Robert Desnick

List of Publications by Year
in descending order

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	Safety and Efficacy of Recombinant Human Î±-Galactosidase A Replacement Therapy in Fabry's Disease. New England Journal of Medicine, 2001, 345, 9-16.	27.0	1,433
2	High Incidence of Later-Onset Fabry Disease Revealed by Newborn Screening*. American Journal of Human Genetics, 2006, 79, 31-40.	6.2	842
3	Fabry Disease, an Under-Recognized Multisystemic Disorder: Expert Recommendations for Diagnosis, Management, and Enzyme Replacement Therapy. Annals of Internal Medicine, 2003, 138, 338.	3.9	619
4	Agalsidase-Beta Therapy for Advanced Fabry Disease. Annals of Internal Medicine, 2007, 146, 77.	3.9	493
5	Recommendations for the Diagnosis and Treatment of the Acute Porphyrias. Annals of Internal Medicine, 2005, 142, 439.	3.9	485
6	Laboratory standards and guidelines for population-based cystic fibrosis carrier screening. Genetics in Medicine, 2001, 3, 149-154.	2.4	440
7	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
8	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
9	Long-Term Safety and Efficacy of Enzyme Replacement Therapyfor Fabry Disease. American Journal of Human Genetics, 2004, 75, 65-74.	6.2	394
10	Fabry disease revisited: Management and treatment recommendations for adult patients. Molecular Genetics and Metabolism, 2018, 123, 416-427.	1.1	391
11	Fabry disease: Detection of undiagnosed hemodialysis patients and identification of a â€œrenal variantâ€• phenotype1. Kidney International, 2003, 64, 801-807.	5.2	368
12	A Phase 1/2 Clinical Trial of Enzyme Replacement in Fabry Disease: Pharmacokinetic, Substrate Clearance, and Safety Studies. American Journal of Human Genetics, 2001, 68, 711-722.	6.2	350
13	An Atypical Variant of Fabry's Disease with Manifestations Confined to the Myocardium. New England Journal of Medicine, 1991, 324, 395-399.	27.0	342
14	Actionable exomic incidental findings in 6503 participants: challenges of variant classification. Genome Research, 2015, 25, 305-315.	5.5	313
15	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
16	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
17	Globotriaosylceramide accumulation in the Fabry kidney is cleared from multiple cell types after enzyme replacement therapy. Kidney International, 2002, 62, 1933-1946.	5.2	291
18	Mutation of the matrix metalloproteinase 2 gene (MMP2) causes a multicentric osteolysis and arthritis syndrome. Nature Genetics, 2001, 28, 261-265.	21.4	277

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19	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
20	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
21	Enzyme replacement and enhancement therapies: lessons from lysosomal disorders. Nature Reviews Genetics, 2002, 3, 954-966.	16.3	254
22	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
23	The porphyrias: advances in diagnosis and treatment. Blood, 2012, 120, 4496-4504.	1.4	207
24	Afamelanotide for Erythropoietic Protoporphyria. New England Journal of Medicine, 2015, 373, 48-59.	27.0	206
25	Mutations in a new gene encoding a thiamine transporter cause thiamine-responsive megaloblastic anaemia syndrome. Nature Genetics, 1999, 22, 309-312.	21.4	201
26	Bone-Marrow Transplantation in the Maroteauxâ€“Lamy Syndrome (Mucopolysaccharidosis Type VI). New England Journal of Medicine, 1984, 311, 1606-1611.	27.0	199
27	Types A and B Niemann-Pick disease. Molecular Genetics and Metabolism, 2017, 120, 27-33.	1.1	196
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	Acute Porphyrias in the USA: Features of 108ÂSubjects from Porphyrias Consortium. American Journal of Medicine, 2014, 127, 1233-1241.	1.5	185
30	Fabry Disease: Preclinical Studies Demonstrate the Effectiveness of Î±-Galactosidase A Replacement in Enzyme-Deficient Mice. American Journal of Human Genetics, 2001, 68, 14-25.	6.2	182
31	Nature and frequency of mutations in the alpha-galactosidase A gene that cause Fabry disease. American Journal of Human Genetics, 1993, 53, 1186-97.	6.2	181
32	Fabry's disease: enzymatic diagnosis of hemizygotes and heterozygotes. Alpha-galactosidase activities in plasma, serum, urine, and leukocytes. Translational Research, 1973, 81, 157-71.	2.3	174
33	Purification and properties of delta-aminolevulinate dehydrase from human erythrocytes.. Journal of Biological Chemistry, 1979, 254, 6924-6930.	3.4	165
34	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
35	Lead Binding to Î´â€Aminolevulinic Acid Dehydratase (ALAD) in Human Erythrocytes. Basic and Clinical Pharmacology and Toxicology, 1997, 81, 153-158.	0.0	155
36	The Ocular Manifestations in Fabry's Disease. JAMA Ophthalmology, 1979, 97, 671-676.	2.4	154

