

Appendix. International Rett Syndrome Foundation Gastrointestinal, Nutrition, and Seizure Survey

Subject Number _____

- 1) What is your daughter's age (in years)?
☐ 0-1 yr
☐ 2-5 yr
☐ 6-10 yr
☐ 11-14 yr
☐ 15-19 yr
☐ 20-29 yr
☐ 30-39 yr
☐ 40 + yr
- 2) Is your daughter positive for a MECP2 mutation?
☐ Yes ☐ No ☐ Don't know
- 3) What is your daughter's weight?
_____ lb ☐ Don't know
- 4) How tall is your daughter?
_____ ft/in ☐ Don't know
- 5) What is your daughter's nutritional status?
☐ Underweight
☐ Normal weight
☐ Overweight
- 6) Is your daughter short for her age?
☐ Yes ☐ No ☐ Don't know
- 7) Do you currently give multivitamin and/or mineral supplements to your daughter?
☐ Yes ☐ No ☐ Don't know
- 8) Do you currently give herbal preparations or health food supplements to your daughter?
☐ Yes ☐ No ☐ Don't know
- 9) Do you currently give a commercial formula preparation (e.g. Pediasure, Ensure, Peptomen, Carnation Instant Breakfast, Scandishake) to your daughter?
☐ As a supplement
☐ As her primary food source
☐ Currently do not use formula preparations

10) Does your daughter have a gastrostomy button?
☐ Yes ☐ No ☐ Don't know

11) Does your daughter have a fundoplication?
☐ Yes ☐ No ☐ Don't know

11) Does your daughter have gastroesophageal (acid) reflux?
☐ Yes ☐ No ☐ Don't know

12) Does your daughter have delayed gastric emptying?
☐ Yes ☐ No ☐ Don't know

13) Does your daughter vomit or regurgitate frequently?
☐ Yes ☐ No ☐ Don't know

14) Does your daughter wake at night, crying inconsolably?
☐ Yes ☐ No ☐ Don't know

15) Has your daughter had an UGI series (x-ray) performed?
☐ Yes ☐ No ☐ Don't know

16) Has your daughter had an upper endoscopy (internal exam of stomach with a telescope-like tube under sedation) performed?
☐ Yes ☐ No ☐ Don't know

17) Has your daughter had a gastric emptying (nuclear medicine) study performed?
☐ Yes ☐ No ☐ Don't know

18) Does your daughter have gallbladder disease?
☐ Yes ☐ No ☐ Don't know

If your daughter has gallbladder disease, does she have gallstones? ☐ Yes
☐ No ☐ Don't know

If your daughter has gallbladder disease, does she have biliary dyskinesia (abnormal motility)?
☐ Yes ☐ No ☐ Don't know

Has your daughter had gallbladder surgery?
☐ Yes ☐ No ☐ Don't know

19) Has your daughter had an abdominal ultrasound of her abdomen?
☐ Yes ☐ No ☐ Don't know

20) Does your daughter have chewing problems?
☐ Yes ☐ No ☐ Don't know

21) How long does it take to feed your child?

Less than 15 minutes ☐

15-20 minutes ☐

30-35 minutes ☐

45-50 minutes ☐

60 minutes or more ☐

22) Does your daughter have swallowing difficulties?

☐ Yes ☐ No ☐ Don't know

23) Does your daughter choke or gag frequently with feeding?

☐ Yes ☐ No ☐ Don't know

24) Has your daughter had a swallowing function study to assess her swallowing ability?

☐ Yes ☐ No ☐ Don't know

25) Does your daughter have constipation?

☐ Yes ☐ No ☐ Don't know

26) Does your daughter have straining or difficulty emptying her bowels?

☐ Yes ☐ No ☐ Don't know

27) Does your daughter have extra firm or hard stools?

☐ Yes ☐ No ☐ Don't know

28) Has your daughter had a colonoscopy (internal exam of large bowel with a telescope-like tube under sedation) performed?

☐ Yes ☐ No ☐ Don't know

29) Has your daughter had abdominal or intestinal surgery other than gallbladder surgery or gastrostomy/fundoplication?

☐ Yes ☐ No ☐ Don't know

30) Does your daughter have diabetes?

☐ Yes ☐ No ☐ Don't know

If your daughter has diabetes, does she require insulin?

☐ Yes ☐ No

31) Does your daughter have osteopenia (bone mineral loss)?

☐ Yes ☐ No ☐ Don't know

32) Has your daughter ever had a bone break or fracture?

☐ Yes ☐ No ☐ Don't know

33) Has your daughter had a DXA (bone density) scan?

☐ Yes ☐ No ☐ Don't know

34) Has your daughter ever had seizures?

☐ Yes ☐ No ☐ Don't know

If your daughter ever had seizures, does she currently have seizures?

☐ Yes ☐ No ☐ Don't know

At what age did your daughter's seizures start?

☐ Less than 10 yr of age

☐ 10-20 yr of age

☐ Greater than 20 yr of age

At what age did your daughter's seizures stop?

☐ Less than 10 yr of age

☐ 10-20 yr of age

☐ Greater than 20 yr of age

☐ Have never stopped

35) Does your daughter take medications for seizures?

☐ Never

☐ In the past, but not now

☐ Currently takes them

36) Has your daughter ever been treated with the ketogenic diet?

☐ Yes ☐ No ☐ Don't know

37) Has your daughter ever had the vagus nerve stimulator placed?

☐ Yes ☐ No ☐ Don't know