Santiago Rodriguez de Cordoba

List of Publications by Year
in descending order

202 papers	24,784 citations	11651 70 h-index	7348 152 g-index
214 all docs	214 docs citations	214 times ranked	30560 citing authors

#	Article	IF	Citations
1	Longitudinal change in proteinuria and kidney outcomes in C3 glomerulopathy. Nephrology Dialysis Transplantation, 2022, 37, 1270-1280.	0.7	13
2	The crystal structure of iC3b-CR3 Î±I reveals a modular recognition of the main opsonin iC3b by the CR3 integrin receptor. Nature Communications, 2022, 13, 1955.	12.8	18
3	The Hidden Side of Complement Regulator C4BP: Dissection and Evaluation of Its Immunomodulatory Activity. Frontiers in Immunology, 2022, 13, 883743.	4.8	5
4	Factor Hâ€“Related Protein 1 Drives Disease Susceptibility and Prognosis in C3 Glomerulopathy. Journal of the American Society of Nephrology: JASN, 2022, 33, 1137-1153.	6.1	12
5	Characteristics, management and outcomes of atypical haemolytic uraemic syndrome in kidney transplant patients: a retrospective national study. CKJ: Clinical Kidney Journal, 2021, 14, 1173-1180.	2.9	12
6	Is the atypical hemolytic uremic syndrome risk polymorphism in Membrane Cofactor Protein MCPggaac relevant in kidney transplantation? A case report. Pediatric Transplantation, 2021, 25, e13903.	1.0	4
7	Detection of Genetic Rearrangements in the Regulators of Complement Activation RCA Cluster by High-Throughput Sequencing and MLPA. Methods in Molecular Biology, 2021, 2227, 159-178.	0.9	7
8	Molecular bases for the association of FHR-1 with atypical hemolytic uremic syndrome and other diseases. Blood, 2021, 137, 3484-3494.	1.4	17
9	Functional characterization of 105 factor H variants associated with aHUS: lessons for variant classification. Blood, 2021, 138, 2185-2201.	1.4	29
10	Complement Factor D (adipsin) Levels Are Elevated in Acquired Partial Lipodystrophy (Barraquerâ€“Simons syndrome). International Journal of Molecular Sciences, 2021, 22, 6608.	4.1	7
11	Case Report: Combined Liver-Kidney Transplantation to Correct a Mutation in Complement Factor B in an Atypical Hemolytic Uremic Syndrome Patient. Frontiers in Immunology, 2021, 12, 751093.	4.8	3
12	Gain-of-Function Mutations R249C and S250C in Complement C2 Protein Increase C3 Deposition in the Presence of C-Reactive Protein. Frontiers in Immunology, 2021, 12, 724361.	4.8	8
13	Noncanonical immunomodulatory activity of complement regulator C4BP(Î²-) limits the development of lupus nephritis. Kidney International, 2020, 97, 551-566.	5.2	11
14	The Relevance of the MCP Risk Polymorphism to the Outcome of aHUS Associated With C3 Mutations. A Case Report. Frontiers in Immunology, 2020, 11, 1348.	4.8	4
15	Mycophenolate Mofetil in C3 Glomerulopathy and Pathogenic Drivers of the Disease. Clinical Journal of the American Society of Nephrology: CJASN, 2020, 15, 1287-1298.	4.5	36
16	Familial risk of developing atypical hemolytic-uremic syndrome. Blood, 2020, 136, 1558-1561.	1.4	18
17	Gain-of-function mutation in complement C2 protein identified in a patient with aHUS. Journal of Allergy and Clinical Immunology, 2020, 146, 916-919.e11.	2.9	11
18	Blocking Complement Factor B Activation Reduces Renal Injury and Inflammation in a Rat Brain Death Model. Frontiers in Immunology, 2019, 10, 2528.	4.8	7

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19	Preface. Molecular Immunology, 2019, 114, 312-313.	2.2	0
20	C3 glomerulopathy â€" understanding a rare complement-driven renal disease. Nature Reviews Nephrology, 2019, 15, 129-143.	9.6	223
21	Severe and malignant hypertension are common in primary atypical hemolytic uremic syndrome. Kidney International, 2019, 96, 995-1004.	5.2	52
22	Kidney, hypertension and complement activation. In search of new therapeutic targets. Nefrologia, 2019, 39, 111-114.	0.4	0
23	Secondary atypical hemolytic uremic syndromes in the era of complement blockade. Kidney International, 2019, 95, 1298-1300.	5.2	12
