

**MECP2 Duplication Syndrome Study**  
**Australian Paediatric Surveillance Unit**

Please contact Dr Helen Leonard 0419 956 946 ([helen.leonard@telethonkids.org.au](mailto:helen.leonard@telethonkids.org.au)) if you have any questions about this form.

*Instructions: Please answer each question by ticking the appropriate box or writing your response in the space provided. DK=Don't Know*

**REPORTING CLINICIAN'S DETAILS:** 1. APSU Dr Code/Name:     /  2. Month/Year of Report:   /

3. Date questionnaire completed:   /   /   4. Paediatric or adult physician: ☐ Paediatric ☐ Adult

5. Specialty: ☐ Clinical geneticist ☐ Neurologist ☐ Developmental Paediatrician ☐ General Paediatrician ☐ Other

**PATIENT DETAILS:** 6. First 2 letters of first name:   7. First 2 letters of surname:   8. Date of Birth:   /   /

9. Sex: ☐ M ☐ F 10. Postcode of family:     11. Country of birth:       ☐ DK

If this patient is primarily cared for by another physician who you believe will report the case, please complete the questionnaire details above this line and return to APSU. Please keep the patient's name and other details in your records. If no other report is received for this child we will contact you for information requested in the remainder of the questionnaire.

The primary clinician caring for this child is: **Name:**

**Hospital:**

**GENERAL INFORMATION:**

12. How old was the patient when you first met him/her?   month(s)   year(s)

13. How old was the patient when their parents first had concerns?   month(s)   year(s)

14. What was the early clinical presentation? Please tick all items that apply.

- ☐ Developmental delay ☐ Hypotonia ☐ Breathing problems ☐ Stereotypic movements  
☐ Feeding problems ☐ Dysmorphic features ☐ Health problems (specify)   
☐ Others (specify)

15. What was your main reason for requesting Array Comparative Genomic Hybridization (CGH) testing?

16. Had you heard of MECP2 duplication syndrome prior to seeing this patient? ☐ Yes ☐ No

**INVESTIGATIONS:** Please forward a de-identified copy of the genetic report with details of MECP2 duplication with this questionnaire

17. Are there any other investigations you have requested for the patient?

- ☐ Karyotype testing ☐ Magnetic Resonance Imaging (MRI Scan) ☐ Electroencephalogram (EEG)  
☐ Other genetic tests or investigations (specify)

**GROSS MOTOR MILESTONE AND MUSCLE TONE:**

18. Did the patient learn to do the following independently? If yes, at what age?

- a. Sit: ☐ Yes -   month(s) ☐ No ☐ DK      b. Crawl: ☐ Yes -   month(s) ☐ No ☐ DK  
c. Walk: ☐ Yes -   month(s) ☐ No ☐ DK

19. Has there been any regression of these skills? If yes, at what age? ☐ Yes -   month(s)   year(s) ☐ No ☐ DK

20. Currently, if the patient is able to walk, is the gait ataxic? ☐ Yes ☐ No ☐ DK

21. If no longer walking, at what age did the patient lose the ability to walk?   month(s)   year(s)

22. Please indicate current muscle tone. ☐ Normal ☐ Increased ☐ Reduced (hypotonic)

23. If not normal, please indicate how affected. ☐ Peripheral ☐ Axial ☐ Both ☐ DK

24. Is muscle tone different between arms and legs? ☐ Yes ☐ No

a. If yes, how?