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37	Cardiac valvular anomalies in Fabry disease. Clinical, morphologic, and biochemical studies.. Circulation, 1976, 54, 818-825.	1.6	151
38	Fabry disease: six gene rearrangements and an exonic point mutation in the alpha-galactosidase gene.. Journal of Clinical Investigation, 1989, 83, 1390-1399.	8.2	149
39	Nucleotide sequence of the human Î±-galactosidase A gene. Nucleic Acids Research, 1989, 17, 3301-3302.	14.5	148
40	Structural organization of the human alpha-galactosidase A gene: further evidence for the absence of a 3' untranslated region.. Proceedings of the National Academy of Sciences of the United States of America, 1988, 85, 3903-3907.	7.1	147
41	Alternative Splicing in the Î±-Galactosidase A Gene: Increased Exon Inclusion Results in the Fabry Cardiac Phenotype. American Journal of Human Genetics, 2002, 70, 994-1002.	6.2	146
42	Combined CYP2C9, VKORC1 and CYP4F2 frequencies among racial and ethnic groups. Pharmacogenomics, 2010, 11, 781-791.	1.3	146
43	Schindler disease: An inherited neuroaxonal dystrophy due to Î±-N-acetylgalactosaminidase deficiency. Journal of Inherited Metabolic Disease, 1990, 13, 549-559.	3.6	145
44	A Genome-Wide Scan of Ashkenazi Jewish Crohn's Disease Suggests Novel Susceptibility Loci. PLoS Genetics, 2012, 8, e1002559.	3.5	144
45	A Mutation ofPCDH15among Ashkenazi Jews with the Type 1 Usher Syndrome. New England Journal of Medicine, 2003, 348, 1664-1670.	27.0	142
46	Purification and properties of delta-aminolevulinate dehydrase from human erythrocytes. Journal of Biological Chemistry, 1979, 254, 6924-30.	3.4	142
47	Toward enzyme therapy for lysosomal storage diseases. Physiological Reviews, 1976, 56, 57-99.	28.8	139
48	Overexpression of human alpha-galactosidase A results in its intracellular aggregation, crystallization in lysosomes, and selective secretion.. Journal of Cell Biology, 1992, 119, 1137-1150.	5.2	137
49	Fabry disease: Characterization of ?-galactosidase A double mutations and the D313Y plasma enzyme pseudodeficiency allele. Human Mutation, 2003, 22, 486-492.	2.5	133
50	Molecular basis of fabry disease: Mutations and polymorphisms in the human Î±-galactosidase A gene. Human Mutation, 1994, 3, 103-111.	2.5	132
51	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
52	Acute Intermittent Porphyria: Predicted Pathogenicity ofHMBSVariants Indicates Extremely Low Penetrance of the Autosomal Dominant Disease. Human Mutation, 2016, 37, 1215-1222.	2.5	129
53	Determination of Bone Markers in Pycnodysostosis: Effects of Cathepsin K Deficiency on Bone Matrix Degradation. Journal of Bone and Mineral Research, 1999, 14, 1902-1908.	2.8	128
54	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

