

Supplementary Table 1. All significant differences for each clinical component of CSS for typical RTT. Average numbers are given in Figure 2. Each of listed pairs is significantly different with $p < 0.05$ as per Tukey-Kramer multiple comparisons test.

<u>Age of Onset</u>		<u>Autonomic Symptoms</u>		<u>Independent sitting</u>	
p.Arg133Cys	p.Arg168X	p.Thr158Met	p.Arg306Cys	p.Arg168X	p.Arg294X
p.Arg133Cys	p.Arg255X	p.Arg168X	p.Arg306Cys	p.Arg168X	p.Arg306Cys
p.Arg133Cys	Deletions	p.Arg255X	p.Arg294X	p.Arg255X	p.Arg294X
p.Arg133Cys	Large Deletions	p.Arg255X	p.Arg306Cys	p.Arg255X	p.Arg306Cys
Deletions	No Mutations	p.Arg294X	Large Deletions	p.Arg270X	p.Arg294X
<u>Ambulation</u>		p.Arg306Cys	Large Deletions	p.Arg270X	p.Arg306Cys
3' Truncations	p.Arg168X	<u>Seizures</u>		p.Arg294X	Deletions
3' Truncations	p.Arg255X	p.Arg255X	p.Arg306Cys	p.Arg294X	Large Deletions
3' Truncations	p.Arg294X	p.Arg294X	p.Arg306Cys	p.Arg294X	No Mutations
3' Truncations	Deletions	p.Arg306Cys	No Mutations	p.Arg306Cys	Deletions
3' Truncations	Large Deletions	<u>Hand Use</u>		p.Arg306Cys	Large Deletions
p.Arg106Trp	p.Arg294X	3' Truncation	p.Arg168X	<u>Onset of stereotypes</u>	
p.Arg133Cys	p.Thr158Met	3' Truncation	p.Arg270X	3' Truncations	p.Arg255X
p.Arg133Cys	p.Arg168X	3' Truncation	Large Deletions	3' Truncations	p.Arg270X
p.Arg133Cys	p.Arg255X	p.Arg106Trp	p.Arg133Cys	p.Arg133Cys	p.Thr158Met
p.Arg133Cys	p.Arg270X	p.Arg133Cys	p.Thr158Met	p.Arg133Cys	p.Arg168X
p.Arg133Cys	Deletions	p.Arg133Cys	p.Arg168X	p.Arg133Cys	p.Arg255X
p.Arg133Cys	Insertions	p.Arg133Cys	p.Arg255X	p.Arg133Cys	p.Arg270X
p.Arg133Cys	Large Deletions	p.Arg133Cys	p.Arg270X	p.Thr158Met	p.Arg294X
p.Arg133Cys	Splice Sites	p.Arg133Cys	Deletions	p.Arg168X	p.Arg294X

p.Arg133Cys	No Mutations	p.Arg133Cys	Insertions	p.Arg255X	p.Arg294X
p.Thr158Met	p.Arg294X	p.Arg133Cys	Large Deletions	p.Arg255X	p.Arg306Cys
p.Arg168X	p.Arg294X	p.Arg133Cys	Splice Sites	p.Arg270X	p.Arg294X
p.Arg168X	p.Arg306Cys	p.Arg168X	p.Arg294X	p.Arg270X	p.Arg306Cys
p.Arg168X	Other Point Mutations	p.Arg168X	p.Arg306Cys	p.Arg270X	No Mutations
p.Arg255X	p.Arg294X	p.Arg168X	Other Point Mutations	p.Arg294X	Large Deletions
p.Arg255X	p.Arg306Cys	p.Arg255X	p.Arg294X	p.Arg294X	Other Point Mutations
p.Arg255X	Other Point Mutations	p.Arg270X	p.Arg294X	<u>Scoliosis</u>	
p.Arg270X	p.Arg294X	p.Arg270X	p.Arg306Cys	3' Truncation	p.Arg306Cys
p.Arg270X	p.Arg306Cys	p.Arg270X	Other Point Mutations	p.Thr158Met	p.Arg306Cys
p.Arg294X	Deletions	p.Arg294X	Large Deletions	p.Arg306Cys	Deletions
p.Arg294X	Insertions	p.Arg294X	Splice Sites	p.Arg306Cys	Large Deletions
p.Arg294X	Large Deletions	p.Arg306Cys	Large Deletions	<u>Somatic growth</u>	
p.Arg294X	Other Point Mutations	Large Deletions	Other Point Mutations	3' Truncation	p.Arg294X
p.Arg294X	Splice Sites	<u>Head Growth</u>			
p.Arg294X	No Mutations	3' Truncation	p.Arg270X		
p.Arg306Cys	Deletions	p.Arg270X	p.Arg294X		
p.Arg306Cys	Insertions				
p.Arg306Cys	Large Deletions				
Deletions	Other Point				

	Mutations				
Large Deletions	Other Point Mutations				

Supplementary Table 2. All significant differences for each clinical component of CSS for atypical RTT. Average numbers are given in Figure 4. Each of listed pairs is significantly different with $p < 0.05$ as per Tukey-Kramer multiple comparisons test.

<u>Autonomic Symptoms</u>		<u>Nonverbal communication</u>	
3' Truncations	p.Arg168X	3' Truncations	p.Arg255X
3' Truncations	p.Arg270X	3' Truncations	No Mutations
<u>Hand Use</u>		<u>Onset of stereotypes</u>	
3' Truncations	p.Arg168X	p.Arg106Trp	p.Arg133Cys
3' Truncations	p.Arg270X	p.Arg106Trp	p.Arg294X
p.Arg133Cys	p.Arg270X	p.Arg133Cys	p.Arg255X
p.Arg270X	p.Arg306Cys	p.Arg133Cys	p.Arg270X
p.Arg270X	Other Point Mutations	p.Arg133Cys	Large Deletions
p.Arg270X	No Mutations	p.Arg133Cys	No Mutations
p.Arg306Cys	Large Deletions	p.Arg255X	p.Arg294X
<u>Language</u>		p.Arg270X	p.Arg294X
3' Truncations	p.Arg168X	p.Arg294X	Large Deletions
p.Arg106Trp	p.Arg168X	p.Arg294X	No Mutations
p.Arg133Cys	p.Arg168X		
p.Arg133Cys	p.Arg255X		
p.Arg133Cys	p.Arg270X		
p.Arg133Cys	Large Deletions		

p.Arg133Cys	No Mutations		
p.Arg168X	p.Arg306Cys		
p.Arg168X	No Mutations		