24	The Antimicrobials Anacardic Acid and Curcumin Are Not-Competitive Inhibitors of Gram-Positive Bacterial Pathogenic Glyceraldehyde-3-Phosphate Dehydrogenase by a Mechanism Unrelated to Human C5a Anaphylatoxin Binding. Frontiers in Microbiology, 2019, 10, 326.	3.5	10
25	RiÃ±Ã³n, hipertensiÃ³n y activaciÃ³n del complemento. En bÃºsqueda de nuevas dianas terapÃ©uticas. Nefrologia, 2019, 39, 111-114.	0.4	0
26	Statistical Validation of Rare Complement Variants Provides Insights into the Molecular Basis of Atypical Hemolytic Uremic Syndrome and C3 Glomerulopathy. Journal of Immunology, 2018, 200, 2464-2478.	0.8	130
27	Human plasma C3 is essential for the development of memory B, but not T, lymphocytes. Journal of Allergy and Clinical Immunology, 2018, 141, 1151-1154.e14.	2.9	26
28	Factor H Competitor Generated by Gene Conversion Events Associates with Atypical Hemolytic Uremic Syndrome. Journal of the American Society of Nephrology: JASN, 2018, 29, 240-249.	6.1	34
29	Complete functional characterization of disease-associated genetic variants in the complement factor H gene. Kidney International, 2018, 93, 470-481.	5.2	45
30	A retrospective study of pregnancy-associated atypical hemolytic uremic syndrome. Kidney International, 2018, 93, 450-459.	5.2	100
31	Absence of CD59 in Guinea Pigs: Analysis of the Cavia porcellus Genome Suggests the Evolution of a CD59 Pseudogene. Journal of Immunology, 2018, 200, 327-335.	0.8	4
32	How novel structures inform understanding of complement function. Seminars in Immunopathology, 2018, 40, 3-14.	6.1	6
33	InvestigaciÃ³n multidisciplinar y traslacional en enfermedades raras. Arbor, 2018, 194, 468.	0.3	0
34	Eculizumab Modifies Outcomes in Adults with Atypical Hemolytic Uremic Syndrome with Acute Kidney Injury. American Journal of Nephrology, 2018, 48, 225-233.	3.1	21
35	High Complement Factor H-Related (FHR)-3 Levels Are Associated With the Atypical Hemolytic-Uremic Syndrome-Risk Allele CFHR3*B. Frontiers in Immunology, 2018, 9, 848.	4.8	26
36	Common and rare genetic variants of complement components in human disease. Molecular Immunology, 2018, 102, 42-57.	2.2	18

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37	Functional and structural characterization of four mouse monoclonal antibodies to complement C3 with potential therapeutic and diagnostic applications. European Journal of Immunology, 2017, 47, 504-515.	2.9	5
38	Elevated factor Hâ€“related protein 1 and factor H pathogenic variants decrease complement regulation inÂIgA nephropathy. Kidney International, 2017, 92, 953-963.	5.2	87
39	FHR-1 Binds to C-Reactive Protein and Enhances Rather than Inhibits Complement Activation. Journal of Immunology, 2017, 199, 292-303.	0.8	43
40	Abnormal glycogen chain length pattern, not hyperphosphorylation, is critical in Lafora disease. EMBO Molecular Medicine, 2017, 9, 906-917.	6.9	59
41	Ionic tethering contributes to the conformational stability and function of complement C3b. Molecular Immunology, 2017, 85, 137-147.	2.2	5
42	Eculizumab in secondary atypical haemolytic uraemic syndrome. Nephrology Dialysis Transplantation, 2017, 32, 466-474.	0.7	121
43	Extravascular hemolysis and complement consumption in Paroxysmal Nocturnal Hemoglobinuria patients undergoing eculizumab treatment. Immunobiology, 2017, 222, 363-371.	1.9	13
44	Crystal Structure of Glyceraldehyde-3-Phosphate Dehydrogenase from the Gram-Positive Bacterial Pathogen A. vaginae, an Immunoevasive Factor that Interacts with the Human C5a Anaphylatoxin. Frontiers in Microbiology, 2017, 8, 541.	3.5	24
45	The Complement Inhibitor Factor H Generates an Anti-Inflammatory and Tolerogenic State in Monocyte-Derived Dendritic Cells. Journal of Immunology, 2016, 196, 4274-4290.	0.8	54
46	Testing the Activity of Complement Convertases in Serum/Plasma for Diagnosis of C4NeF-Mediated C3 Glomerulonephritis. Journal of Clinical Immunology, 2016, 36, 517-527.	3.8	26
47	Complement genetics and susceptibility to inflammatory disease. Lessons from genotypeâ€“phenotype correlations. Immunobiology, 2016, 221, 709-714.	1.9	25
