Uffe Birk Jensen

List of Publications by Year
in descending order

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132 papers	8,689 citations	50276 46 h-index	46799 89 g-index
136 all docs	136 docs citations	136 times ranked	15138 citing authors

#	Article	IF	Citations
1	Breast and Prostate Cancer Risks for MaleBRCA1andBRCA2Pathogenic Variant Carriers Using Polygenic Risk Scores. Journal of the National Cancer Institute, 2022, 114, 109-122.	6.3	19
2	Monoâ€allelic loss of <scp>YTHDF3</scp> and neurodevelopmental disorder: clinical features of four individuals with 8q12.3 deletions. Clinical Genetics, 2022, 101, 208-213.	2.0	2
3	A case-only study to identify genetic modifiers of breast cancer risk for BRCA1/BRCA2 mutation carriers. Nature Communications, 2021, 12, 1078.	12.8	19
4	A Search for Undiagnosed Charcot-Marie-Tooth Disease Among Patients Registered with Unspecified Polyneuropathy in the Danish National Patient Registry. Clinical Epidemiology, 2021, Volume 13, 113-120.	3.0	1
5	The CRISPR/Cas9 Minipigâ€"A Transgenic Minipig to Produce Specific Mutations in Designated Tissues. Cancers, 2021, 13, 3024.	3.7	12
6	The predictive ability of the 313 variantâ€"based polygenic risk score for contralateral breast cancer risk prediction in women of European ancestry with a heterozygous BRCA1 or BRCA2 pathogenic variant. Genetics in Medicine, 2021, 23, 1726-1737.	2.4	16
7	Association of Genomic Domains in BRCA1 and BRCA2 with Prostate Cancer Risk and Aggressiveness. Cancer Research, 2020, 80, 624-638.	0.9	39
8	Generation of eight human induced pluripotent stem cell lines from Parkinson's disease patients carrying familial mutations. Stem Cell Research, 2020, 42, 101657.	0.7	6
9	The role of stem cell antigen-1/Lymphocyte antigen 6A-2/6E-1 knock out in murine epidermis. Stem Cell Research, 2020, 49, 102047.	0.7	0
10	Deleterious misâ€splicing of STK11 caused by a novel singleâ€nucleotide substitution in the 3â€² polypyrimidine tract of intron five. Molecular Genetics & Genomic Medicine, 2020, 8, e1381.	1.2	3
11	Characterization of the Cancer Spectrum in Men With GermlineBRCA1andBRCA2Pathogenic Variants. JAMA Oncology, 2020, 6, 1218.	7.1	48
12	Generation of an induced pluripotent stem cell line (DANi-011A) from a Parkinson's disease patient with a LRRK2 p.G2019S mutation. Stem Cell Research, 2020, 45, 101781.	0.7	1
13	Population frequencies of pathogenic alleles of BRCA1 and BRCA2: analysis of 173 Danish breast cancer pedigrees using the BOADICEA model. Familial Cancer, 2019, 18, 381-388.	1.9	8
14	Exploring the hereditary background of renal cancer in Denmark. PLoS ONE, 2019, 14, e0215725.	2.5	10
15	Genetic analysis of Charcot-Marie-Tooth disease in Denmark and the implementation of a next generation sequencing platform. European Journal of Medical Genetics, 2019, 62, 1-8.	1.3	20
16	Mutational spectrum in a worldwide study of 29,700 families withBRCA1orBRCA2mutations. Human Mutation, 2018, 39, 593-620.	2.5	224
17	The BRCA1 c. 5096G>A p.Arg1699Gln (R1699Q) intermediate risk variant: breast and ovarian cancer risk estimation and recommendations for clinical management from the ENIGMA consortium. Journal of Medical Genetics, 2018, 55, 15-20.	3.2	50
18	CRISPR-C: circularization of genes and chromosome by CRISPR in human cells. Nucleic Acids Research, 2018, 46, e131.	14.5	39

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19	Full in-frame exon 3 skipping of BRCA2 confers high risk of breast and/or ovarian cancer. Oncotarget, 2018, 9, 17334-17348.	1.8	24
20	Current status of treating neurodegenerative disease with induced pluripotent stem cells. Acta Neurologica Scandinavica, 2017, 135, 57-72.	2.1	29
21	Isolating subpopulations of human epidermal basal cells based on polyclonal serum against trypsin-resistant CSPG4 epitopes. Experimental Cell Research, 2017, 350, 368-379.	2.6	2
