Robert Desnick

List of Publications by Year
in descending order

280 papers	22,659 citations	5574 82 h-index	9588 142 g-index
299 all docs	299 docs citations	299 times ranked	14843 citing authors

#	Article	IF	Citations
1	Acute Hepatic Porphyrias: â€œPurple Flagsâ€â€”Clinical Features That Should Prompt Specific Diagnostic Testing. American Journal of the Medical Sciences, 2022, 363, 1-10.	1.1	7
2	Hepatocellular Carcinoma in Acute Hepatic Porphyrias: Results from the Longitudinal Study of the U.S. Porphyrias Consortium. Hepatology, 2021, 73, 1736-1746.	7.3	32
3	Agalsidase beta treatment slows estimated glomerular filtration rate loss in classic Fabry disease patients: results from an individual patient data meta-analysis. CKJ: Clinical Kidney Journal, 2021, 14, 1136-1146.	2.9	9
4	Inherited Porphyrias. , 2021, , 373-411.		1
5	Expression Profiling Identifies TWIST2 Target Genes in Setleis Syndrome Patient Fibroblast and Lymphoblast Cells. International Journal of Environmental Research and Public Health, 2021, 18, 1997.	2.6	3
6	Heme Biosynthesis and the Porphyrias in Children. , 2021, , 530-547.		0
7	ZFN-mediated inÂvivo gene editing in hepatocytes leads to supraphysiologic Î±-Gal A activity and effective substrate reduction in Fabry mice. Molecular Therapy, 2021, 29, 3230-3242.	8.2	9
8	Erythropoietic protoporphyria: time to prodrome, the warning signal to exit sun exposure without painâ€”a patient-reported outcome efficacy measure. Genetics in Medicine, 2021, 23, 1616-1623.	2.4	10
9	ABCB6 Polymorphisms are not Overly Represented in Patients with Porphyria. Blood Advances, 2021, , .	5.2	2
10	Evaluating the Patient-Reported Outcomes Measurement Information System scales in acute intermittent porphyria. Genetics in Medicine, 2020, 22, 590-597.	2.4	8
11	EXPLORE: A Prospective, Multinational, Natural History Study of Patients with Acute Hepatic Porphyria with Recurrent Attacks. Hepatology, 2020, 71, 1546-1558.	7.3	103
12	Porphyric Neuropathy: Pathophysiology, Diagnosis, and Updated Management. Current Neurology and Neuroscience Reports, 2020, 20, 56.	4.2	17
13	5-Aminolevulinate dehydratase porphyria: Update on hepatic 5-aminolevulinic acid synthase induction and long-term response to hemin. Molecular Genetics and Metabolism, 2020, 131, 418-423.	1.1	11
14	Phased Haplotype Resolution of the SLC6A4 Promoter Using Long-Read Single Molecule Real-Time (SMRT) Sequencing. Genes, 2020, 11, 1333.	2.4	5
15	AAV2/6 Gene Therapy in a Murine Model of Fabry Disease Results in Supraphysiological Enzyme Activity and Effective Substrate Reduction. Molecular Therapy - Methods and Clinical Development, 2020, 18, 607-619.	4.1	29
16	Diagnostic yield and clinical utility of whole exome sequencing using an automated variant prioritization system, <scp>EVIDENCE</scp>. Clinical Genetics, 2020, 98, 562-570.	2.0	76
17	Human aminolevulinate synthase structure reveals a eukaryotic-specific autoinhibitory loop regulating substrate binding and product release. Nature Communications, 2020, 11, 2813.	12.8	25
18	Severe hydroxymethylbilane synthase deficiency causes depression-like behavior and mitochondrial dysfunction in a mouse model of homozygous dominant acute intermittent porphyria. Acta Neuropathologica Communications, 2020, 8, 38.	5.2	5

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19	A novel approach to conducting clinical trials in the community setting: utilizing patient-driven platforms and social media to drive web-based patient recruitment. BMC Medical Research Methodology, 2020, 20, 58.	3.1	20
