

Christophe Beroud

List of Publications by Year in descending order

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130
papers

12,751
citations

28736

57
h-index

29333

108
g-index

138
all docs

138
docs citations

138
times ranked

20608
citing authors

#	ARTICLE	IF	CITATIONS
1	Diagnostic interest of whole-body MRI in early- and late-onset LAMA2 muscular dystrophies: a large international cohort. <i>Journal of Neurology</i> , 2022, 269, 2414-2429.	1.8	5
2	The RDConnect Genome-Phenome Analysis Platform: Accelerating diagnosis, research, and gene discovery for rare diseases. <i>Human Mutation</i> , 2022, , .	1.1	18
3	Convergence of patient- and physician-reported outcomes in the French National Registry of Facioscapulohumeral Dystrophy. <i>Orphanet Journal of Rare Diseases</i> , 2022, 17, 96.	1.2	2
4	Highlighting the Dystonic Phenotype Related to <i>GNAO1</i> . <i>Movement Disorders</i> , 2022, 37, 1547-1554.	2.2	25
5	The lncRNA 44s2 Study Applicability to the Design of 45-55 Exon Skipping Therapeutic Strategy for DMD. <i>Biomedicines</i> , 2021, 9, 219.	1.4	4
6	Whole-body muscle MRI characteristics of LAMA2-related congenital muscular dystrophy children: An emerging pattern. <i>Neuromuscular Disorders</i> , 2021, 31, 814-823.	0.3	4
7	Standardisation of pathogenicity classification for somatic alterations in solid tumours and haematologic malignancies. <i>European Journal of Cancer</i> , 2021, 159, 1-15.	1.3	7
8	X-linked muscular dystrophy in a Labrador Retriever strain: phenotypic and molecular characterisation. <i>Skeletal Muscle</i> , 2020, 10, 23.	1.9	12
9	Pathogenic FBN1 Genetic Variation and Aortic Dissection in Patients With Marfan Syndrome. <i>Journal of the American College of Cardiology</i> , 2020, 75, 843-853.	1.2	38
10	The ELIXIR Human Copy Number Variations Community: building bioinformatics infrastructure for research. <i>F1000Research</i> , 2020, 9, 1229.	0.8	5
11	UMD-MEN1 Database: An Overview of the 370 MEN1 Variants Present in 1676 Patients From the French Population. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2019, 104, 753-764.	1.8	39
12	Whole Exome Sequencing Reveals a Large Genetic Heterogeneity and Revisits the Causes of Hypertrophic Cardiomyopathy. <i>Circulation Genomic and Precision Medicine</i> , 2019, 12, e002500.	1.6	9
13	Clinical Significance of Aortic Root Modification Associated With Bicuspid Aortic Valve in Marfan Syndrome. <i>Circulation: Cardiovascular Imaging</i> , 2019, 12, e008129.	1.3	15
14	Proposition of adjustments to the ACMG-CAMP framework for the interpretation of <i>MEN1</i> missense variants. <i>Human Mutation</i> , 2019, 40, 661-674.	1.1	21
15	Single-cell genetic analysis validates cytopathological identification of circulating cancer cells in patients with clear cell renal cell carcinoma. <i>Oncotarget</i> , 2018, 9, 20058-20074.	0.8	20
16	The French National Registry of patients with Facioscapulohumeral muscular dystrophy. <i>Orphanet Journal of Rare Diseases</i> , 2018, 13, 218.	1.2	11
17	Muscular MRI-based algorithm to differentiate inherited myopathies presenting with spinal rigidity. <i>European Radiology</i> , 2018, 28, 5293-5303.	2.3	28
18	Loss of Calmodulin- and Radial-Spoke-Associated Complex Protein CFAP251 Leads to Immotile Spermatozoa Lacking Mitochondria and Infertility in Men. <i>American Journal of Human Genetics</i> , 2018, 103, 413-420.	2.6	74

