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RettBASE: The IRSA MECP2 variation database-a new mutation database in evolution

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#	Paper	IF	Citations
136	Gross rearrangements in the MECP2 gene in three patients with Rett syndrome: implications for routine diagnosis of Rett syndrome. *Human Mutation*, **2003**, 22, 116-20	4.7	48
135	Mutations and polymorphisms in the human methyl CpG-binding protein MECP2. *Human Mutation*, **2003**, 22, 107-15	4.7	68
134	InterRett and RettBASE: International Rett Syndrome Association databases for Rett syndrome. **2003**, 18, 709-13		30
133	MECP2 and beyond: phenotype-genotype correlations in Rett syndrome. **2003**, 18, 669-74		27
132	Refining the phenotype of common mutations in Rett syndrome. **2004**, 41, 25-30		65
131	Phenotypic manifestations of MECP2 mutations in classical and atypical Rett syndrome. **2004**, 126A, 129-40		101
130	MECP2 is progressively expressed in post-migratory neurons and is involved in neuronal maturation rather than cell fate decisions. **2004**, 27, 306-21		341
129	X-chromosome inactivation patterns are unbalanced and affect the phenotypic outcome in a mouse model of rett syndrome. **2004**, 74, 511-20		109
128	Methyl-CpG binding protein 2 gene (MECP2) variations in Japanese patients with Rett syndrome: pathological mutations and polymorphisms. **2005**, 27, 211-7		42
127	Genotype and early development in Rett syndrome: the value of international data. **2005**, 27 Suppl 1, S59-S68		30
126	MECP2 mutation analysis in patients with mental retardation. **2005**, 132A, 121-4		27
125	Does genotype predict phenotype in Rett syndrome?. **2005**, 20, 768-78		29
124	Effectiveness of posthumous molecular diagnosis from a kept baby tooth. **2005**, 366, 1584		3
123	Rett syndrome: clinical review and genetic update. **2005**, 42, 1-7		121
122	Rett syndrome: new clinical and molecular insights. **2006**, 14, 896-903		94
121	The molecular pathology of Rett syndrome: synopsis and update. **2006**, 8, 485-94		16
120	The association between behavior and genotype in Rett syndrome using the Australian Rett Syndrome Database. **2006**, 141B, 177-83		45

119	Recurrent infections, hypotonia, and mental retardation caused by duplication of MECP2 and adjacent region in Xq28. **2006**, 118, e1687-95		158
118	The story of Rett syndrome: from clinic to neurobiology. **2007**, 56, 422-37		936
117	Italian Rett database and biobank. *Human Mutation*, **2007**, 28, 329-35	4.7	23
116	Interpreting missense variants: comparing computational methods in human disease genes CDKN2A, MLH1, MSH2, MECP2, and tyrosinase (TYR). *Human Mutation*, **2007**, 28, 683-93	4.7	111
115	Delineation of large deletions of the MECP2 gene in Rett syndrome patients, including a familial case with a male proband. **2007**, 15, 1218-29		40
114	Low significance of MECP2 mutations as a cause of mental retardation in Brazilian males. **2007**, 29, 293-7		13
113	Connections between epigenetic gene silencing and human disease. **2007**, 618, 163-74		88
112	The diagnosis of autism in a female: could it be Rett syndrome?. **2008**, 167, 661-9		48
111	Investigating genotype-phenotype relationships in Rett syndrome using an international data set. **2008**, 70, 868-75		180
110	Selective cerebral volume reduction in Rett syndrome: a multiple-approach MR imaging study. **2008** , 29, 436-41		72
109	Deciphering Rett syndrome with mouse genetics, epigenomics, and human neurons. **2009**, 89, 147-60		8