48	Guidelines for the use and interpretation of assays for monitoring autophagy (3rd edition). Autophagy, 2016, 12, 1-222.	9.1	4,701
49	Molecular Basis of Factor H R1210C Association with Ocular and Renal Diseases. Journal of the American Society of Nephrology: JASN, 2016, 27, 1305-1311.	6.1	29
50	Atypical hemolytic uraemic syndrome. Medicina ClÃ¬nica (English Edition), 2015, 145, 438-445.	0.2	2
51	Structural insights on complement activation. FEBS Journal, 2015, 282, 3883-3891.	4.7	22
52	Factor H-related proteins determine complement-activating surfaces. Trends in Immunology, 2015, 36, 374-384.	6.8	130
53	Eculizumab in pregnancy-associated atypical hemolytic uremic syndrome: insights for optimizing management. Journal of Nephrology, 2015, 28, 641-645.	2.0	29
54	An update for atypical haemolytic uraemic syndrome: Diagnosis and treatment. A consensus document. Nefrologia, 2015, 35, 421-447.	0.4	77

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55	Effectiveness of mycophenolate mofetil in C3 glomerulonephritis. Kidney International, 2015, 88, 1153-1160.	5.2	130
56	Complement factor H, FHR-3 and FHR-1 variants associate in an extended haplotype conferring increased risk of atypical hemolytic uremic syndrome. Molecular Immunology, 2015, 67, 276-286.	2.2	49
57	The role of complement in C3 glomerulopathy. Molecular Immunology, 2015, 67, 21-30.	2.2	78
58	Case report: lupus nephritis with autoantibodies to complement alternative pathway proteins and C3 gene mutation. BMC Nephrology, 2015, 16, 40.	1.8	18
59	Atypical aHUS: State of the art. Molecular Immunology, 2015, 67, 31-42.	2.2	236
60	The molecular and structural bases for the association of complement C3 mutations with atypical hemolytic uremic syndrome. Molecular Immunology, 2015, 66, 263-273.	2.2	47
61	ActualizaciÃ³n en sÃndrome hemolÃtico urÃ©mico atÃpico: diagnÃ³stico y tratamiento. Documento de consenso. Nefrologia, 2015, 35, 421-447.	0.4	188
62	A Novel Atypical Hemolytic Uremic Syndromeâ€“Associated Hybrid CFHR1/CFH Gene Encoding a Fusion Protein That Antagonizes Factor Hâ€“Dependent Complement Regulation. Journal of the American Society of Nephrology: JASN, 2015, 26, 209-219.	6.1	89
63	Pleiotropic Effects of Cell Wall Amidase LytA on Streptococcus pneumoniae Sensitivity to the Host Immune Response. Infection and Immunity, 2015, 83, 591-603.	2.2	47
64	A Novel Antibody against Human Factor B that Blocks Formation of the C3bB Proconvertase and Inhibits Complement Activation in Disease Models. Journal of Immunology, 2014, 193, 5567-5575.	0.8	14
65	Complement Mutations in Diacylglycerol Kinase-Îµâ€“Associated Atypical Hemolytic Uremic Syndrome. Clinical Journal of the American Society of Nephrology: CJASN, 2014, 9, 1611-1619.	4.5	61
66	Eculizumab long-term therapy for pediatric renal transplant in aHUS with CFH/CFHR1 hybrid gene. Pediatric Nephrology, 2014, 29, 149-153.	1.7	34
67	Genetics of Atypical Hemolytic Uremic Syndrome (aHUS). Seminars in Thrombosis and Hemostasis, 2014, 40, 422-430.	2.7	122
68	Interaction of Shiga toxin 2 with complement regulators of the factor H protein family. Molecular Immunology, 2014, 58, 77-84.	2.2	53
69	An ELISA assay with two monoclonal antibodies allows the estimation of free factor H and identifies patients with acquired deficiency of this complement regulator. Molecular Immunology, 2014, 58, 194-200.	2.2	20
70	The phosphatase activity of laforin is dispensable to rescue Epm2aâ¯'/â¯' mice from Lafora disease. Brain, 2014, 137, 806-818.	7.6	38
71	A Humanized Antibody That Regulates the Alternative Pathway Convertase: Potential for Therapy of Renal Disease Associated with Nephritic Factors. Journal of Immunology, 2014, 192, 4844-4851.	0.8	29
72	Eculizumab in dense-deposit disease after renal transplantation. Pediatric Nephrology, 2014, 29, 2055-2059.	1.7	26

