

Jean-Claude Kaplan

List of Publications by Year in descending order

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57
papers

5,665
citations

126858

33
h-index

114418

63
g-index

67
all docs

67
docs citations

67
times ranked

3717
citing authors

#	ARTICLE	IF	CITATIONS
1	The 2015 version of the gene table of monogenic neuromuscular disorders (nuclear genome). Neuromuscular Disorders, 2014, 24, 1123-1153.	0.3	43
2	Assessment of the structural and functional impact of in-frame mutations of the DMD gene, using the tools included in the eDystrophin online database. Orphanet Journal of Rare Diseases, 2012, 7, 45.	1.2	45
3	Variable phenotype of del45-55 Becker patients correlated with nNOS μ mislocalization and RYR1 hypernitrosylation. Human Molecular Genetics, 2012, 21, 3449-3460.	1.4	43
4	Genotype-phenotype analysis in 2,405 patients with a dystrophinopathy using the UMD-DMD database: a model of nationwide knowledgebase. Human Mutation, 2009, 30, 934-945.	1.1	309
5	Protein- and mRNA-based phenotype-genotype correlations in DMD/BMD with point mutations and molecular basis for BMD with nonsense and frameshift mutations in the DMD gene. Human Mutation, 2007, 28, 183-195.	1.1	107
6	Rescue of Dystrophic Muscle Through U7 snRNA-Mediated Exon Skipping. Science, 2004, 306, 1796-1799.	6.0	454
7	Phase I Study of Dystrophin Plasmid-Based Gene Therapy in Duchenne/Becker Muscular Dystrophy. Human Gene Therapy, 2004, 15, 1065-1076.	1.4	134
8	Dystrophinopathy caused by mid-intronic substitutions activating cryptic exons in the DMD gene. Neuromuscular Disorders, 2004, 14, 10-18.	0.3	46
9	Gene Location. Neuromuscular Disorders, 2004, 14, 85-106.	0.3	2
10	CFTR mutations in patients from Colombia: Implications for local and regional molecular diagnosis programs. Human Mutation, 2003, 22, 259-259.	1.1	20
11	Looking under every rock: Duchenne muscular dystrophy and traditional Chinese medicine. Neuromuscular Disorders, 2003, 13, 705-707.	0.3	11
12	Neuromuscular disorders: gene location. Neuromuscular Disorders, 2002, 12, 82-100.	0.3	1
13	A biochemical, genetic, and clinical survey of autosomal recessive limb girdle muscular dystrophies in Turkey. Annals of Neurology, 1997, 42, 222-229.	2.8	94
14	Absence of β -sarcoglycan (35 DAG) in autosomal recessive muscular dystrophy linked to chromosome 13q12. FEBS Letters, 1996, 381, 15-20.	1.3	48
15	Identification of three novel mutations in the cystic fibrosis transmembrane conductance regulator gene in Argentinian CF patients. , 1996, 7, 376-377.		3
16	Screening for mutations in factor VIII gene using the single-strand conformation polymorphism. Human Mutation, 1995, 5, 357-359.	1.1	0
17	Mutation heterogeneity of cystic fibrosis in France: Screening by denaturing gradient gel electrophoresis using psoralen-modified oligonucleotide. Human Mutation, 1995, 6, 23-29.	1.1	32
18	At least five polymorphic mutants account for the prevalence of glucose-6-phosphate dehydrogenase deficiency in Algeria. Human Genetics, 1994, 94, 513-7.	1.8	36

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19	Mutations in the DAX-1 gene give rise to both X-linked adrenal hypoplasia congenita and hypogonadotropic hypogonadism. <i>Nature</i> , 1994, 372, 672-676.	13.7	722
20	Heterogeneity of the rat NADH-cytochrome-b5-reductase transcripts resulting from multiple alternative first exons. <i>FEBS Journal</i> , 1994, 220, 729-737.	0.2	13
21	Missense mutations in the adhalin gene linked to autosomal recessive muscular dystrophy. <i>Cell</i> , 1994, 78, 625-633.	13.5	463
22	Striking conservation of the brain-specific region of the dystrophin gene. <i>Mammalian Genome</i> , 1993, 4, 393-396.	1.0	4
23	Long-term correction of mouse dystrophic degeneration by adenovirus-mediated transfer of a minidystrophin gene. <i>Nature Genetics</i> , 1993, 5, 130-134.	9.4	215
24	The role of the dystrophin-glycoprotein complex in the molecular pathogenesis of muscular dystrophies. <i>Neuromuscular Disorders</i> , 1993, 3, 533-535.	0.3	51
25	G6PD Aures: a new mutation (48 Ile → Thr) causing mild G6PD deficiency is associated with favism. <i>Human Molecular Genetics</i> , 1993, 2, 81-82.	1.4	27
26	Skipping of exon 9 in CFTR mRNA of human adult and fetal pancreas from non-CF individuals. <i>Human Molecular Genetics</i> , 1993, 2, 2141-2142.	1.4	4
27	Severe childhood autosomal recessive muscular dystrophy with the deficiency of the 50 kDa dystrophin-associated glycoprotein maps to chromosome 13q12. <i>Human Molecular Genetics</i> , 1993, 2, 1423-1428.	1.4	104
28	A YAC contig in Xp21 containing the adrenal hypoplasia congenita and glycerol kinase deficiency genes. <i>Human Molecular Genetics</i> , 1992, 1, 579-585.	1.4	33
29	CFTR illegitimate transcription in lymphoid cells: quantification and applications to the investigation of pathological transcripts. <i>Human Genetics</i> , 1992, 88, 508-512.	1.8	34
30	Deficiency of the 50K dystrophin-associated glycoprotein in severe childhood autosomal recessive muscular dystrophy. <i>Nature</i> , 1992, 359, 320-322.	13.7	262
31	A novel mutation (Arg → Leu in exon 18) in factor VIII gene responsible for moderate hemophilia A. <i>Human Mutation</i> , 1992, 1, 77-78.	1.1	6
32	Illegitimate transcription: Its use in the study of inherited disease. <i>Human Mutation</i> , 1992, 1, 357-360.	1.1	80
33	Illegitimate (or ectopic) transcription proceeds through the usual promoters. <i>Biochemical and Biophysical Research Communications</i> , 1991, 178, 553-557.	1.0	19
34	Immunolocalization and developmental expression of dystrophin related protein in skeletal muscle. <i>Neuromuscular Disorders</i> , 1991, 1, 185-194.	0.3	242
35	Dystrophin gene transcribed from different promoters in neuronal and glial cells. <i>Nature</i> , 1990, 344, 64-65.	13.7	159
36	Quantitative estimation of minor mRNAs by cDNA-polymerase chain reaction. Application to dystrophin mRNA in cultured myogenic and brain cells. <i>FEBS Journal</i> , 1990, 187, 691-698.	0.2	178

