Masafumi Matsuo

List of Publications by Year
in descending order

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94 papers	1,647 citations	331670 21 h-index	377865 34 g-index
97 all docs	97 docs citations	97 times ranked	2112 citing authors

#	Article	IF	Citations
1	Early pathogenesis of Duchenne muscular dystrophy modelled in patient-derived human induced pluripotent stem cells. Scientific Reports, 2015, 5, 12831.	3.3	99
2	Chemical treatment enhances skipping of a mutated exon in the dystrophin gene. Nature Communications, 2011, 2, 308.	12.8	81
3	Establishment of a highly sensitive sandwich ELISA for the N-terminal fragment of titin in urine. Scientific Reports, 2016, 6, 39375.	3.3	58
4	Duchenne/Becker muscular dystrophy: From molecular diagnosis to gene therapy. Brain and Development, 1996, 18, 167-172.	1.1	55
5	A Japanese child with asymptomatic elevation of serum creatine kinase shows PTRF-CAVIN mutation matching with congenital generalized lipodystrophy type 4. Molecular Genetics and Metabolism, 2010, 101, 233-237.	1.1	54
6	A G-to-A transition at the fifth position of intron-32 of the dystrophin gene inactivates a splice-donor site both in vivo and in vitro. Molecular Genetics and Metabolism, 2005, 85, 213-219.	1.1	50
7	Development of an exon skipping therapy for X-linked Alport syndrome with truncating variants in COL4A5. Nature Communications, 2020, 11, 2777.	12.8	46
8	Detection of Splicing Abnormalities and Genotype-Phenotype Correlation in X-linked Alport Syndrome. Journal of the American Society of Nephrology: JASN, 2018, 29, 2244-2254.	6.1	43
9	Involvement of aldehyde dehydrogenase 1A2 in the regulation of cancer stem cell properties in neuroblastoma. International Journal of Oncology, 2015, 46, 1089-1098.	3.3	41
10	Cellular senescence-mediated exacerbation of Duchenne muscular dystrophy. Scientific Reports, 2020, 10, 16385.	3.3	40
11	Intragenic mutations in SMN1 may contribute more significantly to clinical severity than SMN2 copy numbers in some spinal muscular atrophy (SMA) patients. Brain and Development, 2014, 36, 914-920.	1.1	39
12	Common risk variants in NPHS1 and TNFSF15 are associated with childhood steroid-sensitive nephrotic syndrome. Kidney International, 2020, 98, 1308-1322.	5.2	39
13	Diagnostic and clinical significance of the titin fragment in urine of Duchenne muscular dystrophy patients. Clinica Chimica Acta, 2018, 476, 111-116.	1.1	37
14	High Incidence of Electrocardiogram Abnormalities in Young Patients With Duchenne Muscular Dystrophy. Pediatric Neurology, 2008, 39, 399-403.	2.1	36
15	In vivo and in vitro splicing assay of SLC12A1 in an antenatal salt-losing tubulopathy patient with an intronic mutation. Human Genetics, 2009, 126, 533-538.	3.8	36
16	A Deep Intronic Mutation in the SLC12A3 Gene Leads to Gitelman Syndrome. Pediatric Research, 2009, 66, 590-593.	2.3	35
17	Novel missense mutation of the UGT1A1 gene in Thai siblings with Gilbert's syndrome. Pediatrics International, 2002, 44, 427-432.	0.5	33
18	Cardiac Dysfunction in Duchenne Muscular Dystrophy Is Less Frequent in Patients With Mutations in the Dystrophin Dp116 Coding Region Than in Other Regions. Circulation Genomic and Precision Medicine, 2018, 11, e001782.	3.6	32

