Dominique Germain

List of Publications by Year
in descending order

146	12,642	31902 53	24915 109
papers	citations	h-index	g-index

174	174	174	6407
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Diagnosis and Management of Cardiovascular Involvement in Fabry Disease. Heart Failure Clinics, 2022, 18, 39-49.	1.0	18
2	Challenging the traditional approach for interpreting genetic variants: Lessons from Fabry disease. Clinical Genetics, 2022, 101, 390-402.	1.0	26
3	The benefits and challenges of family genetic testing in rare genetic diseasesâ€"lessons from Fabry disease. Molecular Genetics & Genomic Medicine, 2021, 9, e1666.	0.6	26
4	Case Report: First Two Identified Cases of Fabry Disease in Central Asia. Frontiers in Genetics, 2021, 12, 657824.	1.1	4
5	Newborn Screening for Fabry Disease in Northeastern Italy: Results of Five Years of Experience. Biomolecules, 2021, 11, 951.	1.8	20
6	Screening for Fabry disease in male patients with end-stage renal disease in western France. Nephrologie Et Therapeutique, 2021, 17, 180-184.	0.2	9
7	Humoral Immune Response to SARS-CoV-2 Vaccination after a Booster Vaccine Dose in Two Kidney Transplant Recipients with Fabry Disease and Variable Secondary Immunosuppressive Regimens. Vaccines, 2021, 9, 1412.	2.1	3
8	Fabry disease caused by the GLA p.Phe113Leu (p.F113L) variant: Natural history in males. European Journal of Medical Genetics, 2020, 63, 103703.	0.7	21
9	IN VIVO OBSERVATION OF RETINAL VASCULAR DEPOSITS USING ADAPTIVE OPTICS IMAGING IN FABRY DISEASE. Retina, 2020, 40, 1623-1629.	1.0	4
10	MO035HISTORICAL CONTROL ANALYSIS DEMONSTRATES SUPERIOR REDUCTION OF PLASMA GLOBOTRIAOSYLCERAMIDE BY VENGLUSTAT COMPARED WITH PLACEBO OR AGALSIDASE BETA IN CLASSIC FABRY DISEASE PATIENTS. Nephrology Dialysis Transplantation, 2020, 35, .	0.4	2
11	Improvement of gastrointestinal symptoms in a significant proportion of male patients with classic Fabry disease treated with agalsidase beta: A Fabry Registry analysis stratified by phenotype. Molecular Genetics and Metabolism Reports, 2020, 25, 100670.	0.4	6
12	An expert consensus document on the management of cardiovascular manifestations of Fabry disease. European Journal of Heart Failure, 2020, 22, 1076-1096.	2.9	96
13	Cardiomyopathy and kidney function in agalsidase betaâ€treated female Fabry patients: a preâ€treatment vs. postâ€treatment analysis. ESC Heart Failure, 2020, 7, 825-834.	1.4	13
14	Use of a rare disease registry for establishing phenotypic classification of previously unassigned GLA variants: a consensus classification system by a multispecialty Fabry disease genotypeâ€"phenotype workgroup. Journal of Medical Genetics, 2020, 57, 542-551.	1.5	43
15	Fabry disease and COVID-19: international expert recommendations for management based on real-world experience. CKJ: Clinical Kidney Journal, 2020, 13, 913-925.	1.4	11
16	The effect of enzyme replacement therapy on clinical outcomes in paediatric patients with Fabry disease â€" A systematic literature review by a European panel of experts. Molecular Genetics and Metabolism, 2019, 126, 212-223.	0.5	50
17	Therapeutic goals in Fabry disease: Recommendations of a European expert panel, based on current clinical evidence with enzyme replacement therapy. Molecular Genetics and Metabolism, 2019, 126, 210-211.	0.5	13
18	Response to Gurevich and colleagues: The effect of enzyme replacement therapy on clinical outcomes in male patients with Fabry disease: a systematic literature review by a European panel of experts. Molecular Genetics and Metabolism Reports, 2019, 20, 100493.	0.4	0

