
I worry about getting pneumonia.	1	2	3	4	5
I am afraid of choking when I drink liquids.	1	2	3	4	5
I never know when I am going to choke.	1	2	3	4	5

50. In the last month, how often have the following statements **<u>been true</u>** for you because of your ***swallowing problem***?

(circle one number on each line)

	Always true	Often true	Sometimes true	Hardly ever true	Never true
My swallowing problem depresses me.	1	2	3	4	5
Having to be so careful when I eat or drink annoys me.	1	2	3	4	5
I have been discouraged by my swallowing problem.	1	2	3	4	5
My swallowing problem frustrates me.	1	2	3	4	5
I get impatient dealing with my swallowing problem.	1	2	3	4	5

51. Think about your social life in the last month. How strongly would you agree or disagree with the following statements?

(circle one number on each line)

	Strongly agree	Agree	Uncertain	Disagree	Strongly disagree
I do not go out to eat because of my swallowing problem.	1	2	3	4	5
My swallowing problem makes it hard to have a social life.	1	2	3	4	5
My usual work or leisure activities have changed because of my swallowing problem.	1	2	3	4	5
Social gatherings (like holidays and get-togethers) are not enjoyable because of my swallowing problem.	1	2	3	4	5
My role with family and friends has changed because of my swallowing problem.	1	2	3	4	5

52. In the last month, **how often** have you experienced each of the following physical symptoms?

(circle one number on each line)

	All of the time	Most of the time	Some of the time	A little of the time	None of the time
Feel weak?	1	2	3	4	5
Have trouble falling asleep?	1	2	3	4	5
Feel tired?	1	2	3	4	5
Have trouble staying asleep?	1	2	3	4	5
Feel exhausted?	1	2	3	4	5

53. Please circle the letter of the one description below that best describes the consistency or texture of the food you have been eating most often in the last week.

Circle one:

A. Circle this one if you are eating a full normal diet, which would include a wide variety of foods, including hard to chew items like steak, carrots, bread, salad, and popcorn.

B. Circle this one if you are eating soft, easy to chew foods like casseroles, canned fruits, soft cooked vegetables, ground meat, or cream soups.

C. Circle this one if you are eating food that is put through a blender or food processor or anything that is like pudding or pureed foods.

D. Circle this one if you take most of your nutrition by tube, but sometimes eat ice cream, pudding, apple sauce, or other pleasure foods.

E. Circle this one if you take all of your nourishment through a tube.

54. Please circle the letter of the one description below that best describes the consistency of liquids you have been drinking most often in the last week.

Circle one:

A. Circle this if you drink liquids such as water, milk, tea, fruit juice, and coffee.

B. Circle this if the majority of liquids you drink are thick, like tomato juice or apricot nectar. Such thick liquids drip off your spoon in a slow steady stream when you turn it upside down.

C. Circle this if your liquids are moderately thick, like a thick milkshake or smoothie. Such moderately thick liquids are difficult to suck through a straw, like a very thick milkshake, or drip off your spoon slowly drop by drop when you turn it upside down, such as honey.

D. Circle this if your liquids are very thick, like pudding. Such very thick liquids will stick to a spoon when you turn it upside down, such as pudding.

E. Circle this if you did not take any liquids by mouth or if you have been limited to ice chips.

55. The following questions ask about your ability to move around. Please answer based on your **current** condition (how you are doing **today**). Please respond to the items below by circling one number per row.

(Source: PROMIS, NeuroQol)

	Without any difficulty	With a little difficulty	With some difficulty	With much difficulty	Unable to do
Are you able to get on and off the toilet?	5	4	3	2	1
Are you able to step up and down curbs?	5	4	3	2	1
Are you able to get in and out of a car?	5	4	3	2	1
Are you able to get out of bed into a chair?	5	4	3	2	1
Are you able to push open a heavy door?	5	4	3	2	1
Are you able to run errands and shop?	5	4	3	2	1
Are you able to get up off the floor from lying on your back without help?	5	4	3	2	1
Are you able to go for a walk of at least 15 minutes?	5	4	3	2	1
Are you able to squat and get up?	5	4	3	2	1
Are you able to climb up 5 steps?	5	4	3	2	1
Are you able to run a short distance, such as to catch a bus?	5	4	3	2	1

Interest in future studies on OPMD:

56. Have you previously participated in any clinical trials related to OPMD?

- ○ Yes
- ○ No

57. Are you currently participating in any clinical trials related to OPMD?

- ○ Yes
- ○ No

58. Are you willing to be contacted in the future about research related to OPMD?

- ○ Yes
- ○ No

59. Have you donated a sample of blood, tissue, or other biospecimen for research in the past?

- ○ Yes
- ○ No

a. **If yes**, what type was it?

- ○ Blood
- ○ Saliva/Cheek Swab
- ○ Urine
- ○ Other bodily fluid
- ○ Tissue

b. What is the name of the hospital/clinic where the biospecimen was donated? ____________________

60. Are you willing to be contacted about donating a sample of blood, tissue, or other biospecimen for research in the future?

- ○ Yes
- ○ No

You have reached the end of the OPMD Registry Questionnaire. Thank you for taking the time to complete this form. After we receive your questionnaire, we may contact you to clarify some of your responses. We appreciate your efforts!

COMMENTS: Do you have any comments about this questionnaire? We welcome any feedback, especially regarding questions that were unclear or confusing. Also, feel free to include any comments or opinions regarding future research on OPMD.