Jean-Claude Kaplan

List of Publications by Year
in descending order

57 papers	5,665 citations	126907 33 h-index	114465 63 g-index
67 all docs	67 docs citations	67 times ranked	3717 citing authors

#	Article	IF	Citations
1	Transcription of the dystrophin gene in human muscle and non-muscle tissues. Nature, 1988, 333, 858-860.	27.8	760
2	Mutations in the DAX-1 gene give rise to both X-linked adrenal hypoplasia congenita and hypogonadotropic hypogonadism. Nature, 1994, 372, 672-676.	27.8	722
3	Missense mutations in the adhalin gene linked to autosomal recessive muscular dystrophy. Cell, 1994, 78, 625-633.	28.9	463
4	Rescue of Dystrophic Muscle Through U7 snRNA-Mediated Exon Skipping. Science, 2004, 306, 1796-1799.	12.6	454
5	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.	2.5	309
6	Deficiency of the 50K dystrophin-associated glycoprotein in severe childhood autosomal recessive muscular dystrophy. Nature, 1992, 359, 320-322.	27.8	262
7	Immunolocalization and developmental expression of dystrophin related protein in skeletal muscle. Neuromuscular Disorders, 1991, 1, 185-194.	0.6	242
8	Longâ€“term correction of mouse dystrophic degeneration by adenovirusâ€“mediated transfer of a minidystrophin gene. Nature Genetics, 1993, 5, 130-134.	21.4	215
9	Quantitative estimation of minor mRNAs by cDNA-polymerase chain reaction. Application to dystrophin mRNA in cultured myogenic and brain cells. FEBS Journal, 1990, 187, 691-698.	0.2	178
10	Effect of dystrophin gene deletions on mRNA levels and processing in Duchenne and Becker muscular dystrophies. Cell, 1990, 63, 1239-1248.	28.9	165
11	Dystrophin gene transcribed from different promoters in neuronal and glial cells. Nature, 1990, 344, 64-65.	27.8	159
12	Generalised deficiency of cytochrome b5 reductase in congenital methaemoglobinaemia with mental retardation. Nature, 1975, 258, 619-620.	27.8	142
13	Phase I Study of Dystrophin Plasmid-Based Gene Therapy in Duchenne/Becker Muscular Dystrophy. Human Gene Therapy, 2004, 15, 1065-1076.	2.7	134
14	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.	2.1	117
15	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.	2.5	107
16	Severe childhood autosomal recessive muscular dystrophy with the deficiency of the 50 kDa dystrophin-associated glycoprotein maps to chromosome 13q12. Human Molecular Genetics, 1993, 2, 1423-1428.	2.9	104
17	A biochemical, genetic, and clinical survey of autosomal recessive limb girdle muscular dystrophies in Turkey. Annals of Neurology, 1997, 42, 222-229.	5.3	94
18	Illegitimate transcription: Its use in the study of inherited disease. Human Mutation, 1992, 1, 357-360.	2.5	80

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19	Electrophoretic Study of Glutathione Reductase in Human Erythrocytes and Leucocytes. Nature, 1968, 217, 256-258.	27.8	54
20	Titration curves of interacting cytochrome b5 and hemoglobin by isoelectric focusing-electrophoresis. Biochemical and Biophysical Research Communications, 1978, 85, 1575-1581.	2.1	51
21	The role of the dystrophin-glycoprotein complex in the molecular pathogenesis of muscular dystrophies. Neuromuscular Disorders, 1993, 3, 533-535.	0.6	51
22	Absence of Î³â€sarcoglycan (35 DAG) in autosomal recessive muscular dystrophy linked to chromosome 13q12. FEBS Letters, 1996, 381, 15-20.	2.8	48
23	Dystrophinopathy caused by mid-intronic substitutions activating cryptic exons in the DMD gene. Neuromuscular Disorders, 2004, 14, 10-18.	0.6	46
24	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.	2.7	45
25	Variable phenotype of del45-55 Becker patients correlated with nNOSÂµ mislocalization and RYR1 hypernitrosylation. Human Molecular Genetics, 2012, 21, 3449-3460.	2.9	43
26	The 2015 version of the gene table of monogenic neuromuscular disorders (nuclear genome). Neuromuscular Disorders, 2014, 24, 1123-1153.	0.6	43
27	Soluble and microsomal forms of NADH-cytochrome b5 reductase from human placenta Similarity with NADH-methemoglobin reductase from human erythrocytes. Biochimica Et Biophysica Acta - Biomembranes, 1977, 481, 50-62.	2.6	42
28	Assignment of NADH-cytochrome b5 reductase (DIA1 locus) to human chromosome 22. Human Genetics, 1978, 42, 233-239.	3.8	36
29	Red-cell pyrimidine 5â€²-nucleotidase and lead poisoning. Clinica Chimica Acta, 1978, 87, 49-55.	1.1	36
30	At least five polymorphic mutants account for the prevalence of glucose-6-phosphate dehydrogenase deficiency in Algeria. Human Genetics, 1994, 94, 513-7.	3.8	36
31	CFTR illegitimate transcription in lymphoid cells: quantification and applications to the investigation of pathological transcripts. Human Genetics, 1992, 88, 508-512.	3.8	34
32	Human brain and platelet cyclic adenosine 3â€²,5â€²-monophosphate phosphodiesterases: Different response to drugs. Biochimica Et Biophysica Acta - General Subjects, 1972, 279, 217-220.	2.4	33
33	A YAC contig in Xp21 containing the adrenal hypoplasia congenita and glycerol kinase deficiency genes. Human Molecular Genetics, 1992, 1, 579-585.	2.9	33
34	Mutation heterogeneity of cystic fibrosis in France: Screening by denaturing gradient gel electrophoresis using psoralen-modified oligonucleotide. Human Mutation, 1995, 6, 23-29.	2.5	32
35	Presence of red cell type NADH-methemoglobin reductase (NADH-diaphorase) in human non erythroid cells. Biochemical and Biophysical Research Communications, 1972, 49, 945-950.	2.1	28
36	G6PD Aures: a new mutation (48 Ile â†' Thr) causing mild G6PD deficiency is associated with favism. Human Molecular Genetics, 1993, 2, 81-82.	2.9	27