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73	Structural basis for the stabilization of the complement alternative pathway C3 convertase by properdin. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 13504-13509.	7.1	86
74	The Î±7Î²0 Isoform of the Complement Regulator C4b-Binding Protein Induces a Semimature, Anti-Inflammatory State in Dendritic Cells. Journal of Immunology, 2013, 190, 2857-2872.	0.8	33
75	Intracellular Complement Activation Sustains T Cell Homeostasis and Mediates Effector Differentiation. Immunity, 2013, 39, 1143-1157.	14.3	444
76	Combined Complement Gene Mutations in Atypical Hemolytic Uremic Syndrome Influence Clinical Phenotype. Journal of the American Society of Nephrology: JASN, 2013, 24, 475-486.	6.1	308
77	C3 glomerulopathy: consensus report. Kidney International, 2013, 84, 1079-1089.	5.2	505
78	Anti-C5 as Prophylactic Therapy in Atypical Hemolytic Uremic Syndrome in Living-Related Kidney Transplantation. Transplantation, 2013, 96, e26-e29.	1.0	14
79	C3 glomerulopathyâ€“associated CFHR1 mutation alters FHR oligomerization and complement regulation. Journal of Clinical Investigation, 2013, 123, 2434-2446.	8.2	176
80	An update for atypical haemolytic uraemic syndrome: diagnosis and treatment. A consensus document. Nefrologia, 2013, 33, 27-45.	0.4	90
81	Complement factor H variants I890 and L1007 while commonly associated with atypical hemolytic uremic syndrome are polymorphisms with no functional significance. Kidney International, 2012, 81, 56-63.	5.2	34
82	Antibody directs properdin-dependent activation of the complement alternative pathway in a mouse model of abdominal aortic aneurysm. Proceedings of the National Academy of Sciences of the United States of America, 2012, 109, E415-22.	7.1	65
83	Relevance of Complement Factor Hâ€“Related 1 (CFHR1) Genotypes in Age-Related Macular Degeneration. , 2012, 53, 1087.		40
84	Anti-factor H antibody affecting factor H cofactor activity in a patient with dense deposit disease. CKJ: Clinical Kidney Journal, 2012, 5, 133-136.	2.9	20
85	Lafora bodies and neurological defects in malin-deficient mice correlate with impaired autophagy. Human Molecular Genetics, 2012, 21, 1521-1533.	2.9	131
86	Laforin and Malin Deletions in Mice Produce Similar Neurologic Impairments. Journal of Neuropathology and Experimental Neurology, 2012, 71, 413-421.	1.7	43
87	Complement dysregulation and disease: From genes and proteins to diagnostics and drugs. Immunobiology, 2012, 217, 1034-1046.	1.9	109
88	Atypical Hemolytic Uremic Syndrome-Associated Variants and Autoantibodies Impair Binding of Factor H and Factor H-Related Protein 1 to Pentraxin 3. Journal of Immunology, 2012, 189, 1858-1867.	0.8	62
89	Sensitive and specific assays for C3 nephritic factors clarify mechanisms underlying complement dysregulation. Kidney International, 2012, 82, 1084-1092.	5.2	93
90	Guidelines for the use and interpretation of assays for monitoring autophagy. Autophagy, 2012, 8, 445-544.	9.1	3,122

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91	Malin knockout mice support a primary role of autophagy in the pathogenesis of Lafora disease. Autophagy, 2012, 8, 701-703.	9.1	21
92	The complotype: dictating risk for inflammation and infection. Trends in Immunology, 2012, 33, 513-521.	6.8	132
93	Lessons from functional and structural analyses of disease-associated genetic variants in the complement alternative pathway. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2011, 1812, 12-22.	3.8	33
94	Laforin, a dual-specificity phosphatase involved in Lafora disease, is phosphorylated at Ser25 by AMP-activated protein kinase. Biochemical Journal, 2011, 439, 265-275.	3.7	29
95	Common polymorphisms in C3, factor B, and factor H collaborate to determine systemic complement activity and disease risk. Proceedings of the National Academy of Sciences of the United States of America, 2011, 108, 8761-8766.	7.1	198
96	Unique structure of iC3b resolved at a resolution of 24 Ã... by 3D-electron microscopy. Proceedings of the National Academy of Sciences of the United States of America, 2011, 108, 13236-13240.	7.1	49
