RettBASE: IRSA *MECP2* Gene Variation Database

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: _____ (DD/MM/YYYY)

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 ❒ don't know ❒

Reason why sequence change is thought to be mutation or polymorphism: _____

Systematic name of DNA 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 give as complete a description of the sequence change here:

_____

_____

Systematic name of protein variation : _____

Mutation type:

Silent ❒ Missense ❒ Nonsense ❒ Frameshift insertion or deletion ❒
In-frame insertion or deletion ❒ Frameshift combined insertion and deletion ❒
In-frame combined insertion and deletion ❒ Intronic variation ❒ Mutation in 3'UTR ❒
Mutation in 5'UTR ❒

Mutation domain:

5'UTR ❒ N-terminal region ❒ MBD ❒ Inter-domain region ❒
TRD ❒ NLS ❒ C-terminal region ❒ 3'UTR ❒ Intronic ❒

Detection method:

Direct sequencing ❒ SSCP ❒ PTT ❒ DGGE ❒ CCM ❒
EMC ❒ DHPLC ❒ Heteroduplex analysis ❒ PCR / diagnostic restriction ❒
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 ❒

Result if tested _____

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
Yes ❒ No ❒ Not Checked ❒ Not Applicable ❒

Result: not found in _____ normal chromosomes

X-inactivation studies performed Yes ❒ No ❒ Not known ❒ Not relevant ❒

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:

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