108	Mouse models of MeCP2 disorders share gene expression changes in the cerebellum and hypothalamus. *Human Molecular Genetics*, **2009**, 18, 2431-42	5.6	185
107	MeCP2 post-translational regulation through PEST domains: two novel hypotheses: potential relevance and implications for Rett syndrome. **2009**, 31, 561-9		19
106	A novel hypomorphic MECP2 point mutation is associated with a neuropsychiatric phenotype. **2009**, 124, 615-23		22
105	InterRett, a model for international data collection in a rare genetic disorder. **2009**, 3, 639-639		39
104	Rapid detection of deletions in hotspot C-terminal segment region of MECP2 by routine PCR method: report of two classical Rett syndrome patients of Indian origin. **2009**, 13, 277-80		4
103	Hip displacement and scoliosis in Rett syndrome - screening is required. **2010**, 52, 93-8		10
102	De novo duplication of MECP2 in a girl with mental retardation and no obvious dysmorphic features. *Clinical Genetics*, **2010**, 78, 175-80	4	21

101	F2-neuroprostanes mediate neurological severity in Rett syndrome. **2011**, 412, 1399-406		63
100	Ex vivo treatment with a novel synthetic aminoglycoside NB54 in primary fibroblasts from Rett syndrome patients suppresses MECP2 nonsense mutations. *PLoS ONE*, **2011**, 6, e20733	3.7	37
99	A MECP2 missense mutation within the MBD domain in a Brazilian male with autistic disorder. **2011**, 33, 807-9		9
98	Neuronal maturation defect in induced pluripotent stem cells from patients with Rett syndrome. **2011**, 108, 14169-74		167
97	F2-dihomo-isoprostanes as potential early biomarkers of lipid oxidative damage in Rett syndrome. **2011**, 52, 2287-2297		72
96	Analysis of Hungarian patients with Rett syndrome phenotype for MECP2, CDKL5 and FOXG1 gene mutations. **2011**, 56, 183-7		13
95	Oxidative stress in Rett syndrome: natural history, genotype, and variants. **2011**, 16, 145-53		59
94	A novel MECP2 change in an indian boy with variant rett phenotype and congenital blindness: implications for genetic counseling and prenatal diagnosis. **2011**, 26, 209-13		6
93	The phenotype associated with a large deletion on MECP2. **2012**, 20, 921-7		25
92	Rett networked database: an integrated clinical and genetic network of Rett syndrome databases. *Human Mutation*, **2012**, 33, 1031-6	4.7	14
91	Novel non-identical MECP2 mutations in Rett syndrome family: a rare presentation. **2012**, 34, 28-31		4
90	The role of oxidative stress in Rett syndrome: an overview. **2012**, 1259, 121-35		72
89	Barriers to diagnosis of a rare neurological disorder in China--lived experiences of Rett syndrome families. **2012**, 158A, 1-9		13
88	A novel transcript of cyclin-dependent kinase-like 5 (CDKL5) has an alternative C-terminus and is the predominant transcript in brain. **2012**, 131, 187-200		38
87	Role of conserved cis-regulatory elements in the post-transcriptional regulation of the human MECP2 gene involved in autism. **2013**, 7, 19		10
86	Isoprostanes and neuroprostanes: total synthesis, biological activity and biomarkers of oxidative stress in humans. **2013**, 107, 95-102		61
85	Rett syndrome mutations abolish the interaction of MeCP2 with the NCoR/SMRT co-repressor. **2013**, 16, 898-902		252
84	MeCP2 R168X male and female mutant mice exhibit Rett-like behavioral deficits. **2013**, 12, 732-40		37

83	Redox imbalance and morphological changes in skin fibroblasts in typical Rett syndrome. **2014**, 2014, 195935		36
82	Rett Syndrome: Coming to Terms with Treatment. **2014**, 2014, 1-20		7
