

## NIGMS HUMAN GENETIC CELL REPOSITORY Rett Syndrome Clinical Data Elements Form

Date:		Name:					
Sex:		Age:					
MECP2	mutation:		Testing Lab	:			
Please <u>a</u> print leg		opropriate res	sponse and add an	ny additional observations under comments (please			
				Comments			
Period o	of normal development	<u>:</u>					
	e child had a definite no	•	•				
	u noticed the first signs		the age of 6 mon	ths			
∐ if chi	ild never developed no	rmally					
Deceler	rating head circumferer	ice.					
	ad circumference has n		d				
_	☐ if head circumference deceleration was noted after age 1						
	ad circumference dece		_				
	purposeful hand use:						
	ild has normal hand use		d +h a magali (ag migl				
item	ild has limited hand use	e (e.g. can ree	a themselves, pick				
	ild has no hand use						
	ina mas mo mana asc						
Repetit	ive hand motions:						
☐ if chi	ild has no repetitive ha	nd motions					
☐ if chi	ild has occasional repet	itive hand mo	otions				
☐ if chi	ild has often/constant r	epetitive han	d motions				
Non-ve	rhali						
	ild speaks normally for	their age					
	ild regularly speaks usir	_	ls or short sentend	res			
	ild is non-verbal	.6 06.0					
_							
<u>Ambula</u>							
	e child walks well indep	-					
_	e child can walk with as						
11 tile	e child cannot walk at a	11					
Breathi	ng irregularities (hyper	ventilation, br	eath holding, apn	ea):			
	ild has no breathing pro						
☐ if chi	ild has occasional breat	hing problem	S				
☐ if chi	ild often/constantly has	s breathing pr	oblems				
C-:-							
Seizure:	<u>s:</u> ild has no overt seizure	r					
_	ild has occasional seizu		e controlled with				
	ications	ics of they all	c controlled with				
	ild has seizures often o	they are not	controlled with	·			
	ications						



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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	
<del>-</del>	
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	



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<u>Small feet:</u> ☐ 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	
Tromore	
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	
and any or many stocks are more about the	
General health:	
if child is never/rarely sick	
if child is often sick	
if child is always sick	
II cilila is always sick	

**Questions?** 

Contact the NIGMS Human Genetic Cell Repository Genetic Counselor

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