97	Laforin, a dual specificity phosphatase involved in Lafora disease, regulates insulin response and whole-body energy balance in mice. Human Molecular Genetics, 2011, 20, 2571-2584.	2.9	16
98	aHUS: a disorder with many risk factors. Blood, 2010, 115, 158-160.	1.4	31
99	C4BPB/C4BPA is a new susceptibility locus for venous thrombosis with unknown protein Sâ€“independent mechanism: results from genome-wide association and gene expression analyses followed by case-control studies. Blood, 2010, 115, 4644-4650.	1.4	61
100	Complement Factor H Is Expressed in Adipose Tissue in Association With Insulin Resistance. Diabetes, 2010, 59, 200-209.	0.6	88
101	Impaired autophagy in Lafora disease. Autophagy, 2010, 6, 991-993.	9.1	30
102	Variant-specific quantification of factor H in plasma identifies null alleles associated with atypical hemolytic uremic syndrome. Kidney International, 2010, 78, 782-788.	5.2	42
103	Identification of a mutation in complement factor H-related protein 5 in patients of Cypriot origin with glomerulonephritis. Lancet, The, 2010, 376, 794-801.	13.7	298
104	Laforin, the most common protein mutated in Lafora disease, regulates autophagy. Human Molecular Genetics, 2010, 19, 2867-2876.	2.9	170
105	Human C3 mutation reveals a mechanism of dense deposit disease pathogenesis and provides insights into complement activation and regulation. Journal of Clinical Investigation, 2010, 120, 3702-3712.	8.2	195
106	Increased Endoplasmic Reticulum Stress and Decreased Proteasomal Function in Lafora Disease Models Lacking the Phosphatase Laforin. PLoS ONE, 2009, 4, e5907.	2.5	69
107	Functional basis of protection against age-related macular degeneration conferred by a common polymorphism in complement factor B. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 4366-4371.	7.1	98
108	AMP-activated Protein Kinase Phosphorylates R5/PTG, the Glycogen Targeting Subunit of the R5/PTG-Protein Phosphatase 1 Holoenzyme, and Accelerates Its Down-regulation by the Laforin-Malin Complex. Journal of Biological Chemistry, 2009, 284, 8247-8255.	3.4	53

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109	Characterization of complement factor Hâ€“related (CFHR) proteins in plasma reveals novel genetic variations of CFHR1 associated with atypical hemolytic uremic syndrome. Blood, 2009, 114, 4261-4271.	1.4	190
110	Coexistence of Closed and Open Conformations of Complement Factor B in the Alternative Pathway C3bB(Mg2+) Proconvertase. Journal of Immunology, 2009, 183, 7347-7351.	0.8	35
111	Liver-Kidney Transplantation to Cure Atypical Hemolytic Uremic Syndrome. Journal of the American Society of Nephrology: JASN, 2009, 20, 940-949.	6.1	154
112	The disease-protective complement factor H allotypic variant Ile62 shows increased binding affinity for C3b and enhanced cofactor activity. Human Molecular Genetics, 2009, 18, 3452-3461.	2.9	127
113	Successful Renal Transplantation in a Patient with Atypical Hemolytic Uremic Syndrome Carrying Mutations in Both Factor I and MCP. American Journal of Transplantation, 2009, 9, 1477-1483.	4.7	26
114	Lack of association between polymorphisms in C4b-binding protein and atypical haemolytic uraemic syndrome in the Spanish population. Clinical and Experimental Immunology, 2009, 155, 59-64.	2.6	13
115	3D structure of the C3bB complex provides insights into the activation and regulation of the complement alternative pathway convertase. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 882-887.	7.1	76
116	Mutations in Proteins of the Alternative Pathway of Complement and the Pathogenesis of Atypical Hemolytic Uremic Syndrome. American Journal of Kidney Diseases, 2008, 52, 171-180.	1.9	24
117	Genetic deficiency of complement factor H in a patient with age-related macular degeneration and membranoproliferative glomerulonephritis. Molecular Immunology, 2008, 45, 2897-2904.	2.2	46