81	Perspectives on hand function in girls and women with Rett syndrome. **2014**, 17, 210-7		14
80	Rett syndrome and MeCP2. **2014**, 16, 231-64		84
79	Experience of gastrostomy using a quality care framework: the example of rett syndrome. **2014**, 93, e328		19
78	Mutation is a Low-Incidence Genetic Cause in Atypical Rett Syndrome. **2015**, 2, 2329048X14568151		3
77	The Utility of Next-Generation Sequencing in Gene Discovery for Mutation-Negative Patients with Rett Syndrome. **2015**, 9, 266		10
76	Structural, Dynamical, and Energetical Consequences of Rett Syndrome Mutation R133C in MeCP2. **2015**, 2015, 746157		9
75	Rett syndrome: disruption of epigenetic control of postnatal neurological functions. *Human Molecular Genetics*, **2015**, 24, R10-6	5.6	48
74	A Novel Mutation p.A59P in N-Terminal Domain of Methyl-CpG-Binding Protein 2 Confers Phenotypic Variability in 3 Cases of Tunisian Rett Patients: Clinical Evaluations and In Silico Investigations. **2015**, 30, 1715-21		4
73	Neuronal nucleus and cytoplasm volume deficit in children with autism and volume increase in adolescents and adults. **2015**, 3, 2		22
72	Exploring the possible link between MeCP2 and oxidative stress in Rett syndrome. **2015**, 88, 81-90		41
71	Rett syndrome: a complex disorder with simple roots. **2015**, 16, 261-75		216
70	Longitudinal bone mineral content and density in Rett syndrome and their contributing factors. **2015**, 74, 191-8		11
69	Impact of Rett Syndrome Mutations on MeCP2 MBD Stability. **2015**, 54, 6357-68		18
68	Modifiers and Readers of DNA Modifications and Their Impact on Genome Structure, Expression, and Stability in Disease. **2016**, 7, 115		33
67	Characterisation of CDKL5 Transcript Isoforms in Human and Mouse. *PLoS ONE*, **2016**, 11, e0157758	3.7	35
66	The Clinical Predictors That Facilitate a Clinician's Decision to Order Genetic Testing for Rett Syndrome. **2016**, 63, 66-70		6

65	From Function to Phenotype: Impaired DNA Binding and Clustering Correlates with Clinical Severity in Males with Missense Mutations in MECP2. *Scientific Reports*, **2016**, 6, 38590	4.9	12
64	Analysis of 589,306 genomes identifies individuals resilient to severe Mendelian childhood diseases. *Nature Biotechnology*, **2016**, 34, 531-8	44.5	196
63	OxInflammation in Rett syndrome. **2016**, 81, 246-253		36
62	Whole exome sequencing of Rett syndrome-like patients reveals the mutational diversity of the clinical phenotype. **2016**, 135, 1343-1354		44
61	A rare MeCP2_e1 mutation first described in a male patient with severe neonatal encephalopathy. **2016**, 170, 1881-3		4
60	Extensive Variation in the Mutation Rate Between and Within Human Genes Associated with Mendelian Disease. *Human Mutation*, **2016**, 37, 488-94	4.7	11
59	The MECP2 variant c.925C>T (p.Arg309Trp) causes intellectual disability in both males and females without classic features of Rett syndrome. *Clinical Genetics*, **2016**, 89, 733-8	4	12
58	Binding Analysis of Methyl-CpG Binding Domain of MeCP2 and Rett Syndrome Mutations. **2016**, 11, 2706-2715		27
57	MECP2 mutations in Czech patients with Rett syndrome and Rett-like phenotypes: novel mutations, genotype-phenotype correlations and validation of high-resolution melting analysis for mutation scanning. **2016**, 61, 617-25		5
56	Functional outcomes in Rett syndrome. **2016**, 38, 76-81		6
55	Inherited human IRAK-1 deficiency selectively impairs TLR signaling in fibroblasts. **2017**, 114, E514-E523		31