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19	VarAFT: a variant annotation and filtration system for human next generation sequencing data. Nucleic Acids Research, 2018, 46, W545-W553.	6.5	136
20	Clinical Outcomes in Duchenne Muscular Dystrophy: A Study of 5345 Patients from the TREAT-NMD DMD Global Database. Journal of Neuromuscular Diseases, 2017, 4, 293-306.	1.1	125
21	Exome Sequencing Identifies Two Variants of the Alkylglycerol Monooxygenase Gene as a Cause of Relapses in Visceral Leishmaniasis in Children, in Sudan. Journal of Infectious Diseases, 2017, 216, 22-28.	1.9	11
22	New advances in DPYD genotype and risk of severe toxicity under capecitabine. PLoS ONE, 2017, 12, e0175998.	1.1	82
23	Simple Sequence Mutations. , 2017, , 217-230.		0
24	UMDâ€Predictor: A Highâ€Throughput Sequencing Compliant System for Pathogenicity Prediction of any Human cDNA Substitution. Human Mutation, 2016, 37, 439-446.	1.1	104
25	Coverage analysis of lists of genes involved in heterogeneous genetic diseases following benchtop exome sequencing using the ion proton. Journal of Genetics, 2016, 95, 203-208.	0.4	3
26	How to Identify Pathogenic Mutations among All Those Variations: Variant Annotation and Filtration in the Genome Sequencing Era. Human Mutation, 2016, 37, 1272-1282.	1.1	28
27	Novel heterozygous mutation in <i>ANO3</i> responsible for craniocervical dystonia. Movement Disorders, 2016, 31, 1251-1252.	2.2	24
28	BRCA Share: A Collection of Clinical BRCA Gene Variants. Human Mutation, 2016, 37, 1318-1328.	1.1	57
29	Actionable Genes, Core Databases, and Locus-Specific Databases. Human Mutation, 2016, 37, 1299-1307.	1.1	6
30	WES/WGS Reporting of Mutations from Cardiovascular â€Actionableâ€Genes in Clinical Practice: A Key Role for UMD Knowledgebases in the Era of Big Databases. Human Mutation, 2016, 37, 1308-1317.	1.1	5
31	High-Throughput Sequencing in the Context of Human Genetic Diseases: Now and Tomorrow. Human Mutation, 2016, 37, 1247-1247.	1.1	0
32	Consideration surrounding incidental findings throughout multigene panel testing in cancer genetics. Clinical Genetics, 2016, 89, 267-268.	1.0	1
33	Improving molecular diagnosis of distal myopathies by targeted next-generation sequencing: Table 1. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, 340-342.	0.9	20
34	A mutation in the Gardos channel is associated with hereditary xerocytosis. Blood, 2015, 126, 1273-1280.	0.6	97
35	Laminin Î±2 Deficiency-Related Muscular Dystrophy Mimicking Emery-Dreifuss and Collagen VI related Diseases. Journal of Neuromuscular Diseases, 2015, 2, 229-240.	1.1	30
36	Comparing targeted exome and whole exome approaches for genetic diagnosis of neuromuscular disorders. Applied & Translational Genomics, 2015, 7, 26-31.	2.1	18

