Effects of MECP2 mutation type, location and X-inactivation in modulating Rett syndrome phenotype

DOI: 10.1002/ajmg.a.10053
American Journal of Medical Genetics Part A, 2003, 118A, 103-14.

#	Paper	IF	Citations
86	Multiplex ligation-dependent probe amplification (MLPA) detects large deletions in the MECP2 gene of Swedish Rett syndrome patients. *Genetic Testing and Molecular Biomarkers*, **2003**, 7, 329-32		63
85	Hand preference, extent of laterality, and functional hand use in Rett syndrome. *Journal of Child Neurology*, **2003**, 18, 481-7	2.5	13
84	InterRett and RettBASE: International Rett Syndrome Association databases for Rett syndrome. *Journal of Child Neurology*, **2003**, 18, 709-13	2.5	30
83	MECP2 and beyond: phenotype-genotype correlations in Rett syndrome. *Journal of Child Neurology*, **2003**, 18, 669-74	2.5	27
82	MeCP2 expression in human cerebral cortex and lymphoid cells: immunochemical characterization of a novel higher-molecular-weight form. *Journal of Child Neurology*, **2003**, 18, 675-82	2.5	10
81	Influence of MECP2 gene mutation and X-chromosome inactivation on the Rett syndrome phenotype. *Journal of Child Neurology*, **2004**, 19, 503-8	2.5	17
80	Rett syndrome: a prototypical neurodevelopmental disorder. *Neuroscientist*, **2004**, 10, 118-28	7.6	114
79	Mutation analysis of MECP2 and determination of the X-inactivation pattern in Hungarian Rett syndrome patients. **2004**, 131, 106		2
78	The transcriptional repressor Mecp2 regulates terminal neuronal differentiation. *Molecular and Cellular Neurosciences*, **2004**, 27, 44-58	4.8	66
77	X-chromosome inactivation patterns are unbalanced and affect the phenotypic outcome in a mouse model of rett syndrome. *American Journal of Human Genetics*, **2004**, 74, 511-20	11	109
76	Mutations of CDKL5 cause a severe neurodevelopmental disorder with infantile spasms and mental retardation. *American Journal of Human Genetics*, **2004**, 75, 1079-93	11	352
75	Clinical stringency greatly improves mutation detection in Rett syndrome. *Canadian Journal of Neurological Sciences*, **2005**, 32, 321-6	1	5
74	Dimensional phenotypic analysis and functional categorisation of mutations reveal novel genotype-phenotype associations in Rett syndrome. *European Journal of Human Genetics*, **2005**, 13, 1121-30	5.3	45
73	Methyl-CpG binding protein 2 gene (MECP2) variations in Japanese patients with Rett syndrome: pathological mutations and polymorphisms. *Brain and Development*, **2005**, 27, 211-7	2.2	42
72	X chromosome inactivation patterns in brain in Rett syndrome: implications for the disease phenotype. *Brain and Development*, **2005**, 27, 266-70	2.2	30
71	Histone modifications in Rett syndrome lymphocytes: a preliminary evaluation. *Brain and Development*, **2005**, 27, 331-9	2.2	22
70	MeCP2 expression and function during brain development: implications for Rett syndrome's pathogenesis and clinical evolution. *Brain and Development*, **2005**, 27 Suppl 1, S77-S87	2.2	78

69	Genotype and early development in Rett syndrome: the value of international data. *Brain and Development*, **2005**, 27 Suppl 1, S59-S68	2.2	30
68	Macrocephalic mental retardation associated with a novel C-terminal MECP2 frameshift deletion. *European Journal of Pediatrics*, **2005**, 164, 154-7	4.1	6
67	Health service use in Rett syndrome. *Journal of Child Neurology*, **2005**, 20, 42-50	2.5	9
66	Does genotype predict phenotype in Rett syndrome?. *Journal of Child Neurology*, **2005**, 20, 768-78	2.5	29
65	Rett syndrome: clinical review and genetic update. *Journal of Medical Genetics*, **2005**, 42, 1-7	5.8	121
64	Rett syndrome in Australia: a review of the epidemiology. *Journal of Pediatrics*, **2006**, 148, 347-52	3.6	241
63	Predictors of seizure onset in Rett syndrome. *Journal of Pediatrics*, **2006**, 149, 542-7	3.6	79
62	Temporal shift in methyl-CpG binding protein 2 expression in a mouse model of Rett syndrome. *Neuroscience*, **2006**, 139, 1449-60	3.9	29
61	Increased skewing of X chromosome inactivation in Rett syndrome patients and their mothers. *European Journal of Human Genetics*, **2006**, 14, 1189-94	5.3	39
60	The association between behavior and genotype in Rett syndrome using the Australian Rett Syndrome Database. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2006**, 141B, 177-83	3.5	45
59	Mecp2 deficiency is associated with learning and cognitive deficits and altered gene activity in the hippocampal region of mice. *Brain*, **2006**, 129, 887-98	11.2	172
58	Mechanisms of disease: neurogenetics of MeCP2 deficiency. *Nature Clinical Practice Neurology*, **2006**, 2, 212-21		45
57	Predictors of scoliosis in Rett syndrome. *Journal of Child Neurology*, **2006**, 21, 809-13	2.5	51
56	Very mild cases of Rett syndrome with skewed X inactivation. *Journal of Medical Genetics*, **2006**, 43, 814-6	5.8	38
55	Autism-lessons from the X chromosome. *Social Cognitive and Affective Neuroscience*, **2006**, 1, 183-93	4	60
54	Correlation between clinical severity in patients with Rett syndrome with a p.R168X or p.T158M MECP2 mutation, and the direction and degree of skewing of X-chromosome inactivation. *Journal of Medical Genetics*, **2007**, 44, 148-52	5.8	72
53	Feeding experiences and growth status in a Rett syndrome population. *Journal of Pediatric Gastroenterology and Nutrition*, **2007**, 45, 582-90	2.8	35
52	Arts syndrome is caused by loss-of-function mutations in PRPS1. *American Journal of Human Genetics*, **2007**, 81, 507-18	11	65