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19	Co-occurrence of mutations in both dystrophin- and androgen-receptor genes is a novel cause of female Duchenne muscular dystrophy. Human Genetics, 2006, 119, 516-519.	3.8	28
20	Detection of Dystrophin Dp71 in Human Skeletal Muscle Using an Automated Capillary Western Assay System. International Journal of Molecular Sciences, 2018, 19, 1546.	4.1	28
21	Antisense Oligonucleotide-Mediated Exon-skipping Therapies: Precision Medicine Spreading from Duchenne Muscular Dystrophy. JMA Journal, 2021, 4, 232-240.	0.8	27
22	2â€²-O-Methyl RNA/Ethylene-Bridged Nucleic Acid Chimera Antisense Oligonucleotides to Induce Dystrophin Exon 45 Skipping. Genes, 2017, 8, 67.	2.4	26
23	Exon skipping induced by nonsense/frameshift mutations in DMD gene results in Becker muscular dystrophy. Human Genetics, 2020, 139, 247-255.	3.8	23
24	Urinary Titin Is a Novel Biomarker for Muscle Atrophy in Nonsurgical Critically Ill Patients: A Two-Center, Prospective Observational Study. Critical Care Medicine, 2020, 48, 1327-1333.	0.9	22
25	A novel cryptic exon identified in the 3â€² region of intron 2 of the human dystrophin gene. Journal of Human Genetics, 2005, 50, 425-433.	2.3	21
26	Duchenne and Becker Muscular Dystrophy: From Gene Diagnosis to Molecular Therapy. IUBMB Life, 2002, 53, 147-152.	3.4	20
27	Titin fragment in urine: A noninvasive biomarker of muscle degradation. Advances in Clinical Chemistry, 2019, 90, 1-23.	3.7	20
28	Two alternative exons can result from activation of the cryptic splice acceptor site deep within intronÂ2 of the dystrophin gene in a patient with as yet asymptomatic dystrophinopathy. Human Genetics, 2003, 112, 164-170.	3.8	19
29	A nonsense mutation-created intraexonic splice site is active in the lymphocytes, but not in the skeletal muscle of a DMD patient. Human Genetics, 2006, 120, 737-742.	3.8	19
30	Patients with Duchenne muscular dystrophy are significantly shorter than those with Becker muscular dystrophy, with the higher incidence of short stature in Dp71 mutated subgroup. Neuromuscular Disorders, 2017, 27, 1023-1028.	0.6	19
31	Novel double-deletion mutations of the OFD1 gene creating multiple novel transcripts. Human Genetics, 2004, 115, 97-103.	3.8	18
32	Identification of seven novel cryptic exons embedded in the dystrophin gene and characterization of 14 cryptic dystrophin exons. Journal of Human Genetics, 2007, 52, 607-617.	2.3	18
33	Dystrophin Dp116: A yet to Be Investigated Product of the Duchenne Muscular Dystrophy Gene. Genes, 2017, 8, 251.	2.4	16
34	Pathogenic evaluation of synonymous COL4A5 variants in Xâ€linked Alport syndrome using a minigene assay. Molecular Genetics & Genomic Medicine, 2020, 8, e1342.	1.2	16
35	Neuronal SH-SY5Y cells use the C-dystrophin promoter coupled with exon 78 skipping and display multiple patterns of alternative splicing including two intronic insertion events. Human Genetics, 2015, 134, 993-1001.	3.8	15
36	A comparison of splicing assays to detect an intronic variant of the OCRL gene in Lowe syndrome. European Journal of Medical Genetics, 2017, 60, 631-634.	1.3	15

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37	Contribution of Rare Variants of the SLC22A12 Gene to the Missing Heritability of Serum Urate Levels. Genetics, 2020, 214, 1079-1090.	2.9	15
38	HEK293 cells express dystrophin Dp71 with nucleus-specific localization of Dp71ab. Histochemistry and Cell Biology, 2016, 146, 301-309.	1.7	14
39	Contributions of Japanese patients to development of antisense therapy for DMD. Brain and Development, 2016, 38, 4-9.	1.1	14
40	Determination of the pathogenicity of known COL4A5 intronic variants by in vitro splicing assay. Scientific Reports, 2019, 9, 12696.	3.3	14
41	Dystrophin Dp71ab is monoclonally expressed in human satellite cells and enhances proliferation of myoblast cells. Scientific Reports, 2020, 10, 17123.	3.3	14
42	Haploinsufficiency of thec-myctranscriptional repressorFIR, as a dominant negative-alternative splicing model, promoted p53-dependent T-cell acute lymphoblastic leukemia progression by activating Notch1. Oncotarget, 2015, 6, 5102-5117.	1.8	14
43	Last Nucleotide Substitutions of COL4A5 Exons Cause Aberrant Splicing. Kidney International Reports, 2022, 7, 108-116.	0.8	14
44	Tandem duplications of two separate fragments of the dystrophin gene in a patient with Duchenne muscular dystrophy. Journal of Human Genetics, 2008, 53, 215-219.	2.3	13
45	A novel cryptic exon in intron 3 of the dystrophin gene was incorporated into dystrophin mRNA with a single nucleotide deletion in exon 5. Journal of Human Genetics, 2002, 47, 196-201.	2.3	12
46	High prevalence of Southeast Asian ovalocytosis in Malays with distal renal tubular acidosis. Journal of Human Genetics, 2003, 48, 650-653.	2.3	12
47	Early detection of tumor relapse/regrowth by consecutive minimal residual disease monitoring in high-risk neuroblastoma patients. Oncology Letters, 2016, 12, 1119-1123.	1.8	12
48	Skipping of an exon with a nonsense mutation in the DMD gene is induced by the conversion of a splicing enhancer to a splicing silencer. Human Genetics, 2019, 138, 771-785.	3.8	12
49	Molecular assay for an intronic variant in NUP93 that causes steroid resistant nephrotic syndrome. Journal of Human Genetics, 2019, 64, 673-679.	2.3	12
50	Systematic Review of Genotype-Phenotype Correlations in Frasier Syndrome. Kidney International Reports, 2021, 6, 2585-2593.	0.8	12
51	Novel missense mutation of the UGT1A1 gene in Thai siblings with Gilbert's syndrome. Pediatrics International, 2002, 44, 427-432.	0.5	12
52	The ACTN3 577XX Null Genotype Is Associated with Low Left Ventricular Dilation-Free Survival Rate in Patients with Duchenne Muscular Dystrophy. Journal of Cardiac Failure, 2020, 26, 841-848.	1.7	11
53	Differential expression of minimal residual disease markers in peripheral blood and bone marrow samples from high-risk neuroblastoma patients. Oncology Letters, 2015, 10, 3228-3232.	1.8	10
54	An in vitro splicing assay reveals the pathogenicity of a novel intronic variant in ATP6V0A4 for autosomal recessive distal renal tubular acidosis. BMC Nephrology, 2017, 18, 353.	1.8	10