25. Has muscle tone change over time? ☐ Yes ☐ No

**DEVELOPMENTAL AND BEHAVIOURAL FEATURES:**

26. If your patient is younger than 5 years, is there global developmental delay? ☐ Yes ☐ No ☐ DK

27. If your patient is 5 years or older, what is the level of intellectual disability? ☐ None ☐ Mild ☐ Moderate ☐ Severe

28. What is/are the communication skill(s) currently present? Please tick all that apply.

- ☐ Cooing ☐ Babble ☐ Single words ☐ Phrases ☐ 4 or more sentences

29. Has the patient ever been diagnosed with autism? ☐ Yes ☐ No ☐ DK

30. Does the patient have any of the followings? Please tick all that apply.

- ☐ Difficulty using eye gaze for social interactions ☐ Gaze avoidance ☐ Repetitive behaviours ☐ Compulsive behaviours  
☐ Anxiety ☐ Hand stereotypies ☐ Difficulty adjusting to change

**MEDICAL HISTORY:**

31. Has the patient suffered from the following conditions? Please tick all that apply. If condition was present, please indicate number of episodes since birth (tick one box only).

- ☐ Pneumonia: ☐ 0-3 ☐ 4-6 ☐ 7-9 ☐ >9      ☐ Bronchiolitis: ☐ 0-3 ☐ 4-6 ☐ 7-9 ☐ >9  
☐ Other respiratory infection: ☐ 0-3 ☐ 4-6 ☐ 7-9 ☐ >9      ☐ Asthma: ☐ 0-3 ☐ 4-6 ☐ 7-9 ☐ >9  
☐ Ear infection: ☐ 0-3 ☐ 4-6 ☐ 7-9 ☐ >9

32. Please list any respiratory pathogens identified if known: \_\_\_\_\_

33. Does the patient have the followings? Please tick all that apply.

- ☐ Apnoea whilst awake    ☐ Sleep apnoea    ☐ Shallow breathing    ☐ Breathlessness    ☐ Difficulty swallowing  
☐ Gastro-oesophageal reflux    ☐ Constipation    ☐ Abdominal bloating

34. Does the patient have a gastrostomy? ☐ Yes ☐ No ☐ DK

35. Does the patient have a seizure disorder? ☐ Yes ☐ No ☐ DK

a. If yes, what was the age of onset? ☐ ☐ month(s) ☐ ☐ year(s)

**OTHER CLINICAL FEATURES:**

Please tick the box for each sign that your patient may or may have had.

	Occurring now	Occurring previously	Has never occurred	DK
36. Teeth grinding	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
37. Uncontrolled screaming spells	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
38. Sleep disturbances	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
39. Night laughing	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
40. Choreiform movements	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
41. Scoliosis	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

**MEDICATIONS:**

42. Please list the medication(s) your patient is currently using.

**DYSMORPHIC FEATURES:**

43. Does the patient have any dysmorphic features? ☐ Yes ☐ No

If yes, please describe in the space provided.

- a. Head (e.g. macrocephaly, microcephaly, plagiocephaly) \_\_\_\_\_  
 b. Face (e.g. Depressed nasal bridge, thick lips, prognathism) \_\_\_\_\_  
 c. Hands (e.g. tapering fingers) \_\_\_\_\_  
 d. Genitals (e.g. cryptorchidism, hypospadias, micropenis) \_\_\_\_\_  
 e. Feet (e.g. long toes) \_\_\_\_\_  
 f. Other body part \_\_\_\_\_

**FAMILY HISTORY:**

44. Does the patient have any relatives diagnosed with intellectual disability, autism or other developmental disabilities?

☐ Yes ☐ No

If yes, please complete the following table.

Family member	Intellectual disability	Autism	Unexplained death	Other (specify)
a. Mother	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
b. Father	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
c. Brother(s)	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
d. Sister(s)	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
e. Uncle	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
f. Aunt	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

45. Has testing for MECP2 duplication been requested for other family members? ☐ Yes ☐ No

If yes, what was the result? \_\_\_\_\_

**Thank you for your help with this research project. Please return this questionnaire by email to [Helen.Leonard@telethonkids.org.au](mailto:Helen.Leonard@telethonkids.org.au).**

The Australian Paediatric Surveillance Unit is affiliated with the Royal Australasian College of Physicians (Paediatrics and Child Health Division) and Sydney Medical School, The University of Sydney. APSU is funded by the Australian Government Department of Health and Ageing.

This study has been approved by a Human Research Ethics Committee properly constituted under NHMRC guidelines