54	Clinical, Molecular, and Computational Analysis in Patients With a Novel Double Mutation and a New Synonymous Variant in MeCP2: Report of the First Missense Mutation Within the AT-hook1 Cluster in Rett Syndrome. **2017**, 32, 694-703		5
53	Neuroepigenomics in Aging and Disease. **2017**,		3
52	Stem Cell Technology for (Epi)genetic Brain Disorders. **2017**, 978, 443-475		4
51	RettBASE: Rett syndrome database update. *Human Mutation*, **2017**, 38, 922-931	4.7	65
50	Structure of the MeCP2-TBLR1 complex reveals a molecular basis for Rett syndrome and related disorders. **2017**, 114, E3243-E3250		42
49	Clinical and biological progress over 50 years in Rett syndrome. **2017**, 13, 37-51		120
48	The utility of Next Generation Sequencing for molecular diagnostics in Rett syndrome. *Scientific Reports*, **2017**, 7, 12288	4.9	16

47	MeCP2 and CTCF: enhancing the cross-talk of silencers. **2017**, 95, 593-608		7
46	Development of the Tailored Rett Intervention and Assessment Longitudinal (TRIAL) database and the Rett Evaluation of Symptoms and Treatments (REST) Questionnaire. **2017**, 7, e015342		8
45	variants: Improving our understanding of a rare neurologic disorder. **2017**, 3, e200		27
44	Lactonase Activity and Lipoprotein-Phospholipase A as Possible Novel Serum Biomarkers for the Differential Diagnosis of Autism Spectrum Disorders and Rett Syndrome: Results from a Pilot Study. **2017**, 2017, 5694058		16
43	MeCP2 AT-Hook1 mutations in patients with intellectual disability and/or schizophrenia disrupt DNA binding and chromatin compaction in vitro. *Human Mutation*, **2018**, 39, 717-728	4.7	12
42	MECP2 variation in Rett syndrome-An overview of current coverage of genetic and phenotype data within existing databases. *Human Mutation*, **2018**, 39, 914-924	4.7	12
41	Rett Syndrome: A Genetic Update and Clinical Review Focusing on Comorbidities. **2018**, 9, 167-176		44
40	Genetics. **2018**, 125-189		
39	Novel Mutation in the MECP2 Gene Identified in a Group of Rett Syndrome Patients from Ukraine. **2018**, 52, 294-298		2
38	Affinity for DNA Contributes to NLS Independent Nuclear Localization of MeCP2. **2018**, 24, 2213-2220		14
37	Rett Syndrome and Stem Cell Research. **2018**, 27-41		
36	A mutation-led search for novel functional domains in MeCP2. *Human Molecular Genetics*, **2018**, 27, 2531-2545	5.6	14
35	Molecular Testing of MECP2 Gene in Rett Syndrome Phenotypes in Indian Girls. *Indian Pediatrics*, **2018**, 55, 474-477	1.2	3
34	Spectrum of MECP2 mutations in Vietnamese patients with RETT syndrome. *BMC Medical Genetics*, **2018**, 19, 137	2.1	3
33	Diagnostic yield of targeted massively parallel sequencing in children with epileptic encephalopathy. *Seizure: the Journal of the British Epilepsy Association*, **2018**, 59, 132-140	3.2	39
32	Genetic Landscape of Rett Syndrome Spectrum: Improvements and Challenges. *International Journal of Molecular Sciences*, **2019**, 20,	6.3	15
31	Genomic insights into MeCP2 function: A role for the maintenance of chromatin architecture. *Current Opinion in Neurobiology*, **2019**, 59, 174-179	7.6	15
30	P152R Mutation Within MeCP2 Can Cause Loss of DNA-Binding Selectivity. *Interdisciplinary Sciences, Computational Life Sciences*, **2019**, 11, 10-20	3.5	1

29	Whole exome sequencing reveals a de novo missense variant in in a Rett syndrome-like patient. *Clinical Case Reports (discontinued)*, **2019**, 7, 2476-2482	0.7	4
28	An early seizure variant type of a male Rett syndrome patient with a MECP2 p.Arg133His missense mutation. *Molecular Genetics & Genomic Medicine*, **2019**, 7, e532	2.3	2