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55	Infusion of recombinant human acid sphingomyelinase into Niemannâ€Pick disease mice leads to visceral, but not neurological, correction of the pathophysiology. FASEB Journal, 2000, 14, 1988-1995.	0.5	126
56	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
57	Experience with carrier screening and prenatal diagnosis for 16 Ashkenazi Jewish genetic diseases. Human Mutation, 2010, 31, 1240-1250.	2.5	125
58	Fabry Disease: Thirty-Five Mutations in the Î±-Galactosidase A Gene in Patients with Classic and Variant Phenotypes. Molecular Medicine, 1997, 3, 174-182.	4.4	124
59	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
60	Acute hepatic porphyrias: Recommendations for evaluation and longâ€term management. Hepatology, 2017, 66, 1314-1322.	7.3	122
61	Glutamate dehydrogenase deficiency in three patients with spinocerebellar syndrome. Annals of Neurology, 1980, 7, 297-303.	5.3	116
62	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
63	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
64	Congenital erythropoietic porphyria: advances in pathogenesis and treatment. British Journal of Haematology, 2002, 117, 779-795.	2.5	108
65	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
66	Purification and properties of uroporphyrinogen I synthase from human erythrocytes. Identification of stable enzyme-substrate intermediates. Journal of Biological Chemistry, 1980, 255, 1993-9.	3.4	107
67	EXPLORE: A Prospective, Multinational, Natural History Study of Patients with Acute Hepatic Porphyria with Recurrent Attacks. Hepatology, 2020, 71, 1546-1558.	7.3	103
68	Alpha-galactosidase A gene rearrangements causing Fabry disease. Identification of short direct repeats at breakpoints in an Alu-rich gene. Journal of Biological Chemistry, 1990, 265, 9319-26.	3.4	103
69	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
70	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
71	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
72	Genetic homogeneity and phenotypic variability among Ashkenazi Jews with Usher syndrome type III. Journal of Medical Genetics, 2003, 40, 767-772.	3.2	97

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73	Alpha-L-iduronidase Deficiency in a Cat: A Model of Mucopolysaccharidosis I. Pediatric Research, 1979, 13, 1294-1297.	2.3	96
74	Fabry disease in childhood. Journal of Pediatrics, 2004, 144, S20-S26.	1.8	96
75	Human uroporphyrinogen III synthase: molecular cloning, nucleotide sequence, and expression of a full-length cDNA.. Proceedings of the National Academy of Sciences of the United States of America, 1988, 85, 7049-7053.	7.1	94
76	Linkage of pycnodysostosis to chromosome 1q21 by homozygosity mapping. Nature Genetics, 1995, 10, 235-237.	21.4	94
77	Bone density in type 1 gaucher disease. Journal of Bone and Mineral Research, 1996, 11, 1801-1807.	2.8	94
78	Human Î±-galactosidase A: glycosylation site 3 is essential for enzyme solubility. Biochemical Journal, 1998, 332, 789-797.	3.7	92
79	Acute Intermittent Porphyria. Archives of Neurology, 2004, 61, 1764.	4.5	92
80	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
81	Multi-ethnic distribution of clinically relevant CYP2C genotypes and haplotypes. Pharmacogenomics Journal, 2013, 13, 369-377.	2.0	87
82	Fabry disease: 45 novel mutations in the Î±-galactosidase A gene causing the classical phenotype. Molecular Genetics and Metabolism, 2002, 76, 23-30.	1.1	86
83	Long-Read Single Molecule Real-Time Full Gene Sequencing of Cytochrome P450-2D6. Human Mutation, 2016, 37, 315-323.	2.5	86
84	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
85	Uroporphyrinogen III synthase erythroid promoter mutations in adjacent GATA1 and CP2 elements cause congenital erythropoietic porphyria. Journal of Clinical Investigation, 2001, 107, 753-762.	8.2	85
86	Congenital erythropoietic porphyria: identification and expression of 10 mutations in the uroporphyrinogen III synthase gene.. Journal of Clinical Investigation, 1995, 95, 905-912.	8.2	85
87	Later Onset Fabry Disease, Cardiac Damage Progress in Silence. Journal of the American College of Cardiology, 2016, 68, 2554-2563.	2.8	81
88	Carrier Frequency of the Bloom SyndromeblmAshMutation in the Ashkenazi Jewish Population. Molecular Genetics and Metabolism, 1998, 64, 286-290.	1.1	79
89	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
90	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