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37	High Prevalence of PRPH2 in Autosomal Dominant Retinitis Pigmentosa in France and Characterization of Biochemical and Clinical Features. <i>American Journal of Ophthalmology</i> , 2015, 159, 302-314.	1.7	29
38	Rare inherited disorders with renal involvement—approach to the patient. <i>Kidney International</i> , 2015, 87, 901-908.	2.6	18
39	The TREAT-NMD DMD Global Database: Analysis of More than 7,000 Duchenne Muscular Dystrophy Mutations. <i>Human Mutation</i> , 2015, 36, 395-402.	1.1	507
40	Genomic variations integrated database for <i>MUTYH</i> -associated adenomatous polyposis. <i>Journal of Medical Genetics</i> , 2015, 52, 25-27.	1.5	2
41	Identification of Splicing Defects Caused by Mutations in the Dysferlin Gene. <i>Human Mutation</i> , 2014, 35, 1532-1541.	1.1	22
42	Clinical heterogeneity and a high proportion of novel mutations in a Chinese cohort of patients with dysferlinopathy. <i>Neurology India</i> , 2014, 62, 635.	0.2	27
43	The UMD-APC Database, a Model of Nation-Wide Knowledge Base: Update with Data from 3,581 Variations. <i>Human Mutation</i> , 2014, 35, 532-536.	1.1	15
44	Mapping the differences in care for 5,000 Spinal Muscular Atrophy patients, a survey of 24 national registries in North America, Australasia and Europe. <i>Journal of Neurology</i> , 2014, 261, 152-163.	1.8	76
45	RD-Connect: An Integrated Platform Connecting Databases, Registries, Biobanks and Clinical Bioinformatics for Rare Disease Research. <i>Journal of General Internal Medicine</i> , 2014, 29, 780-787.	1.3	159
46	Diagnostic approach to the congenital muscular dystrophies. <i>Neuromuscular Disorders</i> , 2014, 24, 289-311.	0.3	275
47	Dispelling myths about rare disease registry system development. <i>Source Code for Biology and Medicine</i> , 2013, 8, 21.	1.7	49
48	UMD-MLH1/MSH2/MSH6 databases: description and analysis of genetic variations in French Lynch syndrome families. <i>Database: the Journal of Biological Databases and Curation</i> , 2013, 2013, bat036-bat036.	1.4	15
49	The TREAT-NMD Duchenne Muscular Dystrophy Registries: Conception, Design, and Utilization by Industry and Academia. <i>Human Mutation</i> , 2013, 34, 1449-1457.	1.1	94
50	Description and analysis of genetic variants in French hereditary breast and ovarian cancer families recorded in the UMD-BRCA1/BRCA2 databases. <i>Nucleic Acids Research</i> , 2012, 40, D992-D1002.	6.5	84
51	Aortic Event Rate in the Marfan Population. <i>Circulation</i> , 2012, 125, 226-232.	1.6	165
52	VarioML framework for comprehensive variation data representation and exchange. <i>BMC Bioinformatics</i> , 2012, 13, 254.	1.2	17
53	Surgical management of patients with Marfan syndrome: Evolution throughout the years. <i>Archives of Cardiovascular Diseases</i> , 2012, 105, 84-90.	0.7	5
54	In-Frame Mutations in Exon 1 of SKI Cause Dominant Shprintzen-Goldberg Syndrome. <i>American Journal of Human Genetics</i> , 2012, 91, 950-957.	2.6	95

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55	Bioinformatics and Mutations Leading to Exon Skipping. <i>Methods in Molecular Biology</i> , 2012, 867, 17-35.	0.4	12
56	Mutation spectrum in the large GTPase dynamin 2, and genotype-phenotype correlation in autosomal dominant centronuclear myopathy. <i>Human Mutation</i> , 2012, 33, 949-959.	1.1	115
57	Motor and respiratory heterogeneity in Duchenne patients: Implication for clinical trials. <i>European Journal of Paediatric Neurology</i> , 2012, 16, 149-160.	0.7	112
58	The new Ghent criteria for Marfan syndrome: what do they change?. <i>Clinical Genetics</i> , 2012, 81, 433-442.	1.0	90
59	UMD-DYSF, a novel locus specific database for the compilation and interactive analysis of mutations in the dysferlin gene. <i>Human Mutation</i> , 2012, 33, E2317-E2331.	1.1	35
60	Hydrophobic pulses predict transmembrane helix irregularities and channel transmembrane units. <i>BMC Bioinformatics</i> , 2011, 12, 135.	1.2	4
61	Polymorphisms of MAMLD1 gene in hypospadias. <i>Journal of Pediatric Urology</i> , 2011, 7, 585-591.	0.6	33
62	DYT6 dystonia: Review of the literature and creation of the UMD locus-specific database (LSDB) for mutations in the THAP1 gene. <i>Human Mutation</i> , 2011, 32, 1213-1224.	1.1	126
63	Missense mutations of conserved glycine residues in fibrillin-1 highlight a potential subtype of cb-EGF-like domains. <i>Human Mutation</i> , 2010, 31, E1021-E1042.	1.1	9
64	MUT-TP53 2.0: a novel versatile matrix for statistical analysis of TP53 mutations in human cancer. <i>Human Mutation</i> , 2010, 31, 1020-1025.	1.1	26
65	UMD-CFTR: A database dedicated to CF and CFTR-related disorders. <i>Human Mutation</i> , 2010, 31, 1011-1019.	1.1	23
66	Cardiovascular manifestations in men and women carrying a FBN1 mutation. <i>European Heart Journal</i> , 2010, 31, 2223-2229.	1.0	133
67	Consensus Statement on Standard of Care for Congenital Muscular Dystrophies. <i>Journal of Child Neurology</i> , 2010, 25, 1559-1581.	0.7	200
68	Locus Reference Genomic sequences: an improved basis for describing human DNA variants. <i>Genome Medicine</i> , 2010, 2, 24.	3.6	100
69	Comparison of Clinical Presentations and Outcomes Between Patients With <i>TGFBR2</i> and <i>FBN1</i> Mutations in Marfan Syndrome and Related Disorders. <i>Circulation</i> , 2009, 120, 2541-2549.	1.6	203
70	Clinical and Molecular Study of 320 Children With Marfan Syndrome and Related Type I Fibrillinopathies in a Series of 1009 Proband With Pathogenic <i>FBN1</i> Mutations. <i>Pediatrics</i> , 2009, 123, 391-398.	1.0	146
71	Pathogenic <i>FBN1</i> mutations in 146 adults not meeting clinical diagnostic criteria for Marfan syndrome: Further delineation of type 1 fibrillinopathies and focus on patients with an isolated major criterion. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 854-860.	0.7	40
72	The <i>FBN2</i> gene: new mutations, locus-specific database (Universal Mutation Database <i>FBN2</i>), and genotype-phenotype correlations. <i>Human Mutation</i> , 2009, 30, 181-190.	1.1	49