51	Imprinting status of paternally imprinted DLX5 gene in Japanese patients with Rett syndrome. *Brain and Development*, **2007**, 29, 491-5	2.2	6
50	Sleep problems in Rett syndrome. *Brain and Development*, **2007**, 29, 609-16	2.2	91
49	Seizures in Rett syndrome: an overview from a one-year calendar study. *European Journal of Paediatric Neurology*, **2007**, 11, 310-7	3.8	68
48	Development of a video-based evaluation tool in Rett syndrome. *Journal of Autism and Developmental Disorders*, **2007**, 37, 1636-46	4.6	31
47	The diagnosis of autism in a female: could it be Rett syndrome?. *European Journal of Pediatrics*, **2008**, 167, 661-9	4.1	48
46	Deletion of Mecp2 in Sim1-expressing neurons reveals a critical role for MeCP2 in feeding behavior, aggression, and the response to stress. *Neuron*, **2008**, 59, 947-58	13.9	202
45	Multiple de novo mutations in the MECP2 gene. *Genetic Testing and Molecular Biomarkers*, **2008**, 12, 373-5		11
44	X chromosome inactivation in Rett Syndrome and its correlations with MECP2 mutations and phenotype. *Journal of Child Neurology*, **2008**, 23, 22-5	2.5	31
43	Excavating relics of DNA methylation changes during the development of neoplasia. *Seminars in Cancer Biology*, **2009**, 19, 198-208	12.7	18
42	A novel hypomorphic MECP2 point mutation is associated with a neuropsychiatric phenotype. *Human Genetics*, **2009**, 124, 615-23	6.3	22
41	Neurochemical changes in a mouse model of Rett syndrome: changes over time and in response to perinatal choline nutritional supplementation. *Journal of Neurochemistry*, **2009**, 108, 361-71	6	39
40	Psychiatric features in children with genetic syndromes: toward functional phenotypes. *Child and Adolescent Psychiatric Clinics of North America*, **2010**, 19, 229-61, viii	3.3	21
39	Mutational analysis of the MECP2 gene in Tunisian patients with Rett syndrome: a novel double mutation. *Journal of Child Neurology*, **2010**, 25, 1042-6	2.5	8
38	Late-onset mental deterioration and fluctuating dystonia in a female patient with a truncating MECP2 mutation. *Journal of the Neurological Sciences*, **2011**, 308, 168-72	3.2	3
37	Psychiatric features in children with genetic syndromes: toward functional phenotypes. *Pediatric Clinics of North America*, **2011**, 58, 833-64, x	3.6	12
36	F□neuroprostanes mediate neurological severity in Rett syndrome. *Clinica Chimica Acta*, **2011**, 412, 1399-406	6.0	63
35	A case of a Tunisian Rett patient with a novel double-mutation of the MECP2 gene. *Biochemical and Biophysical Research Communications*, **2011**, 409, 270-4	3.4	7
34	Rett syndrome with and without detected MECP2 mutations: an attempt to redefine phenotypes. *Brain and Development*, **2011**, 33, 69-76	2.2	18