20	Fabry disease: Î±-galactosidase A deficiency. , 2020, , 575-587.		2
21	Erythropoietic Protoporphyria: Phase 2 Clinical Trial Results Evaluating the Safety and Effectiveness of Dersimelagon (MT-7117), an Oral MC1R Agonist. Blood, 2020, 136, 51-51.	1.4	8
22	Fabry disease. , 2019, , 287-292.		0
23	The New York pilot newborn screening program for lysosomal storage diseases: Report of the First 65,000 Infants. Genetics in Medicine, 2019, 21, 631-640.	2.4	113
24	Congenital erythropoietic porphyria: Recent advances. Molecular Genetics and Metabolism, 2019, 128, 288-297.	1.1	45
25	Murine models of the human porphyrias: Contributions toward understanding disease pathogenesis and the development of new therapies. Molecular Genetics and Metabolism, 2019, 128, 332-341.	1.1	12
26	International Porphyria Molecular Diagnostic Collaborative: an evidence-based database of verified pathogenic and benign variants for the porphyrias. Genetics in Medicine, 2019, 21, 2605-2613.	2.4	16
27	Harderoporphyria: Case of lifelong photosensitivity associated with compound heterozygous coproporphyrinogen oxidase (CPOX) mutations. Molecular Genetics and Metabolism Reports, 2019, 19, 100457.	1.1	2
28	Phase 1 Trial of an RNA Interference Therapy for Acute Intermittent Porphyria. New England Journal of Medicine, 2019, 380, 549-558.	27.0	194
29	Identification and characterization of 40 novel hydroxymethylbilane synthase mutations that cause acute intermittent porphyria. Journal of Inherited Metabolic Disease, 2019, 42, 186-194.	3.6	17
30	Congenital erythropoietic porphyria and erythropoietic protoporphyria: Identification of 7 uroporphyrinogen III synthase and 20 ferrochelatase novel mutations. Molecular Genetics and Metabolism, 2019, 128, 358-362.	1.1	9
31	Strong correlation of ferrochelatase enzymatic activity with Mitoferrin-1 mRNA in lymphoblasts of patients with protoporphyria. Molecular Genetics and Metabolism, 2019, 128, 391-395.	1.1	7
32	Sex differences in vascular reactivity in mesenteric arteries from a mouse model of acute intermittent porphyria. Molecular Genetics and Metabolism, 2019, 128, 376-381.	1.1	16
33	Characterization of the hepatic transcriptome following phenobarbital induction in mice with AIP. Molecular Genetics and Metabolism, 2019, 128, 382-390.	1.1	7
34	Homozygous hydroxymethylbilane synthase knock-in mice provide pathogenic insights into the severe neurological impairments present in human homozygous dominant acute intermittent porphyria. Human Molecular Genetics, 2019, 28, 1755-1767.	2.9	17
35	Integrated CYP2D6 interrogation for multiethnic copy number and tandem allele detection. Pharmacogenomics, 2019, 20, 9-20.	1.3	9
36	Acute hepatic porphyrias: Identification of 46 hydroxymethylbilane synthase, 11 coproporphyrinogen oxidase, and 20 protoporphyrinogen oxidase novel mutations. Molecular Genetics and Metabolism, 2019, 128, 352-357.	1.1	2

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37	Therapies for Lysosomal Storage Diseases. , 2019, , 205-227.		1
38	Porphyria cutanea tarda and hepatoerythropoietic porphyria: Identification of 19 novel uroporphyrinogen III decarboxylase mutations. Molecular Genetics and Metabolism, 2019, 128, 363-366.	1.1	9