22	A new technique for accelerated liver regeneration: An experimental study in rats. Surgery, 2017, 162, 233-247.	1.9	6
23	Magnetic resonance neurography and diffusion tensor imaging of the peripheral nerves in patients with <scp>C</scp>harcotâ€<scp>M</scp>arieâ€<scp>T</scp>ooth Type 1A. Muscle and Nerve, 2017, 56, E78-E84.	2.2	28
24	Development of hypomelanotic macules is associated with constitutive activated mTORC1 in tuberous sclerosis complex. Molecular Genetics and Metabolism, 2017, 120, 384-391.	1.1	5
25	Identification of 12 new susceptibility loci for different histotypes of epithelial ovarian cancer. Nature Genetics, 2017, 49, 680-691.	21.4	356
26	GRIN2Bencephalopathy: novel findings on phenotype, variant clustering, functional consequences and treatment aspects. Journal of Medical Genetics, 2017, 54, 460-470.	3.2	190
27	Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer. Nature Genetics, 2017, 49, 1767-1778.	21.4	289
28	Constitutive transgene expression of Stem Cell Antigen-1 in the hair follicle alters the sensitivity to tumor formation and progression. Stem Cell Research, 2017, 23, 109-118.	0.7	1
29	Charcot-Marie-Tooth disease in Denmark: a nationwide register-based study of mortality, prevalence and incidence. BMJ Open, 2017, 7, e018048.	1.9	10
30	Association of breast cancer risk in BRCA1 and BRCA2 mutation carriers with genetic variants showing differential allelic expression: identification of a modifier of breast cancer risk at locus 11q22.3. Breast Cancer Research and Treatment, 2017, 161, 117-134.	2.5	18
31	Genetic screening of the FLCN gene identify six novel variants and a Danish founder mutation. Journal of Human Genetics, 2017, 62, 151-157.	2.3	19
32	Mitochondrial Spare Respiratory Capacity Is Negatively Correlated with Nuclear Reprogramming Efficiency. Stem Cells and Development, 2017, 26, 166-176.	2.1	21
33	Preparation of A Spaceflight: Apoptosis Search in Sutured Wound Healing Models. International Journal of Molecular Sciences, 2017, 18, 2604.	4.1	11
34	Gene Expression in the Liver Remnant Is Significantly Affected by the Size of Partial Hepatectomy: An Experimental Rat Study. Gene Expression, 2017, 17, 289-299.	1.2	5
35	Prediction of Breast and Prostate Cancer Risks in Male BRCA1 and BRCA2 Mutation Carriers Using Polygenic Risk Scores. Journal of Clinical Oncology, 2017, 35, 2240-2250.	1.6	152
36	Validation of diagnostic codes for Charcot-Marie-Tooth disease in the Danish National Patient Registry. Clinical Epidemiology, 2016, Volume 8, 783-787.	3.0	4

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37	Fine-Scale Mapping at 9p22.2 Identifies Candidate Causal Variants That Modify Ovarian Cancer Risk in BRCA1 and BRCA2 Mutation Carriers. PLoS ONE, 2016, 11, e0158801.	2.5	10
38	<scp>JPâ€“HHT</scp> phenotype in Danish patients with <scp>SMAD4</scp> mutations. Clinical Genetics, 2016, 90, 55-62.	2.0	30
39	Identification of independent association signals and putative functional variants for breast cancer risk through fine-scale mapping of the 12p11 locus. Breast Cancer Research, 2016, 18, 64.	5.0	31
40	Male breast cancer in BRCA1 and BRCA2 mutation carriers: pathology data from the Consortium of Investigators of Modifiers of BRCA1/2. Breast Cancer Research, 2016, 18, 15.	5.0	88
41	Dataset on gene expression profiling of multiple murine hair follicle populations. Data in Brief, 2016, 9, 328-334.	1.0	1
42	Global gene expression and comparison between multiple populations in the mouse epidermis. Stem Cell Research, 2016, 17, 191-202.	0.7	10
43	Identification of four novel susceptibility loci for oestrogen receptor negative breast cancer. Nature Communications, 2016, 7, 11375.	12.8	93
44	Identification of eight novel SDHB, SDHC, SDHD germline variants in Danish pheochromocytoma/paraganglioma patients. Hereditary Cancer in Clinical Practice, 2016, 14, 13.	1.5	13