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55	Identification of the shortest splice variant of Dp71, together with five known variants, in glioblastoma cells. Biochemical and Biophysical Research Communications, 2019, 508, 640-645.	2.1	10
56	Tissue- and case-specific retention of intron 40 in mature dystrophin mRNA. Journal of Human Genetics, 2015, 60, 327-333.	2.3	9
57	Alternative splicing of a cryptic exon embedded in intron 6 of SMN1 and SMN2. Human Genome Variation, 2016, 3, 16040.	0.7	9
58	Monophasic Pulsed Microcurrent of 1â€“8â€…Hz Increases the Number of Human Dermal Fibroblasts. Progress in Rehabilitation Medicine, 2016, 1, n/a.	0.9	9
59	DMD transcripts in CRL-2061 rhabdomyosarcoma cells show high levels of intron retention by intron-specific PCR amplification. Cancer Cell International, 2017, 17, 58.	4.1	9
60	Receiver operating curve analyses of urinary titin of healthy 3-y-old children may be a noninvasive screening method for Duchenne muscular dystrophy. Clinica Chimica Acta, 2018, 486, 110-114.	1.1	9
61	Functional analysis of suspected splicing variants in CLCN5 gene in Dent disease 1. Clinical and Experimental Nephrology, 2020, 24, 606-612.	1.6	9
62	Urinary Titin N-Fragment as a Biomarker of Muscle Atrophy, Intensive Care Unit-Acquired Weakness, and Possible Application for Post-Intensive Care Syndrome. Journal of Clinical Medicine, 2021, 10, 614.	2.4	9
63	Wide ranges of serum myostatin concentrations in Duchenne muscular dystrophy patients. Clinica Chimica Acta, 2008, 391, 115-117.	1.1	8
64	Renadirsen, a Novel 2â€²OMeRNA/ENAÂ® Chimera Antisense Oligonucleotide, Induces Robust Exon 45 Skipping for Dystrophin In Vivo. Current Issues in Molecular Biology, 2021, 43, 1267-1281.	2.4	8
65	Cryptic splice activation but not exon skipping is observed in minigene assays of dystrophin c.9361+1G>A mutation identified by NGS. Journal of Human Genetics, 2017, 62, 531-537.	2.3	7
66	Functional splicing analysis in an infantile case of atypical hemolytic uremic syndrome caused by digenic mutations in C3 and MCP genes. Journal of Human Genetics, 2018, 63, 755-759.	2.3	7
67	Staurosporine allows dystrophin expression by skipping of nonsense-encoding exon. Brain and Development, 2016, 38, 738-745.	1.1	6
68	Schwann cell-specific Dp116 is expressed in glioblastoma cells, revealing two novel DMD gene splicing patterns. Biochemistry and Biophysics Reports, 2019, 20, 100703.	1.3	6
69	Pathological evaluation of rats carrying in-frame mutations in the dystrophin gene: A new model of Becker muscular dystrophy. DMM Disease Models and Mechanisms, 2020, 13, .	2.4	6
70	A sandwich ELISA kit reveals marked elevation of titin Nâ€terminal fragment levels in the urine of mdx mice. Animal Models and Experimental Medicine, 2022, 5, 48-55.	3.3	6
71	Intron-retained transcripts of the spinal muscular atrophy genes, SMN1 and SMN2. Brain and Development, 2018, 40, 670-677.	1.1	5
72	Detection of a Splice Site Variant in a Patient with Glomerulopathy and Fibronectin Deposits. Nephron, 2018, 138, 166-171.	1.8	5

