

# Resource Summary Report

Generated by [SPARC SAWG](#) on Sep 12, 2026

## RettBASE: IRSF MECP2 Variation Database

RRID:SCR\_001066

Type: Tool

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### Proper Citation

RettBASE: IRSF MECP2 Variation Database (RRID:SCR\_001066)

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### Resource Information

**URL:** <http://mecp2.chw.edu.au/>

**Proper Citation:** RettBASE: IRSF MECP2 Variation Database (RRID:SCR\_001066)

**Description:** Database constructed by merging mutation and polymorphism data from the published literature pertaining to Rett syndrome and related clinical disorders, and by incorporating unpublished mutation and polymorphism data that have been submitted directly. RettBase is updated on a very frequent basis manually by curators to ensure the validity of the data submitted., THIS RESOURCE IS NO LONGER IN SERVICE.  
Documented on September 16,2025.

**Synonyms:** RettBase

**Resource Type:** data or information resource, database

**Defining Citation:** [PMID:28544139](#)

**Keywords:** genetics; metadata, polymorphism

**Related Condition:** Rett Syndrome

**Availability:** THIS RESOURCE IS NO LONGER IN SERVICE

**Resource Name:** RettBASE: IRSF MECP2 Variation Database

**Resource ID:** SCR\_001066

**Alternate IDs:** nif-0000-00156

**Record Creation Time:** 20220129T080205+0000

**Record Last Update:** 20260912T200121+0000

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### Ratings and Alerts

No rating or validation information has been found for RettBASE: IRSF MECP2 Variation Database.

No alerts have been found for RettBASE: IRSF MECP2 Variation Database.

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## Data and Source Information

**Source:** [SciCrunch Registry](#)

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## Usage and Citation Metrics

We found 14 mentions in open access literature.

**Listed below are recent publications.** The full list is available at [SPARC SAWG](#) .

Shah RR, et al. (2017) MeCP2 mutations: progress towards understanding and treating Rett syndrome. *Genome medicine*, 9(1), 17.

Bueno C, et al. (2016) Rett Syndrome Mutant Neural Cells Lacks MeCP2 Immunoreactive Bands. *PloS one*, 11(4), e0153262.

Cobolli Gigli C, et al. (2016) MeCP2 Related Studies Benefit from the Use of CD1 as Genetic Background. *PloS one*, 11(4), e0153473.

Stefanelli G, et al. (2016) Brain phosphorylation of MeCP2 at serine 164 is developmentally regulated and globally alters its chromatin association. *Scientific reports*, 6, 28295.

Bianciardi L, et al. (2016) MECP2 missense mutations outside the canonical MBD and TRD domains in males with intellectual disability. *Journal of human genetics*, 61(2), 95.

Gold WA, et al. (2015) The Utility of Next-Generation Sequencing in Gene Discovery for Mutation-Negative Patients with Rett Syndrome. *Frontiers in cellular neuroscience*, 9, 266.

Viana MC, et al. (2015) MECP2, a gene associated with Rett syndrome in humans, shows conserved coding regions, independent Alu insertions, and a novel transcript across primate evolution. *BMC genetics*, 16, 77.

Kucukkal TG, et al. (2014) Computational and experimental approaches to reveal the effects of single nucleotide polymorphisms with respect to disease diagnostics. *International journal of molecular sciences*, 15(6), 9670.

Bellini E, et al. (2014) MeCP2 post-translational modifications: a mechanism to control its involvement in synaptic plasticity and homeostasis? *Frontiers in cellular neuroscience*, 8, 236.

Baker SA, et al. (2013) An AT-hook domain in MeCP2 determines the clinical course of Rett syndrome and related disorders. *Cell*, 152(5), 984.

Ravn K, et al. (2011) Two new Rett syndrome families and review of the literature: expanding the knowledge of MECP2 frameshift mutations. Orphanet journal of rare diseases, 6, 58.

Fendri-Kriaa N, et al. (2011) A case of a Tunisian Rett patient with a novel double-mutation of the MECP2 gene. Biochemical and biophysical research communications, 409(2), 270.

Ho KL, et al. (2008) MeCP2 binding to DNA depends upon hydration at methyl-CpG. Molecular cell, 29(4), 525.

Young JI, et al. (2004) X-chromosome inactivation patterns are unbalanced and affect the phenotypic outcome in a mouse model of rett syndrome. American journal of human genetics, 74(3), 511.