39	Recent advances on porphyria genetics: Inheritance, penetrance & molecular heterogeneity, including new modifying/causative genes. Molecular Genetics and Metabolism, 2019, 128, 320-331.	1.1	59
40	Fabry disease revisited: Management and treatment recommendations for adult patients. Molecular Genetics and Metabolism, 2018, 123, 416-427.	1.1	391
41	Long-term Outcomes of Kidney Transplantation in Fabry Disease. Transplantation, 2018, 102, 1924-1933.	1.0	18
42	Human hydroxymethylbilane synthase: Molecular dynamics of the pyrrole chain elongation identifies step-specific residues that cause AIP. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, E4071-E4080.	7.1	32
43	Fabry Disease: prevalence of affected males and heterozygotes with pathogenic GLA mutations identified by screening renal, cardiac and stroke clinics, 1995â€“2017. Journal of Medical Genetics, 2018, 55, 261-268.	3.2	91
44	Focal facial dermal dysplasia type 4: identification of novel CYP26C1 mutations in unrelated patients. Journal of Human Genetics, 2018, 63, 257-261.	2.3	3
45	Functional variants in the LRRK2 gene confer shared effects on risk for Crohnâ€™s disease and Parkinsonâ€™s disease. Science Translational Medicine, 2018, 10, .	12.4	273
46	Identification and characterization of 40 novel hydroxymethylbilane synthase mutations that cause acute intermittent porphyria. Journal of Inherited Metabolic Disease, 2018, 42, 186.	3.6	9
47	Identification of Fabry Disease in a Tertiary Referral Cohort of Patients with Hypertrophic Cardiomyopathy. American Journal of Medicine, 2018, 131, 200.e1-200.e8.	1.5	31
48	Reply. Hepatology, 2018, 67, 803-804.	7.3	0
49	Newborn screening for lysosomal storage disorders by tandem mass spectrometry in North East Italy. Journal of Inherited Metabolic Disease, 2018, 41, 209-219.	3.6	114
50	Parkinson's disease prevalence in Fabry disease: A survey study. Molecular Genetics and Metabolism Reports, 2018, 14, 27-30.	1.1	34
51	Design and validation of an open-source modular Microplate Photoirradiation System for high-throughput photobiology experiments. PLoS ONE, 2018, 13, e0203597.	2.5	5
52	Many pitfalls in diagnosis of acute intermittent porphyria: a case report. BMC Research Notes, 2018, 11, 552.	1.4	10
53	Multi-ethnicSULT1A1copy number profiling with multiplex ligation-dependent probe amplification. Pharmacogenomics, 2018, 19, 761-770.	1.3	9
54	Roscoe Owen Brady, MD: Remembrances of co-investigators and colleagues. Molecular Genetics and Metabolism, 2017, 120, 1-7.	1.1	3

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55	Types A and B Niemann-Pick disease. Molecular Genetics and Metabolism, 2017, 120, 27-33.	1.1	196
56	Acute hepatic porphyrias: Recommendations for evaluation and longâ€term management. Hepatology, 2017, 66, 1314-1322.	7.3	122
57	Clinical, Biochemical, and Genetic Characterization of North American Patients With Erythropoietic Protoporphyria and X-linked Protoporphyria. JAMA Dermatology, 2017, 153, 789.	4.1	70
58	Institutional profile: translational pharmacogenomics at the Icahn School of Medicine at Mount Sinai. Pharmacogenomics, 2017, 18, 1381-1386.	1.3	20
59	Fabry disease: characterisation of the plasma proteome pre- and post-enzyme replacement therapy. Journal of Medical Genetics, 2017, 54, 771-780.	3.2	20
60	Evolution of cardiac pathology in classic Fabry disease: Progressive cardiomyocyte enlargement leads to increased cell death and fibrosis, and correlates with severity of ventricular hypertrophyâ€¬â€¬â€¬â€¬â€¬â€¬â€¬â€¬â€¬â€¬â€¬â€¬â€¬. International Journal of Cardiology, 2017, 248, 257-262.	1.7	13
