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