118	Binding of complement regulatory proteins to Group A Streptococcus. Vaccine, 2008, 26, I75-I78.	3.8	18
119	Complement Factor H Binds to Denatured Rather than to Native Pentameric C-reactive Protein. Journal of Biological Chemistry, 2008, 283, 30451-30460.	3.4	82
120	The Complement Factor H R1210C Mutation Is Associated With Atypical Hemolytic Uremic Syndrome. Journal of the American Society of Nephrology: JASN, 2008, 19, 639-646.	6.1	81
121	Regulation of glycogen synthesis by the laforinâ€“malin complex is modulated by the AMP-activated protein kinase pathway. Human Molecular Genetics, 2008, 17, 667-678.	2.9	128
122	Measurement of Factor H Variants in Plasma Using Variant-Specific Monoclonal Antibodies: Application to Assessing Risk of Age-Related Macular Degeneration. , 2008, 49, 1983.		80
123	Hepatic disease as the first manifestation of progressive myoclonus epilepsy of Lafora. Neurology, 2007, 68, 1369-1373.	1.1	32
124	Spontaneous hemolytic uremic syndrome triggered by complement factor H lacking surface recognition domains. Journal of Experimental Medicine, 2007, 204, 1249-1256.	8.5	267
125	Gain-of-function mutations in complement factor B are associated with atypical hemolytic uremic syndrome. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 240-245.	7.1	429
126	Membrane cofactor protein (MCP, CD46) binding to clinical isolates of Streptococcus pyogenes: Binding to M type 18 strains is independent of Emm or Enn proteins. Molecular Immunology, 2007, 44, 3571-3579.	2.2	12

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127	Complement analysis in the 21st century. Molecular Immunology, 2007, 44, 3838-3849.	2.2	132
128	New Approaches to the Treatment of Dense Deposit Disease. Journal of the American Society of Nephrology: JASN, 2007, 18, 2447-2456.	6.1	231
129	The interactive Factor H-atypical hemolytic uremic syndrome mutation database and website: update and integration of membrane cofactor protein and Factor I mutations with structural models. Human Mutation, 2007, 28, 222-234.	2.5	160
130	Reply to â€œReactive oxygen species and the segregation of mtDNA sequence variantsâ€• Nature Genetics, 2007, 39, 572-572.	21.4	0
131	Mechanism suppressing glycogen synthesis in neurons and its demise in progressive myoclonus epilepsy. Nature Neuroscience, 2007, 10, 1407-1413.	14.8	320
132	Successful Liver-Kidney Transplantation in Two Children With aHUS Caused by a Mutation in Complement Factor H. American Journal of Transplantation, 2007, 8, 071105081616017-???.	4.7	83
133	Translational Mini-Review Series on Complement Factor H: Genetics and disease associations of human complement factor H. Clinical and Experimental Immunology, 2007, 151, 1-13.	2.6	252
134	Insights into hemolytic uremic syndrome: Segregation of three independent predisposition factors in a large, multiple affected pedigree. Molecular Immunology, 2006, 43, 1769-1775.	2.2	122
135	Differences in reactive oxygen species production explain the phenotypes associated with common mouse mitochondrial DNA variants. Nature Genetics, 2006, 38, 1261-1268.	21.4	301
136	Ochronotic rheumatism in Algeria: clinical, radiological, biological and molecular studiesâ€”a case study of 14 patients in 11 families. Joint Bone Spine, 2006, 73, 284-292.	1.6	27
137	m.6267G>A: a recurrent mutation in the human mitochondrial DNA that reduces cytochrome c oxidase activity and is associated with tumors. Human Mutation, 2006, 27, 575-582.	2.5	56
138	De novo gene conversion in the RCA gene cluster (1q32) causes mutations in complement factor H associated with atypical hemolytic uremic syndrome. Human Mutation, 2006, 27, 292-293.	2.5	143
139	Expression of the peptide C4b-binding protein Â in the arthritic joint. Annals of the Rheumatic Diseases, 2006, 65, 1279-1285.	0.9	8
140	Genomic sequence of the pathogenic and allergenic filamentous fungus Aspergillus fumigatus. Nature, 2005, 438, 1151-1156.	27.8	1,272