45	Danish retinoblastoma patients 1943â€“2013 â€“ genetic testing and clinical implications. Acta OncolÃ³gica, 2016, 55, 412-417.	1.8	10
46	No clinical utility of KRAS variant rs61764370 for ovarian or breast cancer. Gynecologic Oncology, 2016, 141, 386-401.	1.4	18
47	Difficulties in diagnosing Marfan syndrome using current FBN1 databases. Genetics in Medicine, 2016, 18, 98-102.	2.4	17
48	An original phylogenetic approach identified mitochondrial haplogroup T1a1 as inversely associated with breast cancer risk in BRCA2 mutation carriers. Breast Cancer Research, 2015, 17, 61.	5.0	26
49	Assessing Associations between the AURKA-HMMR-TPX2-TUBG1 Functional Module and Breast Cancer Risk in BRCA1/2 Mutation Carriers. PLoS ONE, 2015, 10, e0120020.	2.5	34
50	Transcriptional regulator PRDM12 is essential for human pain perception. Nature Genetics, 2015, 47, 803-808.	21.4	137
51	Identification of six new susceptibility loci for invasive epithelial ovarian cancer. Nature Genetics, 2015, 47, 164-171.	21.4	221
52	A novel single nucleotide splice site mutation in FHL1 confirms an Emery-Dreifuss plus phenotype with pulmonary artery hypoplasia and facial dysmorphology. European Journal of Medical Genetics, 2015, 58, 222-229.	1.3	11
53	Association of Type and Location ofBRCA1andBRCA2Mutations With Risk of Breast and Ovarian Cancer. JAMA - Journal of the American Medical Association, 2015, 313, 1347.	7.4	390
54	Candidate Genetic Modifiers for Breast and Ovarian Cancer Risk in BRCA1 and BRCA2 Mutation Carriers. Cancer Epidemiology Biomarkers and Prevention, 2015, 24, 308-316.	2.5	22

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55	DNA Glycosylases Involved in Base Excision Repair May Be Associated with Cancer Risk in BRCA1 and BRCA2 Mutation Carriers. PLoS Genetics, 2014, 10, e1004256.	3.5	47
56	Refined histopathological predictors of BRCA1 and BRCA2mutation status: a large-scale analysis of breast cancer characteristics from the BCAC, CIMBA, and ENIGMA consortia. Breast Cancer Research, 2014, 16, 3419.	5.0	97
57	Associations of common breast cancer susceptibility alleles with risk of breast cancer subtypes in BRCA1 and BRCA2 mutation carriers. Breast Cancer Research, 2014, 16, 3416.	5.0	57
58	Persistence of <scp>DNMT</scp>3A mutations at longâ€term remission in adult patients with <scp>AML</scp>. British Journal of Haematology, 2014, 167, 478-486.	2.5	113
59	Truncating Plakophilin-2 Mutations in Arrhythmogenic Cardiomyopathy Are Associated With Protein Haploinsufficiency in Both Myocardium and Epidermis. Circulation: Cardiovascular Genetics, 2014, 7, 230-240.	5.1	36
60	Multiple independent variants at the TERT locus are associated with telomere length and risks of breast and ovarian cancer. Nature Genetics, 2013, 45, 371-384.	21.4	493
61	Mutated Desmoglein-2 Proteins are Incorporated into Desmosomes and Exhibit Dominant-Negative Effects in Arrhythmogenic Right Ventricular Cardiomyopathy. Human Mutation, 2013, 34, 697-705.	2.5	30
62	Monodisperse and LPS-free Aggregatibacter actinomycetemcomitans leukotoxin: Interactions with human Î²2 integrins and erythrocytes. Biochimica Et Biophysica Acta - Proteins and Proteomics, 2013, 1834, 546-558.	2.3	30
63	Isolation and Characterization of Cutaneous Epithelial Stem Cells. Methods in Molecular Biology, 2013, 989, 61-69.	0.9	4
64	Severe Prenatal Renal Anomalies Associated with Mutations in HNF1B or PAX2 Genes. Clinical Journal of the American Society of Nephrology: CJASN, 2013, 8, 1179-1187.	4.5	87
65	Identification of a BRCA2-Specific Modifier Locus at 6p24 Related to Breast Cancer Risk. PLoS Genetics, 2013, 9, e1003173.	3.5	105
66	Tetraploidy in hydatidiform moles. Human Reproduction, 2013, 28, 2010-2020.	0.9	24
67	Genome-Wide Association Study in BRCA1 Mutation Carriers Identifies Novel Loci Associated with Breast and Ovarian Cancer Risk. PLoS Genetics, 2013, 9, e1003212.	3.5	244