61	Fabry Disease in Families With Hypertrophic Cardiomyopathy. Circulation: Cardiovascular Genetics, 2017, 10, .	5.1	17
62	Correlation of Lyso-Gb3 levels in dried blood spots and sera from patients with classic and Later-Onset Fabry disease. Molecular Genetics and Metabolism, 2017, 121, 320-324.	1.1	50
63	The focal facial dermal dysplasias: phenotypic spectrum and molecular genetic heterogeneity. Journal of Medical Genetics, 2017, 54, 585-590.	3.2	10
64	Early manifestations of type 1 Gaucher disease in presymptomatic children diagnosed after parental carrier screening. Genetics in Medicine, 2017, 19, 652-658.	2.4	19
65	Plasma LysoGb3: A useful biomarker for the diagnosis and treatment of Fabry disease heterozygotes. Molecular Genetics and Metabolism, 2017, 120, 57-61.	1.1	99
66	The validation of pharmacogenetics for the identification of Fabry patients to be treated with migalastat. Genetics in Medicine, 2017, 19, 430-438.	2.4	157
67	A Frameshift in CSF2RB Predominant Among Ashkenazi Jews Increases Risk for Crohn's Disease and Reduces Monocyte Signaling via GM-CSF. Gastroenterology, 2016, 151, 710-723.e2.	1.3	51
68	Exome sequencing of extreme clopidogrel response phenotypes identifies B4GALT2 as a determinant of onâ€treatment platelet reactivity. Clinical Pharmacology and Therapeutics, 2016, 100, 287-294.	4.7	22
69	Long-Read Single Molecule Real-Time Full Gene Sequencing of Cytochrome P450-2D6. Human Mutation, 2016, 37, 315-323.	2.5	86
70	Later Onset Fabry Disease, Cardiac Damage Progress in Silence. Journal of the American College of Cardiology, 2016, 68, 2554-2563.	2.8	81
71	Response to â€œLetter to the Editor by Drs Block and Razaniâ€. Journal of Clinical Lipidology, 2016, 10, 1281-1282.	1.5	2
72	Experiences and concerns of patients with recurrent attacks of acute hepatic porphyria: A qualitative study. Molecular Genetics and Metabolism, 2016, 119, 278-283.	1.1	60

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73	Xâ€chromosomal inactivation directly influences the phenotypic manifestation of Xâ€linked protoporphyria. Clinical Genetics, 2016, 89, 20-26.	2.0	25
74	Acute Intermittent Porphyria: Predicted Pathogenicity ofHMBSVariants Indicates Extremely Low Penetrance of the Autosomal Dominant Disease. Human Mutation, 2016, 37, 1215-1222.	2.5	129
75	Acute Intermittent Porphyria in children: A case report and review of the literature. Molecular Genetics and Metabolism, 2016, 119, 295-299.	1.1	31
76	Genetics of GNE myopathy in the non-Jewish Persian population. European Journal of Human Genetics, 2016, 24, 243-251.	2.8	18
77	Focal Facial Dermal Dysplasias. , 2016, , 501-505.		1
78	Interim Data from a Randomized, Placebo Controlled, Phase 1 Study of Aln-AS1, an Investigational RNAi Therapeutic for the Treatment of Acute Hepatic Porphyria. Blood, 2016, 128, 2318-2318.	1.4	3
79	The D519G Polymorphism of Glyceronephosphate O-Acyltransferase Is a Risk Factor for Familial Porphyria Cutanea Tarda. PLoS ONE, 2016, 11, e0163322.	2.5	7
80	Efficacy of Enzyme and Substrate Reduction Therapy with a Novel Antagonist of Glucosylceramide Synthase for Fabry Disease. Molecular Medicine, 2015, 21, 389-399.	4.4	72
81	Liver Transplantation for Acute Intermittent Porphyria: Biochemical and Pathologic Studies of the Explanted Liver. Molecular Medicine, 2015, 21, 487-495.	4.4	51
