

Robert Desnick

List of Publications by Year in descending order

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Version: 2024-02-01

280
papers

22,659
citations

5574

82
h-index

9589

142
g-index

299
all docs

299
docs citations

299
times ranked

14843
citing authors

#	ARTICLE	IF	CITATIONS
1	Acute Hepatic Porphyrrias: “Purple Flags” 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 Porphyrrias: Results from the Longitudinal Study of the U.S. Porphyrrias 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 Porphyrrias. , 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 Porphyrrias in Children. , 2021, , 530-547.		0
7	ZFN-mediated in vivo gene editing in hepatocytes leads to supraphysiologic α -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 pain—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, <sc>EVIDENCE</sc>. 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 Protoporphyrria: 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. <i>Molecular Genetics and Metabolism</i> , 2019, 128, 363-366.	1.1	9
39	Recent advances on porphyria genetics: Inheritance, penetrance & molecular heterogeneity, including new modifying/causative genes. <i>Molecular Genetics and Metabolism</i> , 2019, 128, 320-331.	1.1	59
40	Fabry disease revisited: Management and treatment recommendations for adult patients. <i>Molecular Genetics and Metabolism</i> , 2018, 123, 416-427.	1.1	391
41	Long-term Outcomes of Kidney Transplantation in Fabry Disease. <i>Transplantation</i> , 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. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2018, 115, E4071-E4080.	7.1	32
43	Fabry Disease: prevalence of affected males and heterozygotes with pathogenic <i>GLA</i> mutations identified by screening renal, cardiac and stroke clinics, 1995–2017. <i>Journal of Medical Genetics</i> , 2018, 55, 261-268.	3.2	91
44	Focal facial dermal dysplasia type 4: identification of novel <i>CYP26C1</i> mutations in unrelated patients. <i>Journal of Human Genetics</i> , 2018, 63, 257-261.	2.3	3
45	Functional variants in the <i>LRRK2</i> gene confer shared effects on risk for Crohn's disease and Parkinson's disease. <i>Science Translational Medicine</i> , 2018, 10, .	12.4	273
46	Identification and characterization of 40 novel hydroxymethylbilane synthase mutations that cause acute intermittent porphyria. <i>Journal of Inherited Metabolic Disease</i> , 2018, 42, 186.	3.6	9
47	Identification of Fabry Disease in a Tertiary Referral Cohort of Patients with Hypertrophic Cardiomyopathy. <i>American Journal of Medicine</i> , 2018, 131, 200.e1-200.e8.	1.5	31
48	Reply. <i>Hepatology</i> , 2018, 67, 803-804.	7.3	0
49	Newborn screening for lysosomal storage disorders by tandem mass spectrometry in North East Italy. <i>Journal of Inherited Metabolic Disease</i> , 2018, 41, 209-219.	3.6	114
50	Parkinson's disease prevalence in Fabry disease: A survey study. <i>Molecular Genetics and Metabolism Reports</i> , 2018, 14, 27-30.	1.1	34
51	Design and validation of an open-source modular Microplate Photoirradiation System for high-throughput photobiology experiments. <i>PLoS ONE</i> , 2018, 13, e0203597.	2.5	5
52	Many pitfalls in diagnosis of acute intermittent porphyria: a case report. <i>BMC Research Notes</i> , 2018, 11, 552.	1.4	10
53	Multi-ethnic <i>SULT1A1</i> copy number profiling with multiplex ligation-dependent probe amplification. <i>Pharmacogenomics</i> , 2018, 19, 761-770.	1.3	9
54	Roscoe Owen Brady, MD: Remembrances of co-investigators and colleagues. <i>Molecular Genetics and Metabolism</i> , 2017, 120, 1-7.	1.1	3

