

## University of Edinburgh MeCP2 Mutation Database

Contact person:

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The data should be submitted using above mentioned address or e-mail.

### **Rett Syndrome questionnaire**

Please give each symptom a rating of 0, 1 or 2.  
Feel free to include comments if necessary.

Patients Name (optional):  
Date of Birth:  
*MECP2* Mutation:  
Lab that performed mutation testing:

Period of normal development:  
0 if the child had a definite normal period of development  
1 if you noticed the first signs of RS around the age of 6 months  
2 if child never developed normally

Decelerating head circumference:  
0 if head circumference has not decelerated  
1 if head circumference deceleration was noted after age 1  
2 if head circumference deceleration was noted before age 1

Loss of purposeful hand use:  
0 if child has normal hand use  
1 if child has limited hand use (eg. Can feed themselves, pick items up)  
2 if child has no hand use

Repetitive hand motions:  
0 if child has no repetitive hand motions  
1 if child has occasional repetitive hand motions  
2 if child has often/constant repetitive hand motions

Non verbal:

- 0 if child speaks normally for their age
- 1 if child regularly speaks using single words or short sentences
- 2 if child is non verbal

Ambulation:

- 0 if the child walks well independently
- 1 if the child can walk with assistance
- 2 if the child cannot walk at all

Breathing irregularities (hyperventilation, breath holding, apnea):

- 0 if child has no breathing problems
- 1 if child has occasional breathing problems
- 2 if child often/constantly has breathing problems

Seizures:

- 0 if child has no overt seizures
- 1 if child has occasional seizures or they are controlled with medications
- 2 if child has seizures often or they are not controlled with medications

Abnormal EEG:

- 0 if child's EEG is normal
- 1 if EEG has minor abnormalities
- 2 if EEG has significant abnormalities

Eating difficulties (biting, chewing, swallowing, choking):

- 0 if child has no eating difficulties
- 1 if child has minor eating difficulties
- 2 if child has major difficulties eating

Reflux:

- 0 if child has no reflux issues
- 1 if child has minor reflux or reflux is controlled with medication
- 2 if child has reflux that is not controlled with medication

Constipation:

- 0 if the child has no issues with constipation
- 1 if the problem is controlled through medication or diet
- 2 if constipation is an ongoing problem

Growth retardation:

- 0 if child is normal height/weight for age and taking account size of parents
- 1 if child is slightly below height/weight norms for age and taking account size of parents
- 2 if child is significantly below growth charts for age and taking account size of parents

Scoliosis/kyphosis:

0 if child has no scoliosis or kyphosis

1 if child has a slight curve

2 if child is wearing brace or had surgery to correct curve

Muscle rigidity/spasticity:

0 if child has no issues with rigidity/spasticity

1 if child has some degree of rigidity/spasticity

2 if child has ongoing problems with rigidity/spasticity

Small feet:

0 if child's feet size is proportionate with body size

1 if child's feet are slightly smaller in proportion to rest of the body

2 if child's feet are significantly smaller than you would expect given body size

Poor Circulation:

0 if this is not an issue

1 if child has occasional problems with circulation

2 if child has ongoing issues with circulation

Tremors:

0 if no tremor problems

1 if occasional tremors

2 if tremors occur often

Teeth grinding

0 if child does not grind teeth

1 if child occasionally grinds teeth

2 if child grinds teeth often/constant

Self Injurious Behavior:

0 if child does not exhibit self injurious behavior

1 if child seldom exhibits self injurious behavior

2 if self injurious behavior is an ongoing problem

Irritability/agitation:

0 if child's irritability/agitation is the same as you would expect in peers

1 if irritability/agitation is slightly more than peer would exhibit

2 if irritability/agitation is an ongoing problem

Abnormal sleep patterns

0 if child has typical sleeping pattern for age group

1 if child has occasional difficulty falling asleep or staying asleep or sleeps excessively during the day

2 if child routinely has difficulty sleeping or sleeps excessively during the day or if child only sleeps with medication