27	Mosaic MECP2 variants in males with classical Rett syndrome features, including stereotypical hand movements. *Clinical Genetics*, **2019**, 95, 403-408	4	5
26	Molecular Dynamic Simulations Suggest that P152R Mutation Within MeCP2 Can Lead to Higher DNA Binding Affinity and Loss of Selective Binding to Methylated DNA. *Advances in Intelligent Systems and Computing*, **2019**, 27-34	0.4	
25	The complexity of Rett syndrome models: Primary fibroblasts as a disease-in-a-dish reliable approach. *Drug Discovery Today: Disease Models*, **2020**, 31, 11-19	1.3	2
24	MeCP2 links heterochromatin condensates and neurodevelopmental disease. *Nature*, **2020**, 586, 440-444	50.4	45
23	Complete Profiling of Methyl-CpG-Binding Domains for Combinations of Cytosine Modifications at CpG Dinucleotides Reveals Differential Read-out in Normal and Rett-Associated States. *Scientific Reports*, **2020**, 10, 4053	4.9	6
22	X-linked and mitochondrial disorders. **2021**, 137-149		
21	Induced pluripotent stem cells for modeling of Rett Syndrome. **2021**, 171-216		
20	Circulating 4-F-Neuroprostane and 10-F-Neuroprostane Are Related to Gene Mutation and Natural History in Rett Syndrome. *International Journal of Molecular Sciences*, **2021**, 22,	6.3	0
19	Altered Bone Status in Rett Syndrome. *Life*, **2021**, 11,	3	1
18	Hip Displacement in MECP2 Disorders: Prevalence and Risk Factors. *Journal of Pediatric Orthopaedics*, **2021**, 41, e800-e803	2.4	
17	MECP2-related conditions in males: A systematic literature review and 8 additional cases. *European Journal of Paediatric Neurology*, **2021**, 34, 7-13	3.8	2
16	A catalogue of 863 Rett-syndrome-causing MECP2 mutations and lessons learned from data integration. *Scientific Data*, **2021**, 8, 10	8.2	4
15	Current developments in the genetics of Rett and Rett-like syndrome. *Current Opinion in Psychiatry*, **2018**, 31, 103-108	4.9	25
14	MECP2 disorders: from the clinic to mice and back. *Journal of Clinical Investigation*, **2015**, 125, 2914-23	15.9	143
13	Apoptotic Activity of MeCP2 Is Enhanced by C-Terminal Truncating Mutations. *PLoS ONE*, **2016**, 11, e0159632	3.7	2
12	Rett-causing mutations reveal two domains critical for MeCP2 function and for toxicity in MECP2 duplication syndrome mice. *ELife*, **2014**, 3,	8.9	57

11	Sindrome di Rett. *Medico E Bambino*, **2021**, 40, 519-521	0.4	
10	Engineered pegRNAs improve prime editing efficiency. *Nature Biotechnology*, **2021**,	44.5	37
9	Substantial acetylcholine reduction in multiple brain regions of Mecp2-deficient female rats and associated behavioral abnormalities. *PLoS ONE*, **2021**, 16, e0258830	3.7	1
8	Centro de Estudos do Genoma Humano:. *Revista Neurociencias*, **2012**, 20, 194-199	0	1
7	Neuroprostanes and Neurological Severity in Rett Syndrome. **2014**, 2633-2645		
6	A mutation-led search for novel functional domains in MeCP2.		
5	Rett Syndrome around the world. **2019**, 111-121		
4	Paving Therapeutic Avenues for FOXG1 Syndrome: Untangling Genotypes and Phenotypes from a Molecular Perspective.. *International Journal of Molecular Sciences*, **2022**, 23,	6.3	0
3	Breathing Abnormalities During Sleep and Wakefulness in Rett Syndrome: Clinical Relevance and Paradoxical Relationship With Circulating Pro-oxidant Markers.. *Frontiers in Neurology*, **2022**, 13, 833239	4.1	1
2	Sirtuins as potential therapeutic targets for mitigating OxInflammation in typical Rett syndrome: plausible mechanisms and evidence. **2022**, 2022, R26-R39		
1	Library Screening Reveals Sequence Motifs That Enable ADAR2 Editing at Recalcitrant Sites.		0