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55	Types A and B Niemann-Pick disease. <i>Molecular Genetics and Metabolism</i> , 2017, 120, 27-33.	1.1	196
56	Acute hepatic porphyrias: Recommendations for evaluation and long-term management. <i>Hepatology</i> , 2017, 66, 1314-1322.	7.3	122
57	Clinical, Biochemical, and Genetic Characterization of North American Patients With Erythropoietic Protoporphyrinemia and X-linked Protoporphyrinemia. <i>JAMA Dermatology</i> , 2017, 153, 789.	4.1	70
58	Institutional profile: translational pharmacogenomics at the Icahn School of Medicine at Mount Sinai. <i>Pharmacogenomics</i> , 2017, 18, 1381-1386.	1.3	20
59	Fabry disease: characterisation of the plasma proteome pre- and post-enzyme replacement therapy. <i>Journal of Medical Genetics</i> , 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 hypertrophy. <i>International Journal of Cardiology</i> , 2017, 248, 257-262.		13
61	Fabry Disease in Families With Hypertrophic Cardiomyopathy. <i>Circulation: Cardiovascular Genetics</i> , 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. <i>Molecular Genetics and Metabolism</i> , 2017, 121, 320-324.	1.1	50
63	The focal facial dermal dysplasias: phenotypic spectrum and molecular genetic heterogeneity. <i>Journal of Medical Genetics</i> , 2017, 54, 585-590.	3.2	10
64	Early manifestations of type 1 Gaucher disease in presymptomatic children diagnosed after parental carrier screening. <i>Genetics in Medicine</i> , 2017, 19, 652-658.	2.4	19
65	Plasma LysoGb3: A useful biomarker for the diagnosis and treatment of Fabry disease heterozygotes. <i>Molecular Genetics and Metabolism</i> , 2017, 120, 57-61.	1.1	99
66	The validation of pharmacogenetics for the identification of Fabry patients to be treated with migalastat. <i>Genetics in Medicine</i> , 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. <i>Gastroenterology</i> , 2016, 151, 710-723.e2.	1.3	51
68	Exome sequencing of extreme clopidogrel response phenotypes identifies <i>B4GALT2</i> as a determinant of on-treatment platelet reactivity. <i>Clinical Pharmacology and Therapeutics</i> , 2016, 100, 287-294.	4.7	22
69	Long-Read Single Molecule Real-Time Full Gene Sequencing of Cytochrome P450-2D6. <i>Human Mutation</i> , 2016, 37, 315-323.	2.5	86
70	Later Onset Fabry Disease, Cardiac Damage Progress in Silence. <i>Journal of the American College of Cardiology</i> , 2016, 68, 2554-2563.	2.8	81
71	Response to "Letter to the Editor by Drs Block and Razani". <i>Journal of Clinical Lipidology</i> , 2016, 10, 1281-1282.	1.5	2
72	Experiences and concerns of patients with recurrent attacks of acute hepatic porphyria: A qualitative study. <i>Molecular Genetics and Metabolism</i> , 2016, 119, 278-283.	1.1	60

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73	Xâ€chromosomal inactivation directly influences the phenotypic manifestation of Xâ€linked protoporphyria. <i>Clinical Genetics</i> , 2016, 89, 20-26.	2.0	25
74	Acute Intermittent Porphyria: Predicted Pathogenicity of <i>HMBS</i> Variants Indicates Extremely Low Penetrance of the Autosomal Dominant Disease. <i>Human Mutation</i> , 2016, 37, 1215-1222.	2.5	129
75	Acute Intermittent Porphyria in children: A case report and review of the literature. <i>Molecular Genetics and Metabolism</i> , 2016, 119, 295-299.	1.1	31
76	Genetics of GNE myopathy in the non-Jewish Persian population. <i>European Journal of Human Genetics</i> , 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. <i>Blood</i> , 2016, 128, 2318-2318.	1.4	3
79	The D519G Polymorphism of Glyceronephosphate O-Acyltransferase Is a Risk Factor for Familial Porphyria Cutanea Tarda. <i>PLoS ONE</i> , 2016, 11, e0163322.	2.5	7
80	Efficacy of Enzyme and Substrate Reduction Therapy with a Novel Antagonist of Glucosylceramide Synthase for Fabry Disease. <i>Molecular Medicine</i> , 2015, 21, 389-399.	4.4	72
81	Liver Transplantation for Acute Intermittent Porphyria: Biochemical and Pathologic Studies of the Explanted Liver. <i>Molecular Medicine</i> , 2015, 21, 487-495.	4.4	51
82	Pitfalls in Erythrocyte Protoporphyrin Measurement for Diagnosis and Monitoring of Protoporphyrins. <i>Clinical Chemistry</i> , 2015, 61, 1453-1456.	3.2	29
83	Setleis syndrome due to inheritance of the 1p36.22p36.21 duplication: evidence for lack of penetrance. <i>Journal of Human Genetics</i> , 2015, 60, 717-722.	2.3	12
84	Î±-Galactosidase A Knockout Mice. <i>American Journal of Pathology</i> , 2015, 185, 651-665.	3.8	34
85	Afamelanotide for Erythropoietic Protoporphyria. <i>New England Journal of Medicine</i> , 2015, 373, 48-59.	27.0	206
86	Ten-year outcome of enzyme replacement therapy with agalsidase beta in patients with Fabry disease. <i>Journal of Medical Genetics</i> , 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). <i>BMC Genomics</i> , 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 /Overlaid	1.2	15
90	Actionable exomic incidental findings in 6503 participants: challenges of variant classification. <i>Genome Research</i> , 2015, 25, 305-315.	5.5	313