82	Pitfalls in Erythrocyte Protoporphyrin Measurement for Diagnosis and Monitoring of Protoporphyrias. Clinical Chemistry, 2015, 61, 1453-1456.	3.2	29
83	Setleis syndrome due to inheritance of the 1p36.22p36.21 duplication: evidence for lack of penetrance. Journal of Human Genetics, 2015, 60, 717-722.	2.3	12
84	Î±-Galactosidase A Knockout Mice. American Journal of Pathology, 2015, 185, 651-665.	3.8	34
85	Afamelanotide for Erythropoietic Protoporphyria. New England Journal of Medicine, 2015, 373, 48-59.	27.0	206
86	Ten-year outcome of enzyme replacement therapy with agalsidase beta in patients with Fabry disease. Journal of Medical Genetics, 2015, 52, 353-358.	3.2	266
87	Quantitative and multiplexed DNA methylation analysis using long-read single-molecule real-time bisulfite sequencing (SMRT-BS). BMC Genomics, 2015, 16, 350.	2.8	68
88	Fabry Disease. , 2015, , 419-430.		3
89	Chromosome 1p36.22p36.21 duplications/triplication causes Setleis syndrome (focal facial dermal) Tj ETQq1 1 0.784314 rgBT /Overl	1.2	15
90	Actionable exomic incidental findings in 6503 participants: challenges of variant classification. Genome Research, 2015, 25, 305-315.	5.5	313

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91	Preclinical Development of a Subcutaneous ALAS1 RNAi Therapeutic for Treatment of Hepatic Porphyrias Using Circulating RNA Quantification. Molecular Therapy - Nucleic Acids, 2015, 4, e263.	5.1	107
92	Setleis syndrome: clinical, molecular and structural studies of the first <scp>TWIST2</scp> missense mutation. Clinical Genetics, 2015, 88, 489-493.	2.0	12
93	RNAi-mediated silencing of hepatic Alas1 effectively prevents and treats the induced acute attacks in acute intermittent porphyria mice. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, 7777-7782.	7.1	99
94	An Ashkenazi Jewish SMN1 haplotype specific to duplication alleles improves pan-ethnic carrier screening for spinal muscular atrophy. Genetics in Medicine, 2014, 16, 149-156.	2.4	64
95	Genome-wide mapping of IBD segments in an Ashkenazi PD cohort identifies associated haplotypes. Human Molecular Genetics, 2014, 23, 4693-4702.	2.9	49
96	Acute Porphyrias in the USA: Features of 108ÂSubjects from Porphyrias Consortium. American Journal of Medicine, 2014, 127, 1233-1241.	1.5	185
97	JCL Roundtable: Enzyme replacement therapy for lipid storage disorders. Journal of Clinical Lipidology, 2014, 8, 463-472.	1.5	3
98	Setleis Syndrome: Genetic and Clinical Findings in a New Case With Epilepsy. Pediatric Neurology, 2014, 50, 389-391.	2.1	3
99	Therapies for Lysosomal Storage Diseases. , 2013, , 1-30.		1
100	An Allele-Specific PCR System for Rapid Detection and Discrimination of the CYP2C19âˆ—4A, âˆ—4B, and âˆ—17 Alleles. Journal of Molecular Diagnostics, 2013, 15, 783-789.	2.8	17
101	Diagnosis of feline acute intermittent porphyria presenting with erythrodontia requires molecular analyses. Veterinary Journal, 2013, 198, 720-722.	1.7	8
102	Multi-ethnic distribution of clinically relevant CYP2C genotypes and haplotypes. Pharmacogenomics Journal, 2013, 13, 369-377.	2.0	87
103	Extended haplotype association study in Crohnâ€™s disease identifies a novel, Ashkenazi Jewish-specific missense mutation in the NF-ÎºB pathway gene, HEATR3. Genes and Immunity, 2013, 14, 310-316.	4.1	31
104	Focal facial dermal dysplasia, type IV, is caused by mutations in CYP26C1. Human Molecular Genetics, 2013, 22, 696-703.	2.9	41