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73	Comprehensive genetic analysis using next-generation sequencing for the diagnosis of nephronophthisis-related ciliopathies in the Japanese population. Journal of Human Genetics, 2022, 67, 427-440.	2.3	5
74	Identification of sleep hypoventilation in young individuals with Becker muscular dystrophy: A pilot study. Brain and Development, 2018, 40, 537-543.	1.1	4
75	Rbfox2 mediates exon 11 inclusion in insulin receptor pre-mRNA splicing in hepatoma cells. Biochimie, 2021, 187, 25-32.	2.6	4
76	Intronic Alternative Polyadenylation in the Middle of the DMD Gene Produces Half-Size N-Terminal Dystrophin with a Potential Implication of ECG Abnormalities of DMD Patients. International Journal of Molecular Sciences, 2020, 21, 3555.	4.1	4
77	Study on mutations affecting the muscle promoter/first exon of the dystrophin gene in 92 Japanese dilated cardiomyopathy patients. , 1998, 79, 226-227.		3
78	Two closely spaced nonsense mutations in the DMD gene in a Malaysian family. Molecular Genetics and Metabolism, 2011, 103, 303-304.	1.1	3
79	Ambulatory capacity in Japanese patients with Duchenne muscular dystrophy. Brain and Development, 2018, 40, 465-472.	1.1	3
80	Dual Fluorescence Splicing Reporter Minigene Identifies an Antisense Oligonucleotide to Skip Exon v8 of the CD44 Gene. International Journal of Molecular Sciences, 2020, 21, 9136.	4.1	3
81	Usefulness of functional splicing analysis to confirm precise disease pathogenesis in Diamond-Blackfan anemia caused by intronic variants in RPS19. Pediatric Hematology and Oncology, 2021, 38, 515-527.	0.8	3
82	Urinary titin as a biomarker in Fukuyama congenital muscular dystrophy. Neuromuscular Disorders, 2021, 31, 194-197.	0.6	3
83	Dystrophin Dp71 Subisoforms Localize to the Mitochondria of Human Cells. Life, 2021, 11, 978.	2.4	3
84	Evaluation of suspected autosomal Alport Syndrome synonymous variants. Kidney360, 2022, 3, 10.34067/KID.0005252021.	2.1	3
85	Spinal Muscular Atrophy: New Screening System with Real-Time mCOP-PCR and PCR-RFLP for SMN1 Deletion. Kobe Journal of Medical Sciences, 2019, 65, E44-E48.	0.2	3
86	Human Dystrophin Dp71ab Enhances the Proliferation of Myoblasts Across Species But Not Human Nonmyoblast Cells. Frontiers in Cell and Developmental Biology, 2022, 10, 877612.	3.7	3
87	Clinical features of autosomal recessive polycystic kidney disease in the Japanese population and analysis of splicing in PKHD1 gene for determination of phenotypes. Clinical and Experimental Nephrology, 2022, 26, 140-153.	1.6	2
88	Spinal Muscular Atrophy: Advanced Version of Screening System with Real-Time mCOP-PCR and PCR-RFLP for SMN1 Deletion. Kobe Journal of Medical Sciences, 2019, 65, E49-E53.	0.2	2
89	A disease-causing variant of COL4A5 in a Chinese family with Alport syndrome: a case series. BMC Nephrology, 2021, 22, 380.	1.8	2
90	An Antisense Oligonucleotide against a Splicing Enhancer Sequence within Exon 1 of the MSTN Gene Inhibits Pre-mRNA Maturation to Act as a Novel Myostatin Inhibitor. International Journal of Molecular Sciences, 2022, 23, 5016.	4.1	2

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91	Can urinary titin be used for predicting Duchenne muscular dystrophy?. Clinica Chimica Acta, 2019, 490, 162.	1.1	1
92	Assessment of catabolic state in infants with the use of urinary titin N-fragment. Pediatric Research, 2021, , .	2.3	1
93	Dystrophin Dp116: A yet to Be Investigated Product of the Duchenne Muscular Dystrophy Gene. Genes, 2017, 8, 251.	2.4	0
94	Onset mechanism of a female patient with Dent disease 2. Clinical and Experimental Nephrology, 2020, 24, 946-954.	1.6	0