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91	Preclinical Development of a Subcutaneous ALAS1 RNAi Therapeutic for Treatment of Hepatic Porphyrins Using Circulating RNA Quantification. <i>Molecular Therapy - Nucleic Acids</i> , 2015, 4, e263.	5.1	107
92	Setleis syndrome: clinical, molecular and structural studies of the first <sc>TWIST2</sc> missense mutation. <i>Clinical Genetics</i> , 2015, 88, 489-493.	2.0	12
93	RNAi-mediated silencing of hepatic <i>Alas1</i> effectively prevents and treats the induced acute attacks in acute intermittent porphyria mice. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 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. <i>Genetics in Medicine</i> , 2014, 16, 149-156.	2.4	64
95	Genome-wide mapping of IBD segments in an Ashkenazi PD cohort identifies associated haplotypes. <i>Human Molecular Genetics</i> , 2014, 23, 4693-4702.	2.9	49
96	Acute Porphyrins in the USA: Features of 108 Subjects from Porphyrins Consortium. <i>American Journal of Medicine</i> , 2014, 127, 1233-1241.	1.5	185
97	JCL Roundtable: Enzyme replacement therapy for lipid storage disorders. <i>Journal of Clinical Lipidology</i> , 2014, 8, 463-472.	1.5	3
98	Setleis Syndrome: Genetic and Clinical Findings in a New Case With Epilepsy. <i>Pediatric Neurology</i> , 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 CYP2C19 [∗] 4A, [∗] 4B, and [∗] 17 Alleles. <i>Journal of Molecular Diagnostics</i> , 2013, 15, 783-789.	2.8	17
101	Diagnosis of feline acute intermittent porphyria presenting with erythrodonia requires molecular analyses. <i>Veterinary Journal</i> , 2013, 198, 720-722.	1.7	8
102	Multi-ethnic distribution of clinically relevant CYP2C genotypes and haplotypes. <i>Pharmacogenomics Journal</i> , 2013, 13, 369-377.	2.0	87
103	Extended haplotype association study in Crohn's disease identifies a novel, Ashkenazi Jewish-specific missense mutation in the NF- κ B pathway gene, HEATR3. <i>Genes and Immunity</i> , 2013, 14, 310-316.	4.1	31
104	Focal facial dermal dysplasia, type IV, is caused by mutations in CYP26C1. <i>Human Molecular Genetics</i> , 2013, 22, 696-703.	2.9	41
105	Frequency of the cholesteryl ester storage disease common <i>LIPA</i>E8SJM mutation (c.894G>A) in various racial and ethnic groups. <i>Hepatology</i> , 2013, 58, 958-965.	7.3	85
106	Loss-of-Function Ferrochelatase and Gain-of-Function Erythroid-Specific 5-Aminolevulinat Synthase Mutations Causing Erythropoietic Protoporphyrin and X-Linked Protoporphyrin in North American Patients Reveal Novel Mutations and a High Prevalence of X-Linked Protoporphyrin. <i>Molecular Medicine</i> , 2013, 19, 26-29.	4.4	74
107	Molecular Expression and Characterization of Erythroid-Specific 5-Aminolevulinat Synthase Gain-of-Function Mutations Causing X-Linked Protoporphyrin. <i>Molecular Medicine</i> , 2013, 19, 18-25.	4.4	33
108	A Genome-Wide Scan of Ashkenazi Jewish Crohn's Disease Suggests Novel Susceptibility Loci. <i>PLoS Genetics</i> , 2012, 8, e1002559.	3.5	144

