

Abnormal EEG:

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

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

- if child has no eating difficulties
- if child has minor eating difficulties
- if child has major difficulties

Does your child have a feeding tube:

- if your child does not have a feeding tube
- if child has a feeding tube

Gastrointestinal Reflux:

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

Constipation:

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

Bloating:

- if the child has no issues with bloating
- if the child has some issues with bloating
- if bloating is a severe concern

Growth retardation:

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

Scoliosis/kyphosis:

- if child has no scoliosis or kyphosis
- if child has a slight curve
- if child is wearing brace or had surgery to correct curve

Muscle rigidity/spasticity:

- if child has no issues with rigidity/spasticity
- if child has some degree of rigidity/spasticity
- if child has ongoing problems with rigidity/spasticity

Small feet:

- if child's feet size is proportionate with body size
- if child's feet are slightly smaller in proportion to rest of the body
- if child's feet are significantly smaller than expected given body size

Poor circulation:

- if this is not an issue
- if child has occasional problem with circulation
- if child has ongoing issues with circulation

Tremors:

- if no tremor problems
- if occasional tremors
- if tremors occur often

Teeth grinding:

- if child does no grind teeth
- if child occasionally grinds teeth
- if child grinds teeth often/constant

Self-injurious behavior:

- if child does not exhibit self- injurious behavior
- if child seldom exhibits self-injurious behavior
- if self- injurious behavior is an ongoing problem

Irritability/agitation:

- if child's irritability/agitation is the same expected in peers
- if irritability/agitation is slightly more than peer would exhibit
- if irritability/agitation is an ongoing problem

Abnormal sleep patterns:

- if child has typical sleeping pattern for age group
- if child has occasional difficulty falling asleep or staying asleep or sleeps excessively during the day
- if child routinely has difficulty sleeping/sleeps excessively during the day or if child only sleeps with medication

General health:

- if child is never/rarely sick
- if child is often sick
- if child is always sick

Questions?

Contact the NIGMS Human Genetic Cell Repository Genetic Counselor

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