**Parent Survey: Biliary Tract (Gallbladder) Disease and Cancer in Rett Syndrome**

We are interested in learning about certain disorders in individuals with Rett syndrome, namely the occurrence of biliary tract (gallbladder) disease and the occurrence of cancer. Please complete this questionnaire to indicate if your child has had either of these disorders.

1. Please provide the initials of your child’s first and last name: \_\_\_\_\_\_\_

2. Please provide your child’s birthdate: \_\_\_\_/\_\_\_\_/\_\_\_\_

Mo Day Yr (2 digits)

3. Gender: Female\_\_\_\_ Male\_\_\_\_\_

4. Rett syndrome diagnosis information (choose one answer only):

\_\_\_\_Meets clinical criteria for Rett syndrome but does NOT have a known genetic mutation in *MECP2* or C*DKL5*

\_\_\_\_Meets clinical criteria AND has a genetic mutation in *MECP2* or *CDKL5*

\_\_\_\_Does NOT meet clinical criteria for Rett syndrome but HAS a genetic mutation in *MECP2 or CDKL5*

\_\_\_\_Not sure

5. What statement about genetic testing results best describes your child?

\_\_\_\_Not tested (no MECP2 or CDKL5 genetic testing done)

\_\_\_\_Tested, but no mutation found

\_\_\_\_Tested and mutation found (choose from answers below)

*MECP2* mutations:

R106W\_\_\_

R133C\_\_\_

R168X\_\_\_

R255X\_\_\_

R270X\_\_\_

R294X\_\_\_

R306C\_\_\_

R306H\_\_\_

T158M\_\_\_

Other MECP2 mutations\_\_\_\_ Please specify if known\_\_\_\_\_\_\_\_\_\_\_\_

Duplication\_\_\_

Don’t know/not sure\_\_\_\_ (you may write in the results here if you are unsure)

Non *MECP2* mutations:

CDKL5 \_\_\_

6. Has your child ever been diagnosed with cancer?

Yes\_\_\_ No\_\_\_ Don’t know\_\_\_

7. If the answer is “yes”, please indicate what type\_\_\_\_\_\_\_\_\_\_\_ and at what age (in years) \_\_\_\_\_\_\_\_\_.

8. Have you or other members of your family (NOT including your child with Rett syndrome) had gallbladder disease?

Yes\_\_\_ No\_\_\_ Don’t know\_\_\_

9. Has your child with Rett syndrome been evaluated for biliary tract (gallbladder) disease?

Yes\_\_\_ No\_\_\_ Don’t know \_\_\_

**If the answer is “NO” or “don’t know” to the question about your child’s gallbladder (# 9 ), STOP and return the survey as directed; if the answer is**

**“YES” to the question about your child’s gallbladder (question #9 ) , PLEASE COMPLETE the entire survey.**

10. DID your child EVER have OR does your child CURRENTLY HAVE biliary tract (gallbladder) disease? Yes\_\_\_ No\_\_\_ Don’t know \_\_\_

11. What was your child’s age in years when the doctor identified biliary tract disease?\_\_\_

12. What type of biliary symptoms did your daughter have?

Abdominal pain Yes\_\_\_ No\_\_\_ Don’t know\_\_\_

Vomiting Yes\_\_\_ No\_\_\_ Don’t know\_\_\_

Fever Yes\_\_\_ No\_\_\_ Don’t know\_\_\_

Irritability (pain) Yes\_\_\_ No\_\_\_ Don’t know\_\_\_

Jaundice Yes\_\_\_ No\_\_\_ Don’t know\_\_\_

Other: please specify \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

13. How long in days, months, or years (please specify) did your daughter have symptoms before she was diagnosed with biliary tract disease? \_\_\_\_\_\_\_\_\_

14. Did your child have blood work such as alanine aminotransferase (ALT), aspartate aminotransferase (AST), and/or bilirubin performed for the symptoms?

Yes\_\_\_ No\_\_\_ Don’t know\_\_\_

15. What were the results of the blood work?

ALT– Normal\_\_\_ Abnormal\_\_\_ Don’t know\_\_\_ N/A\_\_\_

AST – Normal\_\_\_ Abnormal\_\_\_ Don’t know\_\_\_ N/A\_\_\_

Bilirubin– Normal\_\_\_ Abnormal\_\_\_ Don’t know\_\_\_ N/A\_\_\_

16. Did your child have an abdominal ultrasound performed for the symptoms?

Yes\_\_\_ No\_\_\_ Don’t know\_\_\_

17. What were the results of the abdominal ultrasound?

Normal\_\_\_ Abnormal\_\_\_ Don’t know\_\_\_ N/A\_\_\_

18. Did your child have a CT scan performed for the symptoms?

Yes\_\_\_ No\_\_\_ Don’t know\_\_\_

19. What were the results of the CT scan?

Normal\_\_\_ Abnormal\_\_\_ Don’t know\_\_\_ N/A\_\_\_

20. Did your child have a HIDA (biliary) scan performed for the symptoms?

Yes\_\_\_ No\_\_\_ Don’t know\_\_\_

21. What were the results of the HIDA scan?

Normal\_\_\_ Abnormal\_\_\_ Don’t know\_\_\_ N/A\_\_\_

22. What percent of the radionuclide material was emptied from the gallbladder during the HIDA scan (also known as % function)? \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ Don’t know\_\_\_ N/A\_\_\_

