Guide to column names

Citation or Short Citation: A citation of the source of the sequence change data, if applicable.

Nucleotide change: The systematic name for the change being described in the entry in terms of the effect on the nucleotide sequence.

Amino acid change: The systematic name for the change being described in the entry in terms of the effect on the amino acid sequence.

Type of sequence change: Describes the type of sequence variation. Categories include:
- silent
- missense
- nonsense
- frameshift insertion or deletion
- in-frame insertion or deletion
- frameshift combined insertion and deletion
- in-frame combined insertion and deletion
- 3'UTR variation
- 5'UTR variation
- intronic variation

Mutation / polymorphism: Asks whether the sequence variation described is likely to be a mutation (capable of causing a diseased phenotype), a polymorphism (not capable of causing a diseased phenotype) or a silent polymorphism (a variation in the coding sequence of the gene that does not change the protein and therefore can not have any deleterious effect). If a researcher does a search for polymorphisms the results will include polymorphisms and silent polymorphisms.

Domain change location: Describes the area(s) affected by the sequence variation. Categories include:
- 5' untranslated region (5'UTR)
- N-terminal region (amino acids 1 to 77, nucleotides 1-231)
- methyl-binding domain (MBD) (amino acids 78 to 162, nucleotides 232-486)
- region between the MBD and TRD (amino acids 163 to 206, nucleotides 487-618)
- transcription repression domain (TRD) (amino acids 207 to 310, nucleotides 619-930)
- nuclear localisation signal (NLS) (amino acids 255 to 271, nucleotides 763-813)
- C-terminal region (amino acids 311 to 486, nucleotides 931-1458)
- 3'UTR
- intronic sequence changes

Additional sequence variation: Describes whether the patient had any other *MECP2* sequence variations, and if so, whether the variation was on the same allele (*cis*) or the other allele (*trans*).

Phenotype classification: Description of the phenotype of the person. Possible variants recognised in Rett syndrome are:
- classical
- atypical
- forme fruste
- preserved speech
- congenital
- male variant
- specific type of Rett syndrome not known.

Possible phenotypes for individuals who do not have Rett syndrome are:
- 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.

Evidence of pathogenicity: If the sequence variation has been determined to be a mutation or a polymorphism, the reasoning is briefly described here.

Sporadic or familial: Describes if only one person in the family has an abnormal phenotype.

Sex: The gender of the person with the sequence change.

X-inactivation ratio: Describes the X-inactivation ratio of the person if it has been measured.

X-inactivation ratio of relatives: Describes the X-inactivation ratios in relatives of the person if they have been measured.

Carrier status of family: Describes if family members were screened for the sequence variation found in the proband, and if so, what the results were.

Detection method: The sequence analysis method used to screen for sequence variations.

Extent of coding region screened: Describes which parts of the gene were screened for the sequence change.

Source of DNA: The tissue from which the DNA sample was extracted.

Entry ID: A record number for the entry in the database.

Patient ID (from cited work): This is the ID number allocated to this patient in the published paper.