68	A Nonsynonymous Polymorphism in IRS1 Modifies Risk of Developing Breast and Ovarian Cancers in BRCA1 and Ovarian Cancer in BRCA2 Mutation Carriers. Cancer Epidemiology Biomarkers and Prevention, 2012, 21, 1362-1370.	2.5	23
69	Phylloid Hypermelanosis in a Child with Psychomotor Delay, Cicatricial Alopecia, Hearing Loss and Polythelia. Acta Dermato-Venereologica, 2012, 92, 191-192.	1.3	7
70	Identification of epidermal progenitors for the Merkel cell lineage. Development (Cambridge), 2012, 139, 622-622.	2.5	1
71	Common Variants at the 19p13.1 and ZNF365 Loci Are Associated with ER Subtypes of Breast Cancer and Ovarian Cancer Risk in BRCA1 and BRCA2 Mutation Carriers. Cancer Epidemiology Biomarkers and Prevention, 2012, 21, 645-657.	2.5	47
72	Assessment of the Effect of 24-Hour Aldosterone Administration on Protein Abundance in Fluorescence-Sorted Mouse Distal Renal Tubules by Mass Spectrometry. Nephron Physiology, 2012, 121, p9-p15.	1.2	7

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73	Clinical and genetic findings in a large cohort of patients with ryanodine receptor 1 gene-associated myopathies. Human Mutation, 2012, 33, 981-988.	2.5	145
74	Characterization of BRCA1 and BRCA2 splicing variants: a collaborative report by ENIGMA consortium members. Breast Cancer Research and Treatment, 2012, 132, 1009-1023.	2.5	56
75	Common variants at 12p11, 12q24, 9p21, 9q31.2 and in ZNF365 are associated with breast cancer risk for BRCA1 and/or BRCA2mutation carriers. Breast Cancer Research, 2012, 14, R33.	5.0	78
76	Update of PAX2 mutations in renal coloboma syndrome and establishment of a locus-specific database. Human Mutation, 2012, 33, 457-466.	2.5	109
77	Ovarian cancer susceptibility alleles and risk of ovarian cancer inBRCA1andBRCA2mutation carriers. Human Mutation, 2012, 33, 690-702.	2.5	34
78	Identification of proteins regulated by 24â€hour aldosterone treatment in late distal convoluted tubules, connecting tubules and initial cortical collecting ducts. FASEB Journal, 2012, 26, 885.9.	0.5	0
79	Zinc Fixation for Flow Cytometry Analysis of Intracellular and Surface Epitopes, DNA Content, and Cell Proliferation. Current Protocols in Cytometry, 2011, 57, Unit 7.40.	3.7	3
80	Common breast cancer susceptibility alleles are associated with tumour subtypes in BRCA1 and BRCA2 mutation carriers: results from the Consortium of Investigators of Modifiers of BRCA1/2. Breast Cancer Research, 2011, 13, R110.	5.0	71
81	A BRCA2 mutation incorrectly mapped in the original BRCA2 reference sequence, is a common West Danish founder mutation disrupting mRNA splicing. Breast Cancer Research and Treatment, 2011, 128, 179-185.	2.5	4
82	Common alleles at 6q25.1 and 1p11.2 are associated with breast cancer risk for BRCA1 and BRCA2 mutation carriers. Human Molecular Genetics, 2011, 20, 3304-3321.	2.9	68
83	Genetic Variation at 9p22.2 and Ovarian Cancer Risk for BRCA1 and BRCA2 Mutation Carriers. Journal of the National Cancer Institute, 2011, 103, 105-116.	6.3	40
84	Mosaics and moles. European Journal of Human Genetics, 2011, 19, 1026-1031.	2.8	39
85	Mismatch repair defective breast cancer in the hereditary nonpolyposis colorectal cancer syndrome. Breast Cancer Research and Treatment, 2010, 120, 777-782.	2.5	39
86	Zinc fixation preserves flow cytometry scatter and fluorescence parameters and allows simultaneous analysis of DNA content and synthesis, and intracellular and surface epitopes. Cytometry Part A: the Journal of the International Society for Analytical Cytology, 2010, 77A, 798-804.	1.5	22
87	The load of short telomeres, estimated by a new method, Universal STELA, correlates with number of senescent cells. Aging Cell, 2010, 9, 383-397.	6.7	101
88	Common Breast Cancer Susceptibility Alleles and the Risk of Breast Cancer for BRCA1 and BRCA2 Mutation Carriers: Implications for Risk Prediction. Cancer Research, 2010, 70, 9742-9754.	0.9	169