23. What type of biliary tract disease did or does your child have? (check all that apply)

Cholecystitis (inflammation) Yes\_\_\_ No\_\_\_ Don’t know\_\_\_

Cholelithiasis (gallstones) Yes\_\_\_ No\_\_\_ Don’t know\_\_\_

Biliary dyskinesia (poor motility) Yes\_\_\_ No\_\_\_ Don’t know\_\_\_

24. Has your child had surgery for gallbladder disease?

Yes\_\_\_ No\_\_\_ Don’t know\_\_\_

**IF THE ANSWER TO QUESTION #24 is “NO”, SKIP TO QUESTION #41**

25. If your child had surgery for gallbladder disease, at what age in years was surgery performed? \_\_\_\_

26. What type of surgical procedure was done to remove the gallbladder?

Open surgery Yes\_\_\_ No\_\_\_ Don’t know\_\_\_

Laparoscopy Yes\_\_\_ No\_\_\_ Don’t know\_\_\_

27. Did your child have an intravenous cholecystogram (x-ray) during surgery to look for gallstones in the bile ducts?

Yes\_\_\_ No\_\_\_ Don’t know\_\_\_\_

28. If an intravenous cholecystogram was done during surgery, did the surgeon find gallstones in the bile ducts?

Yes\_\_\_ No\_\_\_ Don’t know\_\_\_ N/A\_\_\_\_

29. Did your child have any problems (complications) after gallbladder surgery?

Yes\_\_\_ No\_\_\_ Don’t know\_\_\_

30. Which of the following problems did your child have after surgery?

Fever Yes\_\_\_ No\_\_\_ Don’t know\_\_\_ N/A\_\_\_

Infection Yes\_\_\_ No\_\_\_ Don’t know\_\_\_ N/A\_\_\_

Liver problems Yes\_\_\_ No\_\_\_ Don’t know\_\_\_ N/A\_\_\_

Yellow jaundice Yes\_\_\_ No\_\_\_ Don’t know\_\_\_ N/A\_\_\_

Abdominal pain Yes\_\_\_ No\_\_\_ Don’t know\_\_\_ N/A\_\_\_

31. Did your child have an ERCP (endoscopic retrograde cholangiopancreatography) performed after gallbladder surgery to look for more gallstones in the bile ducts?

Yes\_\_\_ No\_\_\_ Don’t know\_\_\_

32. If an ERCP was performed, did the physician find additional gallstones in the bile duct?

Yes \_\_\_ No\_\_\_ Don’t know\_\_\_ N/A\_\_\_

33. Did surgery correct your child’s biliary tract symptoms?

Yes\_\_\_ No\_\_\_ Don’t know\_\_\_

34. How long in days, months or years has it been since your child has had gallbladder surgery?\_\_\_\_\_\_

35. Does you child still have symptoms at the present time?

Yes\_\_\_ No\_\_\_ Don’t know\_\_\_

36. What symptoms does your child have at the present time?

Abdominal pain Yes\_\_\_ No\_\_\_ Don’t know\_\_\_ N/A\_\_\_

Vomiting Yes\_\_\_ No\_\_\_ Don’t know\_\_\_ N/A\_\_\_

Fever Yes\_\_\_ No\_\_\_ Don’t know\_\_\_ N/A\_\_\_

Irritability Yes\_\_\_ No\_\_\_ Don’t know\_\_\_ N/A\_\_\_

Jaundice Yes\_\_\_ No\_\_\_ Don’t know\_\_\_ N/A\_\_\_

Other – please specify\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

37. Are these symptoms of the same **frequency** as before surgery?

Yes\_\_\_ No\_\_\_ Don’t know\_\_\_

38. If the answer is “no”, indicate for each of the symptom whether the frequency is less or more often than before surgery.

Abdominal pain Less often \_\_\_ More often \_\_\_

Vomiting Less often \_\_\_ More often \_\_\_

Fever Less often \_\_\_ More often \_\_\_

Irritability Less often \_\_\_ More often \_\_\_

Jaundice Less often \_\_\_ More often \_\_\_

39. Are these symptoms of the same **severity** as the ones before surgery?

Yes\_\_\_ No\_\_\_ Don’t know\_\_\_

40. If the answer was “no”, indicate for each symptom whether the severity is more or less severe than before surgery.

Abdominal pain Less severe \_\_\_ More severe \_\_\_

Vomiting Less severe \_\_\_ More severe \_\_\_

Fever Less severe \_\_\_ More severe \_\_\_

Irritability Less severe \_\_\_ More severe \_\_\_

Jaundice Less severe \_\_\_ More severe \_\_\_

41. Would you be willing to have the Baylor College of Medicine Rett Center team review your child’s medical records related to the gallbladder disease under a research protocol?

Yes\_\_\_ No\_\_\_

42. If yes, please provide an email address or telephone number where one of the researchers may contact you to provide further information or please contact the researchers directly at The Blue Bird Circle Rett Center at Baylor College of Medicine, Houston, TX, TEL: 832-822-RETT, FAX: 832-825-RETT

Email\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ Telephone\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_