105	Frequency of the cholesteryl ester storage disease commonLIPAE8SJM mutation (c.894G>A) in various racial and ethnic groups. Hepatology, 2013, 58, 958-965.	7.3	85
106	Loss-of-Function Ferrochelatase and Gain-of-Function Erythroid-Specific 5-Aminolevulinate Synthase Mutations Causing Erythropoietic Protoporphyria and X-Linked Protoporphyria in North American Patients Reveal Novel Mutations and a High Prevalence of X-Linked Protoporphyria. Molecular Medicine, 2013, 19, 26-29.	4.4	74
107	Molecular Expression and Characterization of Erythroid-Specific 5-Aminolevulinate Synthase Gain-of-Function Mutations Causing X-Linked Protoporphyria. Molecular Medicine, 2013, 19, 18-25.	4.4	33
108	A Genome-Wide Scan of Ashkenazi Jewish Crohn's Disease Suggests Novel Susceptibility Loci. PLoS Genetics, 2012, 8, e1002559.	3.5	144

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109	Identification of CYP2C19*4B: pharmacogenetic implications for drug metabolism including clopidogrel responsiveness. Pharmacogenomics Journal, 2012, 12, 297-305.	2.0	40
110	The porphyrias: advances in diagnosis and treatment. Blood, 2012, 120, 4496-4504.	1.4	207
111	Copy number variation and warfarin dosing: evaluation ofCYP2C9,VKORC1,CYP4F2,GGCXandCALU. Pharmacogenomics, 2012, 13, 297-307.	1.3	13
112	Enzyme Replacement Therapy for Lysosomal Diseases: Lessons from 20 Years of Experience and Remaining Challenges. Annual Review of Genomics and Human Genetics, 2012, 13, 307-335.	6.2	229
113	Fabry Disease: Incidence of the Common Later-Onset Î±-Galactosidase A IVS4+919Gâ†'A Mutation in Taiwanese Newbornsâ€"Superiority of DNA-Based to Enzyme-Based Newborn Screening for Common Mutations. Molecular Medicine, 2012, 18, 780-784.	4.4	71
114	The porphyrias: advances in diagnosis and treatment. Hematology American Society of Hematology Education Program, 2012, 2012, 19-27.	2.5	34
115	Setleis syndrome in Mexican-Nahua sibs due to a homozygous TWIST2 frameshift mutation and partial expression in heterozygotes: review of the focal facial dermal dysplasias and subtype reclassification. Journal of Medical Genetics, 2011, 48, 716-720.	3.2	27
116	Increasing Tamoxifen Dose in Breast Cancer Patients Based on CYP2D6 Genotypes and Endoxifen Levels: Effect on Active Metabolite Isomers and the Antiestrogenic Activity Score. Clinical Pharmacology and Therapeutics, 2011, 90, 605-611.	4.7	73
117	Nonsense mutations of the bHLH transcription factor TWIST2 found in Setleis Syndrome patients cause dysregulation of periostin. International Journal of Biochemistry and Cell Biology, 2011, 43, 1523-1531.	2.8	18
118	Diagnostic dilemma: A young woman with Fabry disease symptoms, no family history, and a â€œsequencing crypticâ€•Î±-galactosidase a large deletion. Molecular Genetics and Metabolism, 2011, 104, 314-318.	1.1	11
119	Tay-Sachs disease in an Arab family due to c.78G>A HEXA nonsense mutation encoding a p.W26X early truncation enzyme peptide. Molecular Genetics and Metabolism, 2011, 104, 700-702.	1.1	8
120	CYP1A2*1F and GSTM1 Alleles Are Associated with Susceptibility to Porphyria Cutanea Tarda. Molecular Medicine, 2011, 17, 241-247.	4.4	19
121	Congenital Erythropoietic Porphyria: Characterization of Murine Models of the Severe Common (C73R/C73R) and Later-Onset Genotypes. Molecular Medicine, 2011, 17, 748-756.	4.4	10
122	Harderoporphyria due to homozygosity for coproporphyrinogen oxidase missense mutation H327R. Journal of Inherited Metabolic Disease, 2011, 34, 225-231.	3.6	22