89	Escherichia coli Î±-Hemolysin Triggers Shrinkage of Erythrocytes via KCa3.1 and TMEM16A Channels with Subsequent Phosphatidylserine Exposure. Journal of Biological Chemistry, 2010, 285, 15557-15565.	3.4	53
90	Identification of epidermal progenitors for the Merkel cell lineage. Development (Cambridge), 2010, 137, 3965-3971.	2.5	71

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91	p53 and PPP1R13L (alias iASPP or RAI) form a feedback loop to regulate genotoxic stress responses. Biochimica Et Biophysica Acta - General Subjects, 2010, 1800, 1231-1240.	2.4	14
92	Epsilon Haemoglobin Specific Antibodies with Applications in Noninvasive Prenatal Diagnosis. Journal of Biomedicine and Biotechnology, 2009, 2009, 1-8.	3.0	10
93	Enforced expression of PPP1R13L increases tumorigenesis and invasion through p53â€dependent and p53â€independent mechanisms. Molecular Carcinogenesis, 2009, 48, 832-842.	2.7	18
94	Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy resulting in stroke in an 11â€yearâ€old male. Developmental Medicine and Child Neurology, 2009, 51, 754-757.	2.1	26
95	An Important Role for Type III Interferon (IFN-Î»/IL-28) in TLR-Induced Antiviral Activity. Journal of Immunology, 2008, 180, 2474-2485.	0.8	387
96	A distinct population of clonogenic and multipotent murine follicular keratinocytes residing in the upper isthmus. Journal of Cell Science, 2008, 121, 609-617.	2.0	166
97	Enforced expression of PPP1R13L increases tumorigenesis and invasion through p53-dependent and p53-independent mechanisms.. Nature Precedings, 2008, , .	0.1	0
98	Binding between the Integrin Î±XÎ²2 (CD11c/CD18) and Heparin. Journal of Biological Chemistry, 2007, 282, 30869-30877.	3.4	43
99	IL-20 Gene Expression Is Induced by IL-1Î² through Mitogen-Activated Protein Kinase and NF-ÎºB-Dependent Mechanisms. Journal of Investigative Dermatology, 2007, 127, 1326-1336.	0.7	52
100	Expression of the RAI gene is conducive to apoptosis: Studies of induction and interference. Experimental Cell Research, 2007, 313, 2611-2621.	2.6	23
101	Mitogen- and Stress-Activated Protein Kinase 1 Is Activated in Lesional Psoriatic Epidermis and Regulates the Expression of Pro-Inflammatory Cytokines. Journal of Investigative Dermatology, 2006, 126, 1784-1791.	0.7	58
102	Heat-Shock Protein 70 Genes and Human Longevity: A View from Denmark. Annals of the New York Academy of Sciences, 2006, 1067, 301-308.	3.8	43
103	Protein Expression of TNF-Î± in Psoriatic Skin Is Regulated at a Posttranscriptional Level by MAPK-Activated Protein Kinase 2. Journal of Immunology, 2006, 176, 1431-1438.	0.8	130
104	Reduced heat shock response in human mononuclear cells during aging and its association with polymorphisms in HSP70 genes. Cell Stress and Chaperones, 2006, 11, 208.	2.9	66
105	Side population cells in human and mouse epidermis lack stem cell characteristics. Experimental Cell Research, 2004, 295, 79-90.	2.6	129
106	Functional testing of keratin 14 mutant proteins associated with the three major subtypes of epidermolysis bullosa simplex. Experimental Dermatology, 2003, 12, 472-479.	2.9	14
107	Epidermolysis bullosa simplex keratinocytes with extended lifespan established by ectopic expression of telomerase. Experimental Dermatology, 2003, 12, 71-77.	2.9	4
108	Platelet Derived Growth Factor (PDGF) Responsive Epidermis Formed from Human Keratinocytes Transduced with the PDGFÎ² Receptor Gene. Journal of Investigative Dermatology, 2003, 120, 742-749.	0.7	29

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109	Expression and Localization of Peroxisome Proliferator-Activated Receptors and Nuclear Factor Î¤B in Normal and Lesional Psoriatic Skin. Journal of Investigative Dermatology, 2003, 121, 1104-1117.	0.7	105
110	Manipulation of stem cell proliferation and lineage commitment:visualisation of label-retaining cells in wholemounts of mouse epidermis. Development (Cambridge), 2003, 130, 5241-5255.	2.5	382