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73	Analysis of the <i>DYSF</i> mutational spectrum in a large cohort of patients. <i>Human Mutation</i> , 2009, 30, E345-E375.	1.1	97
74	UMD-predictor, a new prediction tool for nucleotide substitution pathogenicity-application to four genes: <i>FBN1</i> , <i>FBN2</i> , <i>TGFBR1</i> , and <i>TGFBR2</i> . <i>Human Mutation</i> , 2009, 30, 952-959.	1.1	80
75	Genotype-phenotype analysis in 2,405 patients with a dystrophinopathy using the UMD-DMD database: a model of nationwide knowledgebase. <i>Human Mutation</i> , 2009, 30, 934-945.	1.1	309
76	Clinical and mutation-type analysis from an international series of 198 probands with a pathogenic <i>FBN1</i> exons 24-32 mutation. <i>European Journal of Human Genetics</i> , 2009, 17, 491-501.	1.4	66
77	Human Splicing Finder: an online bioinformatics tool to predict splicing signals. <i>Nucleic Acids Research</i> , 2009, 37, e67-e67.	6.5	2,206
78	A new locus-specific database (LSDB) for mutations in the <i>TGFBR2</i> gene: UMD- <i>TGFBR2</i> . <i>Human Mutation</i> , 2008, 29, 33-38.	1.1	27
79	UMD-USHbases: a comprehensive set of databases to record and analyse pathogenic mutations and unclassified variants in seven Usher syndrome causing genes. <i>Human Mutation</i> , 2008, 29, E76-E87.	1.1	38
80	Contribution of molecular analyses in diagnosing Marfan syndrome and type I fibrillinopathies: an international study of 1009 probands. <i>Journal of Medical Genetics</i> , 2008, 45, 384-390.	1.5	83
81	Effect of Mutation Type and Location on Clinical Outcome in 1,013 Probands with Marfan Syndrome or Related Phenotypes and <i>FBN1</i> Mutations: An International Study. <i>American Journal of Human Genetics</i> , 2007, 81, 454-466.	2.6	485
82	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. <i>Human Mutation</i> , 2007, 28, 183-195.	1.1	107
83	Multixon skipping leading to an artificial DMD protein lacking amino acids from exons 45 through 55 could rescue up to 63% of patients with Duchenne muscular dystrophy. <i>Human Mutation</i> , 2007, 28, 196-202.	1.1	178
84	Large genomic rearrangements in the <i>CFTR</i> gene contribute to CBAVD. <i>BMC Medical Genetics</i> , 2007, 8, 22.	2.1	42
85	Trichloroethylene exposure and somatic mutations of the <i>VHL</i> gene in patients with Renal Cell Carcinoma. <i>Journal of Occupational Medicine and Toxicology</i> , 2007, 2, 13.	0.9	23
86	Negative genetic neonatal screening for cystic fibrosis caused by compound heterozygosity for two large <i>CFTR</i> rearrangements. <i>Clinical Genetics</i> , 2007, 72, 374-377.	1.0	11
87	Transcriptional explorations of <i>CAPN3</i> identify novel splicing mutations, a large-sized genomic deletion and evidence for messenger RNA decay. <i>Clinical Genetics</i> , 2007, 72, 582-592.	1.0	25
88	Locus-specific mutation databases: pitfalls and good practice based on the p53 experience. <i>Nature Reviews Cancer</i> , 2006, 6, 83-90.	12.8	134
89	The UMD TP53 database and website: update and revisions. <i>Human Mutation</i> , 2006, 27, 14-20.	1.1	125
90	Meta-analysis of the p53 Mutation Database for Mutant p53 Biological Activity Reveals a Methodologic Bias in Mutation Detection. <i>Clinical Cancer Research</i> , 2006, 12, 62-69.	3.2	67