123	Gaucher disease: when molecular testing and clinical presentation disagree -the novel c.1226A>G(p.N370S)--RecNcil allele. Journal of Inherited Metabolic Disease, 2011, 34, 789-793.	3.6	9
124	Evaluation of 22 genetic variants with Crohn's Disease risk in the Ashkenazi Jewish population: a case-control study. BMC Medical Genetics, 2011, 12, 63.	2.1	41
125	Detection of large gene rearrangements in X-linked genes by dosage analysis: identification of novel Î±-galactosidase A (GLA) deletions causing Fabry disease. Human Mutation, 2011, 32, 688-695.	2.5	13
126	A LCâ€"MS/MS method for the specific, sensitive, and simultaneous quantification of 5-aminolevulinic acid and porphobilinogen. Journal of Chromatography B: Analytical Technologies in the Biomedical and Life Sciences, 2011, 879, 2389-2396.	2.3	37

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127	Identification of two HEXA mutations causing infantile-onset Tayâ€“Sachs disease in the Persian population. Journal of Human Genetics, 2011, 56, 682-684.	2.3	5
128	Congenital erythropoietic porphyria: a novel uroporphyrinogen III synthase branchpoint mutation reveals underlying wild-type alternatively spliced transcripts. Blood, 2010, 115, 1062-1069.	1.4	15
129	Homozygous Nonsense Mutations in TWIST2 Cause Setleis Syndrome. American Journal of Human Genetics, 2010, 87, 289-296.	6.2	65
130	Experience with carrier screening and prenatal diagnosis for 16 Ashkenazi Jewish genetic diseases. Human Mutation, 2010, 31, 1240-1250.	2.5	125
131	Feline Congenital Erythropoietic Porphyria: Two Homozygous UROS Missense Mutations Cause the Enzyme Deficiency and Porphyrin Accumulation. Molecular Medicine, 2010, 16, 381-388.	4.4	23
132	Feline acute intermittent porphyria: a phenocopy masquerading as an erythropoietic porphyria due to dominant and recessive hydroxymethylbilane synthase mutations. Human Molecular Genetics, 2010, 19, 584-596.	2.9	32
133	The Pharmacological Chaperone 1-Deoxygalactonojirimycin Reduces Tissue Globotriaosylceramide Levels in a Mouse Model of Fabry Disease. Molecular Therapy, 2010, 18, 23-33.	8.2	126
134	Combined CYP2C9, VKORC1 and CYP4F2 frequencies among racial and ethnic groups. Pharmacogenomics, 2010, 11, 781-791.	1.3	146
135	Hepatoerythropoietic Porphyria Misdiagnosed as Child Abuse. Archives of Dermatology, 2010, 146, 529-33.	1.4	11
136	AAV8-mediated Gene Therapy Prevents Induced Biochemical Attacks of Acute Intermittent Porphyria and Improves Neuromotor Function. Molecular Therapy, 2010, 18, 17-22.	8.2	52
137	Type 1 Gaucher Disease. Archives of Internal Medicine, 2010, 170, 1463-9.	3.8	58
138	Tamoxifen Metabolite Isomer Separation and Quantification by Liquid Chromatographyâ^'Tandem Mass Spectrometry. Analytical Chemistry, 2010, 82, 10186-10193.	6.5	28
139	Pompe disease: Dramatic improvement in gastrointestinal function following enzyme replacement therapy. A report of three later-onset patients. Molecular Genetics and Metabolism, 2010, 101, 130-133.	1.1	52
140	Newborn screening for Fabry disease in Taiwan reveals a high incidence of the later-onsetGLAmutation c.936+919G>A (IVS4+919G>A). Human Mutation, 2009, 30, 1397-1405.	2.5	299
141	Fabry disease: progression of nephropathy, and prevalence of cardiac and cerebrovascular events before enzyme replacement therapy. Nephrology Dialysis Transplantation, 2009, 24, 2102-2111.	0.7	297
