RettBASE – Mutation Entry Form
Data Entry Form
(modified from the HUGO Mutation Database Initiative form at http://ariel.ucs.unimelb.edu.au:80/~cotton/entry.htm)

Submitter Details
Title: ___________________________ Family name: _________________________________
Given name: ___________________________
Submitter ID Number: ______________________ (if you have a submitter ID number just enter your name and ID number in this section)
Address:
   Dept: _______________________________________________________________________
   Institution: ____________________________________________________________________
   Street Address: __________________________________________________________________
   City: __________________________________________________________________________
   State: ______________ Postcode/Zipcode: _______ Country: ____________________________
Email address: __________________________
Fax number: ____________________________
Role: (eg clinician, lab scientist) ____________________________
Do you want email notification of changes to the database? yes ☐ no ☐

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General Information
Proband Details (confidential):
Please enter either the proband’s details or the proband’s ID (as generated by the Excel spreadsheet available at this site). If you choose to enter the proband’s details please fill in all details and then press generate button. The proband’s ID number will be generated and the proband’s name will be deleted. The proband’s name is never recorded in RettBASE. The proband ID will be used to link with the proposed clinical database.

Family Name: ___________________________ Proband’s Date of Birth: _______________________
Given Name: ___________________________ Middle Name: ____________________________

OR
ID number (Created by the Excel ID number generator) ________________________________
(see Instructions for Submitters)

Mutation associated with disease ☐ polymorphism not causing disease ☐
silent polymorphism ☐ not certain ☐
Reason why sequence change is thought to be mutation or polymorphism: ____________________________
____________________________________________________________________________________
Systematic name of DNA variation: ____________________________
Reference sequence (e.g. GenBank accession ID): ____________________________
Systematic name of protein variation: ____________________________

For details of accepted nomenclatures please review the papers cited under “Mutation Nomenclature” in the section “Useful MECP2 Resources for Researchers”. 
If you are uncertain of the correct nomenclature, please complete a description of the sequence change here:

<table>
<thead>
<tr>
<th>Mutation type:</th>
</tr>
</thead>
<tbody>
<tr>
<td>Silent ☐ Missense ☐ Nonsense ☐ Frameshift insertion or deletion ☐</td>
</tr>
<tr>
<td>In-frame insertion or deletion ☐ Frameshift combined insertion and deletion ☐</td>
</tr>
<tr>
<td>In-frame combined insertion and deletion ☐ Intron variation ☐ Mutation in 3’UTR ☐</td>
</tr>
<tr>
<td>Mutation in 5’UTR ☐</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Mutation domain:</th>
</tr>
</thead>
<tbody>
<tr>
<td>5’UTR ☐ N-terminal region ☐ MBD ☐ Inter-domain region ☐</td>
</tr>
<tr>
<td>TRD ☐ NLS ☐ C-terminal region ☐ 3’UTR ☐ Intrinsic ☐</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Detection method:</th>
</tr>
</thead>
<tbody>
<tr>
<td>Direct sequencing ☐ SSCP ☐ PTT ☐ DGGE ☐ CCM ☐ EMC ☐ DHPLC ☐ Heteroduplex analysis ☐ PCR / diagnostic restriction ☐</td>
</tr>
</tbody>
</table>

If other please specify: ________________________________

Extent of DNA analyzed: (% coding seq., 5'UTR, etc.): ________________________________

Laboratory performing the mutation screening: ________________________________

Published: yes ☐ no ☐ (if published please provide citation)

Author(s) : ________________________________

Title : ________________________________

Journal: ________________________________

Volume: ______ Pages: ___________ PubMed: ___________ Year: ________

Patient ID number in the Journal Article or Laboratory sample ID number (if not published): ________________________________

(As the database will also incorporate all published mutations, there is a risk of double entries. To prevent this from happening, please tell us which patient this data form refers to in the above journal article.)

Chromosomal abnormality: Yes ☐ No ☐ Not known ☐

If yes, please elaborate: ________________________________

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Quality Assurance Checklist for Mutation/Polymorphism

Mutation/polymorphism found on repeat PCR sample (not an artifact)

Yes ☐ No ☐ Not Checked ☐ Not Applicable ☐

Parents, grandparents, siblings, or other family members checked for carrier status

Yes ☐ No ☐ Not Checked ☐ Not Applicable ☐
**RettBase Mutation Entry Form**

Result if tested: ______________________________________________________________________________________

<table>
<thead>
<tr>
<th>MECP2 screened</th>
<th>Yes ☐</th>
<th>No ☐</th>
<th>Not known ☐</th>
<th>Not applicable ☐</th>
</tr>
</thead>
</table>

MECP2 screening results (please include extent of screening, e.g. entire coding region, all 4 exons screened, etc):

Other genes screened: ____________________________________________________________

Other mutation found | Yes ☐ | No ☐ | Not known ☐ | Not applicable ☐ |
|----------------------|-------|------|-------------|------------------|

on same allele (in cis) | Yes ☐ | No ☐ | Not Checked ☐ | Not applicable ☐ |

50 (or other ___________) normal chromosomes tested

<table>
<thead>
<tr>
<th>Yes ☐</th>
<th>No ☐</th>
<th>Not Checked ☐</th>
<th>Not Applicable ☐</th>
</tr>
</thead>
</table>

Result: not found in normal chromosomes

<table>
<thead>
<tr>
<th>X-inactivation studies performed</th>
<th>Yes ☐</th>
<th>No ☐</th>
<th>Not known ☐</th>
<th>Not relevant ☐</th>
</tr>
</thead>
</table>

Result: _____%/_____%

Source of DNA for testing: blood ☐ other ☐ please specify ________________

X-inactivation studies performed in other relatives | Yes ☐ | No ☐ | Not known ☐ | Not relevant ☐ |

| Please give results: __________________________________________________________|

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**Proband Clinical Data:**

Sex: male ☐ female ☐ not known ☐

Phenotype occurrence: sporadic ☐ familial ☐ not known ☐

Phenotype classification: Rett syndrome ☐ Not Rett syndrome ☐ not known ☐

If Rett syndrome, please classify as:

- Classical ☐ atypical ☐ “preserved speech variant” ☐ “forme fruste” ☐
- congenital onset ☐ male variant ☐ not certain ☐

If not Rett syndrome, please classify as:

- Sporadic mental retardation ☐ X-linked mental retardation ☐ unaffected family member ☐
- Angelman syndrome ☐ Mental retardation and autism combined ☐ autism only ☐
- not certain ☐ Non Rett syndrome control ☐

Progressive encephalopathy of neonatal onset ☐ Non-progressive encephalopathy of neonatal onset ☐

If phenotype does not fit under the above categories, please elaborate: ____________________________________________

__________________________________________________________________________
Other Comments:

Thank you for completing this form.

Please send this completed form to:
Professor John Christodoulou
Western Sydney Genetics Program
Children’s Hospital at Westmead
Locked Bag 4001
Westmead NSW 2145  AUSTRALIA
Fax:  [612] 9845 1864
Email: john.christodoulou@health.nsw.gov.au

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