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91	UMD (Universal Mutation Database): 2005 update. <i>Human Mutation</i> , 2005, 26, 184-191.	1.1	101
92	Dysferlin mutations in LGMD2B, Miyoshi myopathy, and atypical dysferlinopathies. <i>Human Mutation</i> , 2005, 26, 165-165.	1.1	104
93	Introducing the online version of the gene table for neuromuscular disease (nuclear genes only). <i>Neuromuscular Disorders</i> , 2005, 15, 88.	0.3	2
94	Impact of cytomorphological detection of circulating tumor cells in patients with liver cancer. <i>Hepatology</i> , 2004, 39, 792-797.	3.6	184
95	Genotype-phenotype correlation in von Hippel-Lindau families with renal lesions. <i>Human Mutation</i> , 2004, 24, 215-224.	1.1	81
96	High incidence of renal tumours in vitamins A and E synthesis workers: A new cause of occupational cancer?. <i>International Journal of Cancer</i> , 2004, 108, 942-944.	2.3	14
97	Dystrophinopathy caused by mid-intronic substitutions activating cryptic exons in the DMD gene. <i>Neuromuscular Disorders</i> , 2004, 14, 10-18.	0.3	46
98	The UMD-p53 database: New mutations and analysis tools. <i>Human Mutation</i> , 2003, 21, 176-181.	1.1	136
99	Significance of TP53 mutations in human cancer: A critical analysis of mutations at CpG dinucleotides. <i>Human Mutation</i> , 2003, 21, 192-200.	1.1	94
100	Update of the UMD-FBN1mutation database and creation of anFBN1polymorphism database. <i>Human Mutation</i> , 2003, 22, 199-208.	1.1	299
101	Prenatal diagnosis of spinal muscular atrophy by genetic analysis of circulating fetal cells. <i>Lancet</i> , The, 2003, 361, 1013-1014.	6.3	45
102	108th ENMC International Workshop, 3rd Workshop of the MYO-CLUSTER project: EUROMEN, 7th International Emery-Dreifuss Muscular Dystrophy (EDMD) Workshop, 13-15 September 2002, Naarden, The Netherlands. <i>Neuromuscular Disorders</i> , 2003, 13, 508-515.	0.3	78
103	Low mitochondrial respiratory chain content correlates with tumor aggressiveness in renal cell carcinoma. <i>Carcinogenesis</i> , 2002, 23, 759-768.	1.3	298
104	Enrichment, Immunomorphological, and Genetic Characterization of Fetal Cells Circulating in Maternal Blood. <i>American Journal of Pathology</i> , 2002, 160, 51-58.	1.9	78
105	Diagnosis of pheochromocytoma and laparoscopic adrenalectomy in two anephric patients with von hippel-lindau disease. <i>American Journal of Kidney Diseases</i> , 2002, 39, e6.1-e6.4.	2.1	4
106	Germline mutation profile ofMEN1in multiple endocrine neoplasia type 1: search for correlation between phenotype and the functional domains of the MEN1 protein. <i>Human Mutation</i> , 2002, 20, 35-47.	1.1	137
107	The UMD-LDLR database: additions to the software and 490 new entries to the database. <i>Human Mutation</i> , 2002, 20, 81-87.	1.1	105
108	Retinal hemangioblastoma in von Hippel-Lindau disease: a clinical and molecular study. <i>Investigative Ophthalmology and Visual Science</i> , 2002, 43, 3067-74.	3.3	91