142	Genomics and personalized medicine: a perspective. Personalized Medicine, 2009, 6, 135-137.	1.5	0
143	Identification and Characterization of Feline Acute Intermittent Porphyria: The First Naturally-Occurring Animal Model.. Blood, 2009, 114, 3014-3014.	1.4	0
144	Human uroporphyrinogen III synthase: NMRâ€based mapping of the active site. Proteins: Structure, Function and Bioinformatics, 2008, 71, 855-873.	2.6	16

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145	Warfarin Pharmacogenetics: CYP2C9 and VKORC1 Genotypes Predict Different Sensitivity and Resistance Frequencies in the Ashkenazi and Sephardi Jewish Populations. American Journal of Human Genetics, 2008, 82, 495-500.	6.2	122
146	Sustained, Long-Term Renal Stabilization After 54 Months of Agalsidase Î² Therapy in Patients with Fabry Disease. Journal of the American Society of Nephrology: JASN, 2007, 18, 1547-1557.	6.1	396
147	CYP2C9,CYP2C19andCYP2D6allele frequencies in the Ashkenazi Jewish population. Pharmacogenomics, 2007, 8, 721-730.	1.3	43
148	Agalsidase-Beta Therapy for Advanced Fabry Disease. Annals of Internal Medicine, 2007, 146, 77.	3.9	493
149	Type 1 Gaucher disease: null and hypomorphic novel chitotriosidase mutations-implications for diagnosis and therapeutic monitoring. Human Mutation, 2007, 28, 866-873.	2.5	52
150	Acute intermittent porphyria: vector optimization for gene therapy. Journal of Gene Medicine, 2007, 9, 806-811.	2.8	6
151	Prenatal diagnosis of Fabry disease. Prenatal Diagnosis, 2007, 27, 693-694.	2.3	36
152	High Incidence of Later-Onset Fabry Disease Revealed by Newborn Screening*. American Journal of Human Genetics, 2006, 79, 31-40.	6.2	842
153	Fabry disease: Identification of 50 novel Î±-galactosidase A mutations causing the classic phenotype and three-dimensional structural analysis of 29 missense mutations. Human Genomics, 2006, 2, 297.	2.9	130
154	Fabry disease: clinical spectrum and evidence-based enzyme replacement therapy. Nephrologie Et Therapeutique, 2006, 2 Suppl 2, S172-85.	0.5	8
155	Detection of ?-galactosidase a mutations causing fabry disease by denaturing high performance liquid chromatography. Human Mutation, 2005, 25, 299-305.	2.5	51
156	Gastrointestinal manifestations of Fabry disease: Clinical response to enzyme replacement therapy. Molecular Genetics and Metabolism, 2005, 85, 255-259.	1.1	73
157	Recommendations for the Diagnosis and Treatment of the Acute Porphyrias. Annals of Internal Medicine, 2005, 142, 439.	3.9	485
158	Acute Intermittent Porphyria. Archives of Neurology, 2004, 61, 1764.	4.5	92
159	AAV2 Vector Harboring a Liver-Restricted Promoter Facilitates Sustained Expression of Therapeutic Levels of Î±-Galactosidase A and the Induction of Immune Tolerance in Fabry Mice. Molecular Therapy, 2004, 9, 231-240.	8.2	127
160	Fabry disease: renal involvement limited to podocyte pathology and proteinuria in a septuagenarian cardiac variant. Pathologic and therapeutic implications. American Journal of Kidney Diseases, 2004, 43, 164-171.	1.9	43
161	Enzyme replacement therapy for Fabry disease: lessons from two Î±-galactosidase A orphan products and one FDA approval. Expert Opinion on Biological Therapy, 2004, 4, 1167-1176.	3.1	99
162	Cystic fibrosis population carrier screening: 2004 revision of American College of Medical Genetics mutation panel. Genetics in Medicine, 2004, 6, 387-391.	2.4	413

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