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91	Gastrointestinal manifestations of Fabry disease: Clinical response to enzyme replacement therapy. Molecular Genetics and Metabolism, 2005, 85, 255-259.	1.1	73
92	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
93	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
94	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
95	Clinical, Biochemical, and Genetic Characterization of North American Patients With Erythropoietic Protoporphyria and X-linked Protoporphyria. JAMA Dermatology, 2017, 153, 789.	4.1	70
96	Electrocardiographic and vectorcardiographic abnormalities in Fabry's disease. American Heart Journal, 1977, 93, 699-705.	2.7	69
97	Congenital erythropoietic porphyria: identification and expression of exonic mutations in the uroporphyrinogen III synthase gene.. Journal of Clinical Investigation, 1992, 89, 693-700.	8.2	69
98	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
99	Homozygous Nonsense Mutations in TWIST2 Cause Setleis Syndrome. American Journal of Human Genetics, 2010, 87, 289-296.	6.2	65
100	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
101	Acute intermittent porphyria: identification and expression of exonic mutations in the hydroxymethylbilane synthase gene. An initiation codon missense mutation in the housekeeping transcript causes "variant acute intermittent porphyria" with normal expression of the erythroid-specific enzyme.. Journal of Clinical Investigation, 1994, 94, 1927-1937.	8.2	64
102	Treatment of severe congenital erythropoietic porphyria by bone marrow transplantation. Journal of the American Academy of Dermatology, 2001, 45, 279-282.	1.2	63
103	Delta-Aminolevulinic Acid Dehydratase Polymorphism: Influence on Lead Levels and Kidney Function in Humans. Archives of Environmental Health, 1997, 52, 91-96.	0.4	60
104	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
105	Inherited multicentric osteolysis with arthritis: A variant resembling Torg syndrome in a Saudi family. American Journal of Medical Genetics Part A, 2000, 93, 11-18.	2.4	59
106	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
107	Type 1 Gaucher Disease. Archives of Internal Medicine, 2010, 170, 1463-9.	3.8	58
108	Regional assignment of the human uroporphyrinogen III synthase (UROS) gene to chromosome 10q25.2?q26.3. Human Genetics, 1991, 87, 18-22.	3.8	56

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109	Human Uroporphyrinogen-III Synthase: Genomic Organization, Alternative Promoters, and Erythroid-Specific Expression. Genomics, 2000, 70, 223-231.	2.9	56
110	Familial dysautonomia: Detection of theIKBKAP IVS20+6T ? C and R696P mutations and frequencies among Ashkenazi Jews. American Journal of Medical Genetics Part A, 2002, 110, 253-257.	2.4	56
111	Molecular Genetics of Congenital Erythropoietic Porphyria. Seminars in Liver Disease, 1998, 18, 77-84.	3.6	52
112	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
113	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
114	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
115	Familial Porphyria Cutanea Tarda: Characterization of Seven Novel Uroporphyrinogen Decarboxylase Mutations and Frequency of Common Hemochromatosis Alleles. American Journal of Human Genetics, 1998, 63, 1363-1375.	6.2	51
116	Detection of ?-galactosidase a mutations causing fabry disease by denaturing high performance liquid chromatography. Human Mutation, 2005, 25, 299-305.	2.5	51
117	Liver Transplantation for Acute Intermittent Porphyria: Biochemical and Pathologic Studies of the Explanted Liver. Molecular Medicine, 2015, 21, 487-495.	4.4	51
118	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
119	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
120	Genome-wide mapping of IBD segments in an Ashkenazi PD cohort identifies associated haplotypes. Human Molecular Genetics, 2014, 23, 4693-4702.	2.9	49
121	Coupled-enzyme and direct assays for uroporphyrinogen III synthase activity in human erythrocytes and cultured lymphoblasts. Analytical Biochemistry, 1987, 166, 120-133.	2.4	48
122	Reduced plasma concentrations of total, low density lipoprotein and high density lipoprotein cholesterol in patients with Gaucher type I disease. Clinical Genetics, 1984, 26, 109-116.	2.0	47
123	Nonâ€“pseudogene-derived complex acid Î²-glucosidase mutations causing mild type 1 and severe type 2 Gaucher disease. Journal of Clinical Investigation, 1999, 103, 817-823.	8.2	46
124	Congenital erythropoietic porphyria: Recent advances. Molecular Genetics and Metabolism, 2019, 128, 288-297.	1.1	45
125	Purification and properties of uroporphyrinogen III synthase from human erythrocytes.. Journal of Biological Chemistry, 1987, 262, 1268-1273.	3.4	45
126	Type A Niemann-Pick disease: A frameshift mutation in the acid sphingomyelinase gene (fsP330) occurs in Ashkenazi Jewish patients. Human Mutation, 1993, 2, 317-319.	2.5	44