141	Predisposition to atypical hemolytic uremic syndrome involves the concurrence of different susceptibility alleles in the regulators of complement activation gene cluster in 1q32. Human Molecular Genetics, 2005, 14, 703-712.	2.9	272
142	Predisposition to atypical hemolytic uremic syndrome involves the concurrence of different susceptibility alleles in the regulators of complement activation gene cluster in 1q32. Human Molecular Genetics, 2005, 14, 1107-1107.	2.9	7
143	Interaction between Complement Regulators andStreptococcus pyogenes: Binding of C4b-Binding Protein and Factor H/Factor H-Like Protein 1 to M18 Strains Involves Two Different Cell Surface Molecules. Journal of Immunology, 2004, 173, 6899-6904.	0.8	53
144	Genetic and environmental factors influencing the human factorï¿½H plasma levels. Immunogenetics, 2004, 56, 77-82.	2.4	145

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145	Analysis of the developmentalSIX6homeobox gene in patients with anophthalmia/microphthalmia. , 2004, 129A, 92-94.		52
146	Functional analysis in serum from atypical Hemolytic Uremic Syndrome patients reveals impaired protection of host cells associated with mutations in factor H. Molecular Immunology, 2004, 41, 81-84.	2.2	181
147	The human complement factor H: functional roles, genetic variations and disease associations. Molecular Immunology, 2004, 41, 355-367.	2.2	514
148	Molecular analyses of the HGO gene mutations in Turkish alkaptonuria patients suggest that the R58fs mutation originated from Central Asia and was spread throughout Europe and Anatolia by human migrations. Journal of Inherited Metabolic Disease, 2003, 26, 17-23.	3.6	19
149	Genetic determinants of variation in the plasma levels of the C4b-binding protein (C4BP) in Spanish families. Immunogenetics, 2003, 54, 862-866.	2.4	17
150	Functional analysis of MCCA and MCCB mutations causing methylcrotonylglycinuria. Molecular Genetics and Metabolism, 2003, 80, 315-320.	1.1	20
151	Laforin, the dual-phosphatase responsible for Lafora disease, interacts with R5 (PTG), a regulatory subunit of protein phosphatase-1 that enhances glycogen accumulation. Human Molecular Genetics, 2003, 12, 3161-3171.	2.9	102
152	Structural and Functional Characterization of Factor H Mutations Associated with Atypical Hemolytic Uremic Syndrome. American Journal of Human Genetics, 2002, 71, 1285-1295.	6.2	208
153	Cloning, characterization and chromosome mapping of the human SMAP1 gene. Gene, 2002, 292, 167-171.	2.2	12
154	Clustering of Missense Mutations in the C-Terminal Region of Factor H in Atypical Hemolytic Uremic Syndrome. American Journal of Human Genetics, 2001, 68, 478-484.	6.2	280
155	The Molecular Basis of 3-Methylcrotonylglycinuria, a Disorder of Leucine Catabolism. American Journal of Human Genetics, 2001, 68, 334-346.	6.2	73
156	Assessment of the interaction of human complement regulatory proteins with group AStreptococcus. Identification of a high-affinity group AStreptococcus binding site in FHL-1. European Journal of Immunology, 2000, 30, 1243-1253.	2.9	59
157	Cytokine-mediated up-regulation of CD55 and CD59 protects human hepatoma cells from complement attack. Clinical and Experimental Immunology, 2000, 121, 234-241.	2.6	64
158	Crystal structure of human homogentisate dioxygenase. Nature Structural Biology, 2000, 7, 542-546.	9.7	137
159	Mutational spectrum of the EPM2A gene in progressive myoclonus epilepsy of Lafora: high degree of allelic heterogeneity and prevalence of deletions. European Journal of Human Genetics, 2000, 8, 946-954.	2.8	55
160	Molecular basis for factor H and FHL-1 deficiency in an Italian family. Immunogenetics, 2000, 51, 366-369.	2.4	24
161	Structural and functional analysis of mutations in alkaptonuria. Human Molecular Genetics, 2000, 9, 2341-2350.	2.9	80
162	High Frequency of Alkaptonuria in Slovakia: Evidence for the Appearance of Multiple Mutations in HGO Involving Different Mutational Hot Spots. American Journal of Human Genetics, 2000, 67, 1333-1339.	6.2	62

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