111	Role of melanoma chondroitin sulphate proteoglycan in patterning stem cells in human interfollicular epidermis. Development (Cambridge), 2003, 130, 6049-6063.	2.5	129
112	RUNX/AML and C/EBP factors regulate CD11a integrin expression in myeloid cells through overlapping regulatory elements. Blood, 2003, 102, 3252-3261.	1.4	50
113	Production of Retroviral Vectors in Primary Human Keratinocytes after DNA-Mediated Gene Transfer Leads to Prolonged Gene Expression. Acta Dermato-Venereologica, 2003, 83, 83-87.	1.3	2
114	Functional requirement of aquaporin-5 in plasma membranes of sweat glands. Proceedings of the National Academy of Sciences of the United States of America, 2002, 99, 511-516.	7.1	194
115	Skin Genetically Engineered as a Bioreactor or a â€˜Metabolic Sinkâ€™. Cells Tissues Organs, 2002, 172, 96-104.	2.3	13
116	Physiological effects of human growth hormone produced after hydrodynamic gene transfer of a plasmid vector containing the human ubiquitin promotor. Journal of Molecular Medicine, 2002, 80, 665-670.	3.9	28
117	Recombinant expression of human mannan-binding lectin. International Immunopharmacology, 2001, 1, 677-687.	3.8	60
118	Cutaneous gene therapy â€“ an update. Histochemistry and Cell Biology, 2001, 115, 73-82.	1.7	17
119	Modulation of Keratinocyte Gene Expression and Differentiation by PPAR-Selective Ligands and Tetradecylthioacetic Acid. Journal of Investigative Dermatology, 2001, 116, 702-712.	0.7	213
120	Tail-Vein Injection of Mannan-Binding Lectin DNA Leads to High Expression Levels of Multimeric Protein in Liver. Molecular Therapy, 2001, 3, 867-874.	8.2	25
121	Immunolocalization of aquaporin-8 in rat kidney, gastrointestinal tract, testis, and airways. American Journal of Physiology - Renal Physiology, 2001, 281, F1047-F1057.	2.7	188
122	Transgene expression in human epidermal keratinocytes: cell cycle arrest of productively transfected cells. Experimental Dermatology, 2000, 9, 298-310.	2.9	17
123	Immunolocalization of AQP9 in Liver, Epididymis, Testis, Spleen, and Brain. Biochemical and Biophysical Research Communications, 2000, 276, 1118-1128.	2.1	296
124	The Î±2 and Î±5 integrin genes: identification of transcription factors that regulate promoter activity in epidermal keratinocytes. FEBS Letters, 2000, 474, 201-207.	2.8	39
125	Rapid Degradation of Short-chain Acyl-CoA Dehydrogenase Variants with Temperature-sensitive Folding Defects Occurs after Import into Mitochondria. Journal of Biological Chemistry, 1998, 273, 13065-13071.	3.4	48
126	Acute effects of vasopressin V₂-receptor antagonist on kidney AQP2 expression and subcellular distribution. American Journal of Physiology - Renal Physiology, 1998, 275, F285-F297.	2.7	33

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127	Influence of Lewis Î±1-3/4-L-Fucosyltransferase (FUT3) Gene Mutations on Enzyme Activity, Erythrocyte Phenotyping, and Circulating Tumor Marker Sialyl-Lewis a Levels. Journal of Biological Chemistry, 1996, 271, 32260-32268.	3.4	94
128	Comparison between medium-chain acyl-CoA dehydrogenase mutant proteins overexpressed in bacterial and mammalian cells. Human Mutation, 1995, 6, 226-231.	2.5	21
129	A nonsense mutation in the COL4A5 collagen gene in a family with X-linked juvenile Alport syndrome. Kidney International, 1995, 47, 327-332.	5.2	12
130	Gene Transfer into Cultured Human Epidermis and its Transplantation onto Immunodeficient Mice: An Experimental Model for Somatic Gene Therapy. Journal of Investigative Dermatology, 1994, 103, 391-394.	0.7	44
131	Correction of Steroid Sulfatase Deficiency by Gene Transfer into Basal Cells of Tissue-Cultured Epidermis from Patients with Recessive X-Linked Ichthyosis. Experimental Cell Research, 1993, 209, 392-397.	2.6	35
132	Expression of wild-type and mutant medium-chain acyl-CoA dehydrogenase (MCAD) cDNA in eucaryotic cells. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 1992, 1180, 65-72.	3.8	26