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127	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
128	CYP2C9,CYP2C19andCYP2D6allele frequencies in the Ashkenazi Jewish population. Pharmacogenomics, 2007, 8, 721-730.	1.3	43
129	Genetic heterogeneity in acute intermittent porphyria: characterisation and frequency of porphobilinogen deaminase mutations in Finland.. BMJ: British Medical Journal, 1985, 291, 505-509.	2.3	41
130	Uroporphyrinogen III Synthase. Journal of Biological Chemistry, 2000, 275, 2295-2304.	3.4	41
131	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
132	Focal facial dermal dysplasia, type IV, is caused by mutations in CYP26C1. Human Molecular Genetics, 2013, 22, 696-703.	2.9	41
133	Purification and properties of uroporphyrinogen III synthase from human erythrocytes. Journal of Biological Chemistry, 1987, 262, 1268-73.	3.4	41
134	Identification of CYP2C19*4B: pharmacogenetic implications for drug metabolism including clopidogrel responsiveness. Pharmacogenomics Journal, 2012, 12, 297-305.	2.0	40
135	Genetic mapping of the cleidocranial dysplasia (CCD) locus on chromosome band 6p21 to include a microdeletion. American Journal of Medical Genetics Part A, 1995, 58, 200-205.	2.4	39
136	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
137	Molecular basis of congenital erythropoietic porphyria: Mutations in the human uroporphyrinogen III synthase gene. Human Mutation, 1996, 7, 187-192.	2.5	36
138	Malignant fibrous histiocytoma: Inherited and sporadic forms have loss of heterozygosity at chromosome bands 9p21-22?evidence for a common genetic defect. Genes Chromosomes and Cancer, 2000, 27, 191-195.	2.8	36
139	Prenatal diagnosis of Fabry disease. Prenatal Diagnosis, 2007, 27, 693-694.	2.3	36
140	Identification and characterization of hydroxymethylbilane synthase mutations causing acute intermittent porphyria: Evidence for an ancestral founder of the common G111R mutation. , 1999, 86, 366-375.		35
141	Carrier Screening for Mucolipidosis Type IV in the American Ashkenazi Jewish Population. American Journal of Human Genetics, 2002, 70, 1023-1027.	6.2	35
142	Congenital erythropoietic porphyria: identification and expression of eight novel mutations in the uroporphyrinogen III synthase gene. British Journal of Haematology, 2002, 117, 980-987.	2.5	35
143	Î±-Galactosidase A Knockout Mice. American Journal of Pathology, 2015, 185, 651-665.	3.8	34
144	Parkinson's disease prevalence in Fabry disease: A survey study. Molecular Genetics and Metabolism Reports, 2018, 14, 27-30.	1.1	34

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145	The porphyrias: advances in diagnosis and treatment. Hematology American Society of Hematology Education Program, 2012, 2012, 19-27.	2.5	34
146	Gaucher Type 1 (Ashkenazi) disease: a new method for heterozygote detection using a novel fluorescent natural substrate. Clinica Chimica Acta, 1982, 124, 123-135.	1.1	33
147	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
148	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
149	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
150	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
151	Extended haplotype association study in Crohnâ€™s disease identifies a novel, Ashkenazi Jewish-specific missense mutation in the NF-Î²B pathway gene, HEATR3. Genes and Immunity, 2013, 14, 310-316.	4.1	31
152	Acute Intermittent Porphyria in children: A case report and review of the literature. Molecular Genetics and Metabolism, 2016, 119, 295-299.	1.1	31
153	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
154	Identification of a missense mutation (S436R) in the acid sphingomyelinase gene from a Japanese patient with type B Niemann-Pick disease. Human Mutation, 1992, 1, 70-71.	2.5	30
155	Cystic fibrosis population carrier screening: Here at lastâ€”Are we ready?. Genetics in Medicine, 2001, 3, 87-90.	2.4	29
156	Pitfalls in Erythrocyte Protoporphyrin Measurement for Diagnosis and Monitoring of Protoporphyrias. Clinical Chemistry, 2015, 61, 1453-1456.	3.2	29
157	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
158	Fabry disease: Detection of gene rearrangements in the human Î±-Galactosidase A Gene by Multiplex PCR Amplification. Human Mutation, 1993, 2, 108-111.	2.5	28
159	Tamoxifen Metabolite Isomer Separation and Quantification by Liquid Chromatographyâ€“Tandem Mass Spectrometry. Analytical Chemistry, 2010, 82, 10186-10193.	6.5	28
160	Molecular basis of acute intermittent porphyria: Mutations and polymorphisms in the human hydroxymethylbilane synthase gene. Human Mutation, 1994, 4, 243-252.	2.5	27
161	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
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