33	Long non-coding RNA modifies chromatin: epigenetic silencing by long non-coding RNAs. *BioEssays*, **2011**, 33, 830-9	4.1	148
32	Analysis of Hungarian patients with Rett syndrome phenotype for MECP2, CDKL5 and FOXG1 gene mutations. *Journal of Human Genetics*, **2011**, 56, 183-7	4.3	13
31	Molecular diagnostic dilemmas in Rett syndrome. *Brain and Development*, **2012**, 34, 750-5	2.2	3
30	Novel double deletions in the MECP2 gene in Tunisian Rett patient. *Gene*, **2012**, 502, 163-7	3.8	4
29	Recurrent mutations in the CDKL5 gene: genotype-phenotype relationships. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 1612-9	2.5	35
28	The role of oxidative stress in Rett syndrome: an overview. *Annals of the New York Academy of Sciences*, **2012**, 1259, 121-35	6.5	72
27	Detection of rarely identified multiple mutations in MECP2 gene do not contribute to enhanced severity in Rett syndrome. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 1638-46	2.5	4
26	Regional MeCP2 expression levels in the female MeCP2-deficient mouse brain correlate with specific behavioral impairments. *Experimental Neurology*, **2013**, 239, 49-59	5.7	23
25	Recent insights into genotype-phenotype relationships in patients with Rett syndrome using a fine grain scale. *Research in Developmental Disabilities*, **2014**, 35, 2976-86	2.7	29
24	Rett syndrome and MeCP2. *NeuroMolecular Medicine*, **2014**, 16, 231-64	4.6	84
23	Methyl-CpG-binding protein 2 (MECP2) mutation type is associated with disease severity in Rett syndrome. *Journal of Medical Genetics*, **2014**, 51, 152-8	5.8	181
22	Understanding the Complex Circuitry of lncRNAs at the X-inactivation Center and Its Implications in Disease Conditions. *Current Topics in Microbiology and Immunology*, **2016**, 394, 1-27	3.3	7
21	Genetic alterations of DNA methylation machinery in human diseases. *Epigenomics*, **2015**, 7, 247-65	4.4	156
20	Characterization of Rett Syndrome-like phenotypes in Mecp2-knockout rats. *Journal of Neurodevelopmental Disorders*, **2016**, 8, 23	4.6	38
19	Effects of chronic exposure to low dose THIP on brainstem neuronal excitability in mouse models of Rett syndrome: Evidence from symptomatic females. *Neuropharmacology*, **2017**, 116, 288-299	5.5	2
18	De novo loss-of-function variants in STAG2 are associated with developmental delay, microcephaly, and congenital anomalies. *American Journal of Medical Genetics, Part A*, **2017**, 173, 1319-1327	2.5	24
17	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. *Journal of Child Neurology*, **2017**, 32, 694-703	2.5	5
16	RettBASE: Rett syndrome database update. *Human Mutation*, **2017**, 38, 922-931	4.7	65

15	Zebrafish (Danio rerio, Hamilton-Buchanan, 1822) as a model to study bone diseases associated with Rett syndrome. *Journal of Applied Ichthyology*, **2018**, 34, 489-500	0.9	
14	X chromosome inactivation does not necessarily determine the severity of the phenotype in Rett syndrome patients. *Scientific Reports*, **2019**, 9, 11983	4.9	3
13	Novel Mutation c.1162_1172del; p.Pro388* in Two Patients with Symptoms of Atypical Rett Syndrome. *Molecular Syndromology*, **2019**, 10, 223-228	1.5	1
12	First description of an unusual novel double mutation in MECP2 co-occurring with the m.827A>G mutation in the MT-RNR1 gene associated with angelman-like syndrome. *International Journal of Developmental Neuroscience*, **2019**, 79, 37-44	2.7	2
11	Proteomic analyses reveal misregulation of LIN28 expression and delayed timing of glial differentiation in human iPS cells with MECP2 loss-of-function. *PLoS ONE*, **2019**, 14, e0212553	3.7	18
10	Mutations in STAG2 cause an X-linked cohesinopathy associated with undergrowth, developmental delay, and dysmorphia: Expanding the phenotype in males. *Molecular Genetics & Genomic Medicine*, **2019**, 7, e00501	2.3	17
9	Leveraging the genetic basis of Rett syndrome to ascertain pathophysiology. *Neurobiology of Learning and Memory*, **2019**, 165, 106961	3.1	2
8	Diurnal variation in autonomic regulation among patients with genotyped Rett syndrome. *Journal of Medical Genetics*, **2020**, 57, 786-793	5.8	7
7	Abnormalities of mitochondrial dynamics and bioenergetics in neuronal cells from CDKL5 deficiency disorder. *Neurobiology of Disease*, **2021**, 155, 105370	7.5	3
6	The Impact of X-Chromosome Inactivation on Phenotypic Expression of X-Linked Neurodevelopmental Disorders. *Brain Sciences*, **2021**, 11,	3.4	2
5	DNA methylation signatures in autism spectrum disorders. *Nucleus (India)*, 1	1.7	0
4	A catalogue of 863 Rett-syndrome-causing MECP2 mutations and lessons learned from data integration. *Scientific Data*, **2021**, 8, 10	8.2	4
3	Apoptotic Activity of MeCP2 Is Enhanced by C-Terminal Truncating Mutations. *PLoS ONE*, **2016**, 11, e0159632	3.7	2
2	R. **2007**, 680-714		
1	Degenerative Disorders Primarily of Gray Matter. **2012**, 518-543		