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37	Effect of dystrophin gene deletions on mRNA levels and processing in Duchenne and Becker muscular dystrophies. <i>Cell</i> , 1990, 63, 1239-1248.	13.5	165
38	Transcription of the dystrophin gene in human muscle and non-muscle tissues. <i>Nature</i> , 1988, 333, 858-860.	13.7	760
39	Expression of NADH-cytochrome b5 reductase during dimethyl sulfoxide-induced differentiation of Friend erythroleukemia cells. <i>FEBS Letters</i> , 1982, 143, 35-39.	1.3	2
40	3',5'-cyclic nucleotide phosphodiesterase from human platelets: effect of heat upon the multiple forms and their interconversion. <i>Biochimie</i> , 1981, 63, 603-609.	1.3	3
41	Diaphorase P: A new fetal isozyme identified in human placenta. <i>Biochimica Et Biophysica Acta - Biomembranes</i> , 1980, 613, 18-25.	1.4	5
42	Direct Enzyme Titration Curve of NADH: Cytochrome b5 Reductase by Combined Isoelectric Focusing/Electrophoresis. Interactions between Enzyme and Cytochrome b5. <i>FEBS Journal</i> , 1980, 112, 179-183.	0.2	14
43	Study of a case with severe red-cell pyrimidine 5'-nucleotidase deficiency. <i>Clinica Chimica Acta</i> , 1979, 95, 83-88.	0.5	10
44	Assignment of NADH-cytochrome b5 reductase (DIA1 locus) to human chromosome 22. <i>Human Genetics</i> , 1978, 42, 233-239.	1.8	36
45	Titration curves of interacting cytochrome b5 and hemoglobin by isoelectric focusing-electrophoresis. <i>Biochemical and Biophysical Research Communications</i> , 1978, 85, 1575-1581.	1.0	51
46	Red-cell pyrimidine 5'-nucleotidase and lead poisoning. <i>Clinica Chimica Acta</i> , 1978, 87, 49-55.	0.5	36
47	A radioassay for pyrimidine-5'-nucleotidase activity. <i>Clinica Chimica Acta</i> , 1978, 85, 193-196.	0.5	13
48	Phenobarbital-induced increase of NADH-cytochrome b5 reductase activity in rat liver microsomes. <i>Biochemical Pharmacology</i> , 1978, 27, 367-368.	2.0	7
49	Soluble and microsomal forms of NADH-cytochrome b5 reductase from human placenta Similarity with NADH-methemoglobin reductase from human erythrocytes. <i>Biochimica Et Biophysica Acta - Biomembranes</i> , 1977, 481, 50-62.	1.4	42
50	Generalised deficiency of cytochrome b5 reductase in congenital methaemoglobinaemia with mental retardation. <i>Nature</i> , 1975, 258, 619-620.	13.7	142
51	Effect of N ⁶ , 2'-O-dibutyl cyclic AMP upon the interconvertible forms of cyclic AMP phosphodiesterase from human platelets. <i>Biochemical and Biophysical Research Communications</i> , 1975, 64, 342-346.	1.0	10
52	Kinetic and electrophoretic abnormality of cyclic AMP phosphodiesterase in genetically obese mouse adipocytes. <i>Biochemical and Biophysical Research Communications</i> , 1973, 51, 1008-1014.	1.0	18
53	Multiple forms of cyclic adenosine 3',5'-monophosphate phosphodiesterase from human blood platelets. I. Kinetic and electrophoretic characterization of two molecular species. <i>Biochimica Et Biophysica Acta - Biomembranes</i> , 1973, 315, 370-377.	1.4	26
54	Human brain and platelet cyclic adenosine 3',5'-monophosphate phosphodiesterases: Different response to drugs. <i>Biochimica Et Biophysica Acta - General Subjects</i> , 1972, 279, 217-220.	1.1	33

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55	Presence of red cell type NADH-methemoglobin reductase (NADH-diaphorase) in human non erythroid cells. Biochemical and Biophysical Research Communications, 1972, 49, 945-950.	1.0	28
56	Electrophoretic Study of Glutathione Reductase in Human Erythrocytes and Leucocytes. Nature, 1968, 217, 256-258.	13.7	54
57	Electrophoresis of red cell NADH- and NADPH-diaphorases in normal subjects and patients with congenital methemoglobinemia. Biochemical and Biophysical Research Communications, 1967, 29, 605-610.	1.0	117