MECP2 Duplication Syndrome Study
Australian Paediatric Surveillance Unit

Please contact Dr Helen Leonard 0419 956 946 (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 the 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 MILESTONES 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 following? 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 since birth? If yes, please indicate number of episodes (tick one box only).

Pneumonia:	☐ No	☐ Yes ->	☐ 1-3	☐ 4-6	☐ 7-9	☐ >9	☐ DK
Other respiratory infection:	☐ No	☐ Yes ->	☐ 1-3	☐ 4-6	☐ 7-9	☐ >9	☐ DK
Bronchiolitis:	☐ No	☐ Yes ->	☐ 1-3	☐ 4-6	☐ 7-9	☐ >9	☐ DK
Asthma:	☐ No	☐ Yes ->	☐ 1-3	☐ 4-6	☐ 7-9	☐ >9	☐ DK
Ear infection:	☐ No	☐ Yes ->	☐ 1-3	☐ 4-6	☐ 7-9	☐ >9	☐ DK

32. Please list any respiratory pathogens identified if known: _____

33. Does the patient have the following? 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	☐	☐	☐	☐
37. Uncontrolled screaming spells	☐	☐	☐	☐
38. Sleep disturbances	☐	☐	☐	☐
39. Night laughing	☐	☐	☐	☐
40. Choreiform movements	☐	☐	☐	☐
41. Scoliosis	☐	☐	☐	☐

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	☐	☐	☐	☐
b. Father	☐	☐	☐	☐
c. Brother(s)	☐	☐	☐	☐
d. Sister(s)	☐	☐	☐	☐
e. Uncle	☐	☐	☐	☐
f. Aunt	☐	☐	☐	☐

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.

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