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109	Identification of CYP2C19*4B: pharmacogenetic implications for drug metabolism including clopidogrel responsiveness. <i>Pharmacogenomics Journal</i> , 2012, 12, 297-305.	2.0	40
110	The porphyrias: advances in diagnosis and treatment. <i>Blood</i> , 2012, 120, 4496-4504.	1.4	207
111	Copy number variation and warfarin dosing: evaluation of <i>CYP2C9</i> , <i>VKORC1</i> , <i>CYP4F2</i> and <i>GGCX</i> and <i>CALU</i> . <i>Pharmacogenomics</i> , 2012, 13, 297-307.	1.3	13
112	Enzyme Replacement Therapy for Lysosomal Diseases: Lessons from 20 Years of Experience and Remaining Challenges. <i>Annual Review of Genomics and Human Genetics</i> , 2012, 13, 307-335.	6.2	229
113	Fabry Disease: Incidence of the Common Later-Onset α -Galactosidase A IVS4+919G>A Mutation in Taiwanese Newborns—Superiority of DNA-Based to Enzyme-Based Newborn Screening for Common Mutations. <i>Molecular Medicine</i> , 2012, 18, 780-784.	4.4	71
114	The porphyrias: advances in diagnosis and treatment. <i>Hematology American Society of Hematology Education Program</i> , 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. <i>Journal of Medical Genetics</i> , 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. <i>Clinical Pharmacology and Therapeutics</i> , 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. <i>International Journal of Biochemistry and Cell Biology</i> , 2011, 43, 1523-1531.	2.8	18
118	Diagnostic dilemma: A young woman with Fabry disease symptoms, no family history, and a α -galactosidase a large deletion. <i>Molecular Genetics and Metabolism</i> , 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. <i>Molecular Genetics and Metabolism</i> , 2011, 104, 700-702.	1.1	8
120	CYP1A2*1F and GSTM1 Alleles Are Associated with Susceptibility to Porphyria Cutanea Tarda. <i>Molecular Medicine</i> , 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. <i>Molecular Medicine</i> , 2011, 17, 748-756.	4.4	10
122	Harderoporphyria due to homozygosity for coproporphyrinogen oxidase missense mutation H327R. <i>Journal of Inherited Metabolic Disease</i> , 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. <i>Journal of Inherited Metabolic Disease</i> , 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. <i>BMC Medical Genetics</i> , 2011, 12, 63.	2.1	41
125	Detection of large gene rearrangements in X-linked genes by dosage analysis: identification of novel α -galactosidase A (GLA) deletions causing Fabry disease. <i>Human Mutation</i> , 2011, 32, 688-695.	2.5	13
126	A LC-MS/MS method for the specific, sensitive, and simultaneous quantification of 5-aminolevulinic acid and porphobilinogen. <i>Journal of Chromatography B: Analytical Technologies in the Biomedical and Life Sciences</i> , 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. <i>Journal of Human Genetics</i> , 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. <i>Blood</i> , 2010, 115, 1062-1069.	1.4	15
129	Homozygous Nonsense Mutations in TWIST2 Cause Setleis Syndrome. <i>American Journal of Human Genetics</i> , 2010, 87, 289-296.	6.2	65
130	Experience with carrier screening and prenatal diagnosis for 16 Ashkenazi Jewish genetic diseases. <i>Human Mutation</i> , 2010, 31, 1240-1250.	2.5	125
131	Feline Congenital Erythropoietic Porphyria: Two Homozygous UROS Missense Mutations Cause the Enzyme Deficiency and Porphyrin Accumulation. <i>Molecular Medicine</i> , 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. <i>Human Molecular Genetics</i> , 2010, 19, 584-596.	2.9	32
133	The Pharmacological Chaperone 1-Deoxygalactonojirimycin Reduces Tissue Globotriaosylceramide Levels in a Mouse Model of Fabry Disease. <i>Molecular Therapy</i> , 2010, 18, 23-33.	8.2	126
134	Combined <i>CYP2C9</i> , <i>VKORC1</i> and <i>CYP4F2</i> frequencies among racial and ethnic groups. <i>Pharmacogenomics</i> , 2010, 11, 781-791.	1.3	146
135	Hepatoerythropoietic Porphyria Misdiagnosed as Child Abuse. <i>Archives of Dermatology</i> , 2010, 146, 529-33.	1.4	11
136	AAV8-mediated Gene Therapy Prevents Induced Biochemical Attacks of Acute Intermittent Porphyria and Improves Neuromotor Function. <i>Molecular Therapy</i> , 2010, 18, 17-22.	8.2	52
137	Type 1 Gaucher Disease. <i>Archives of Internal Medicine</i> , 2010, 170, 1463-9.	3.8	58
138	Tamoxifen Metabolite Isomer Separation and Quantification by Liquid Chromatographyâˆ“Tandem Mass Spectrometry. <i>Analytical Chemistry</i> , 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. <i>Molecular Genetics and Metabolism</i> , 2010, 101, 130-133.	1.1	52
140	Newborn screening for Fabry disease in Taiwan reveals a high incidence of the later-onset <i>GLA</i> mutation c.936+919G>A (IVS4+919G>A). <i>Human Mutation</i> , 2009, 30, 1397-1405.	2.5	299
141	Fabry disease: progression of nephropathy, and prevalence of cardiac and cerebrovascular events before enzyme replacement therapy. <i>Nephrology Dialysis Transplantation</i> , 2009, 24, 2102-2111.	0.7	297
142	Genomics and personalized medicine: a perspective. <i>Personalized Medicine</i> , 2009, 6, 135-137.	1.5	0
143	Identification and Characterization of Feline Acute Intermittent Porphyria: The First Naturally-Occurring Animal Model.. <i>Blood</i> , 2009, 114, 3014-3014.	1.4	0
144	Human uroporphyrinogen III synthase: NMR-based mapping of the active site. <i>Proteins: Structure, Function and Bioinformatics</i> , 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. <i>American Journal of Human Genetics</i> , 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. <i>Journal of the American Society of Nephrology: JASN</i> , 2007, 18, 1547-1557.	6.1	396
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