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37	Multiple forms of cyclic adenosine 3â€²,5â€²-monophosphate phosphodiesterase from human blood platelets. I. Kinetic and electrophoretic characterization of two molecular species. Biochimica Et Biophysica Acta - Biomembranes, 1973, 315, 370-377.	2.6	26
38	CFTR mutations in patients from Colombia: Implications for local and regional molecular diagnosis programs. Human Mutation, 2003, 22, 259-259.	2.5	20
39	Illegitimate (or ectopic) transcription proceeds through the usual promoters. Biochemical and Biophysical Research Communications, 1991, 178, 553-557.	2.1	19
40	Kinetic and electrophoretic abnormality of cyclic AMP phosphodiesterase in genetically obese mouse adipocytes. Biochemical and Biophysical Research Communications, 1973, 51, 1008-1014.	2.1	18
41	Direct Enzyme Titration Curve of NADH: Cytochrome b5 Reductase by Combined Isoelectric Focusing/Electrophoresis. Interactions between Enzyme and Cytochrome b5. FEBS Journal, 1980, 112, 179-183.	0.2	14
42	A radioassay for pyrimidine-5â€²-nucleotidase activity. Clinica Chimica Acta, 1978, 85, 193-196.	1.1	13
43	Heterogeneity of the rat NADH-cytochrome-b5-reductase transcripts resulting from multiple alternative first exons. FEBS Journal, 1994, 220, 729-737.	0.2	13
44	Looking under every rock: Duchenne muscular dystrophy and traditional Chinese medicine. Neuromuscular Disorders, 2003, 13, 705-707.	0.6	11
45	Effect of N6, 2â€²-O-dibutyryl cyclic AMP upon the interconvertible forms of cyclic AMP phosphodiesterase from human platelets. Biochemical and Biophysical Research Communications, 1975, 64, 342-346.	2.1	10
46	Study of a case with severe red-cell pyrimidine 5'-nucleotidase deficiency. Clinica Chimica Acta, 1979, 95, 83-88.	1.1	10
47	Phenobarbital-induced increase of NADH-cytochrome b5 reductase activity in rat liver microsonies. Biochemical Pharmacology, 1978, 27, 367-368.	4.4	7
48	A novel mutation (Argâ†'Leu in exon 18) in factor VIII gene responsible for moderate hemophilia A. Human Mutation, 1992, 1, 77-78.	2.5	6
49	Diaphorase P: A new fetal isozyme identified in human placenta. Biochimica Et Biophysica Acta - Biomembranes, 1980, 613, 18-25.	2.6	5
50	Striking conservation of the brain-specific region of the dystrophin gene. Mammalian Genome, 1993, 4, 393-396.	2.2	4
51	Skipping of exon 9 in CFTR mRNA of human adult and fetal pancreas from non-CF individuals. Human Molecular Genetics, 1993, 2, 2141-2142.	2.9	4
52	3â€²,5â€² cyclic nucleotide phosphodiesterase from human platelets: effect of heat upon the multiple forms and their interconversion. Biochimie, 1981, 63, 603-609.	2.6	3
53	Identification of three novel mutations in the cystic fibrosis transmembrane conductance regulator gene in Argentinian CF patients. , 1996, 7, 376-377.		3
54	Expression of NADH-cytochromeb5reductase during dimethyl sulfoxide-induced differentiation of Friend erythroleukemia cells. FEBS Letters, 1982, 143, 35-39.	2.8	2

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55	Gene Location. Neuromuscular Disorders, 2004, 14, 85-106.	0.6	2
56	Neuromuscular disorders: gene location. Neuromuscular Disorders, 2002, 12, 82-100.	0.6	1
57	Screening for mutations in factor VIII gene using the single-strand conformation polymorphism. Human Mutation, 1995, 5, 357-359.	2.5	0