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109	Association of GSTT1 non-null and NAT1 slow/rapid genotypes with von Hippel-Lindau tumour suppressor gene transversions in sporadic renal cell carcinoma. <i>Pharmacogenetics and Genomics</i> , 2001, 11, 521-535.	5.7	26
110	Assessing TP53 status in human tumours to evaluate clinical outcome. <i>Nature Reviews Cancer</i> , 2001, 1, 233-239.	12.8	587
111	p53 Website and analysis of p53 gene mutations in human cancer: Forging a link between epidemiology and carcinogenesis. , 2000, 15, 105-113.		231
112	UMD (Universal Mutation Database): A generic software to build and analyze locus-specific databases. <i>Human Mutation</i> , 2000, 15, 86-94.	1.1	184
113	Central nervous system hemangioblastomas, endolymphatic sac tumors, and von Hippel-Lindau disease. <i>Neurosurgical Review</i> , 2000, 23, 1-22.	1.2	147
114	Mutations of the VHL gene in sporadic renal cell carcinoma: Definition of a risk factor for VHL patients to develop an RCC. , 1999, 13, 464-475.		126
115	APC gene: database of germline and somatic mutations in human tumors and cell lines. <i>Nucleic Acids Research</i> , 1998, 26, 269-270.	6.5	119
116	Marfan Database (third edition): new mutations and new routines for the software. <i>Nucleic Acids Research</i> , 1998, 26, 229-233.	6.5	97
117	LDLR Database (second edition): new additions to the database and the software, and results of the first molecular analysis. <i>Nucleic Acids Research</i> , 1998, 26, 248-252.	6.5	77
118	p53 gene mutation: software and database. <i>Nucleic Acids Research</i> , 1998, 26, 200-204.	6.5	132
119	Software and database for the analysis of mutations in the VHL gene. <i>Nucleic Acids Research</i> , 1998, 26, 256-258.	6.5	119
120	Software and database for the analysis of mutations in the human WT1 gene. <i>Nucleic Acids Research</i> , 1998, 26, 271-274.	6.5	40
121	Marfan Database (second edition): software and database for the analysis of mutations in the human FBN1 gene. <i>Nucleic Acids Research</i> , 1997, 25, 147-150.	6.5	32
122	p53 and APC gene mutations: software and databases. <i>Nucleic Acids Research</i> , 1997, 25, 138-138.	6.5	17
123	Software and database for the analysis of mutations in the human LDL receptor gene. <i>Nucleic Acids Research</i> , 1997, 25, 172-180.	6.5	50
124	APC gene: database of germline and somatic mutations in human tumors and cell lines. <i>Nucleic Acids Research</i> , 1996, 24, 121-124.	6.5	286
125	Report of the Sixth International Workshop on Human Chromosome 3 Mapping 1995. <i>Cytogenetic and Genome Research</i> , 1996, 72, 255-270.	0.6	17
126	Correlations of allelic imbalance of chromosome 14 with adverse prognostic parameters in 148 renal cell carcinomas. , 1996, 17, 215-224.		41

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127	Software and database for the analysis of mutations in the human FBN1 gene. Nucleic Acids Research, 1996, 24, 137-140.	6.5	41
128	p53 gene mutation: software and database. Nucleic Acids Research, 1996, 24, 147-150.	6.5	82
129	Correlations of allelic imbalance of chromosome 14 with adverse prognostic parameters in 148 renal cell carcinomas. , 1996, 17, 215.		2
130	Constitutional and somatic deletions of two different regions of maternal chromosome 11 in Wilms tumor. Genomics, 1990, 7, 434-438.	1.3	25