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19	Semen and male genital tract characteristics of patients with Fabry disease: the FERTIFABRY multicentre observational study. Basic and Clinical Andrology, 2019, 29, 7.	0.8	5
20	Why systematic literature reviews in Fabry disease should include all published evidence. European Journal of Medical Genetics, 2019, 62, 103702.	0.7	12
21	Atypical COL3A1 variants (glutamic acid to lysine) cause vascular Ehlersâ€“Danlos syndrome with a consistent phenotype of tissue fragility and skin hyperextensibility. Genetics in Medicine, 2019, 21, 2081-2091.	1.1	16
22	Consensus recommendations for diagnosis, management and treatment of Fabry disease in paediatric patients. Clinical Genetics, 2019, 96, 107-117.	1.0	87
23	Efficacy of the pharmacologic chaperone migalastat in a subset of male patients with the classic phenotype of Fabry disease and migalastat-amenable variants: data from the phase 3 randomized, multicenter, double-blind clinical trial and extension study. Genetics in Medicine, 2019, 21, 1987-1997.	1.1	66
24	Fabry disease in cardiology practice: Literature review and expert point of view. Archives of Cardiovascular Diseases, 2019, 112, 278-287.	0.7	69
25	The effect of enzyme replacement therapy on clinical outcomes in male patients with Fabry disease: A systematic literature review by a European panel of experts. Molecular Genetics and Metabolism Reports, 2019, 19, 100454.	0.4	120
26	Treatment needs and expectations for Fabry disease in France: development of a new Patient Needs Questionnaire. Orphanet Journal of Rare Diseases, 2019, 14, 284.	1.2	6
27	The effect of enzyme replacement therapy on clinical outcomes in female patients with Fabry disease â€“ A systematic literature review by a European panel of experts. Molecular Genetics and Metabolism, 2019, 126, 224-235.	0.5	65
28	Fabry disease revisited: Management and treatment recommendations for adult patients. Molecular Genetics and Metabolism, 2018, 123, 416-427.	0.5	391
29	Phenotypic characteristics of the p.Asn215Ser (p.N215S) G<scp>LA</scp> mutation in male and female patients with Fabry disease: A multicenter Fabry Registry study. Molecular Genetics & Genomic Medicine, 2018, 6, 492-503.	0.6	70
30	A survivor analysis for major clinical events in heterozygous female patients with Fabry disease using group consensus phenotype classifications from hemizygous male patients. Molecular Genetics and Metabolism, 2018, 123, S65-S66.	0.5	0
31	The role of CNVs in the etiology of rare autosomal recessive disorders: the example of TRAPPC9-associated intellectual disability. European Journal of Human Genetics, 2018, 26, 143-148.	1.4	26
32	Principles of Human Genetics and Mendelian Inheritance. , 2018, , 1-28.		5
33	European expert consensus statement on therapeutic goals in Fabry disease. Molecular Genetics and Metabolism, 2018, 124, 189-203.	0.5	122
34	Mutation spectrum in the ABCC6 gene and genotypeâ€“phenotype correlations in a French cohort with pseudoxanthoma elasticum. Genetics in Medicine, 2017, 19, 909-917.	1.1	54
35	Basilar Artery Changes in Fabry Disease. American Journal of Neuroradiology, 2017, 38, 531-536.	1.2	39
36	Oral pharmacological chaperone migalastat compared with enzyme replacement therapy in Fabry disease: 18-month results from the randomised phase III ATTRACT study. Journal of Medical Genetics, 2017, 54, 288-296.	1.5	262

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37	Improvement of Fabry Disease-Related Gastrointestinal Symptoms in a Significant Proportion of Female Patients Treated with Agalsidase Beta: Data from the Fabry Registry. JIMD Reports, 2017, 38, 45-51.	0.7	18
38	Efficacy and safety of migalastat, an oral pharmacologic chaperone for Fabry disease: results from two randomized phase 3 studies, FACETS and ATTRACT. Molecular Genetics and Metabolism, 2017, 120, S45-S46.	0.5	1
39	The phenotypic characteristics of the p.N215S Fabry disease genotype in male and female patients: a multi-center Fabry Registry study. Molecular Genetics and Metabolism, 2017, 120, S51-S52.	0.5	3
40	Clinical utility gene card for: Fabry disease â€“ update 2016. European Journal of Human Genetics, 2017, 25, e1-e1.	1.4	10
41	Pseudoxanthoma elasticum. Orphanet Journal of Rare Diseases, 2017, 12, 85.	1.2	118
42	The validation of pharmacogenetics for the identification of Fabry patients to be treated with migalastat. Genetics in Medicine, 2017, 19, 430-438.	1.1	157
43	Fabry disease: Four case reports of meningioma and a review of the literature on other malignancies. Molecular Genetics and Metabolism Reports, 2017, 11, 75-80.	0.4	5
44	LC-MS/MS multiplex analysis of lysosphingolipids in plasma and amniotic fluid: A novel tool for the screening of sphingolipidoses and Niemann-Pick type C disease. PLoS ONE, 2017, 12, e0181700.	1.1	55
45	Time to treatment benefit for adult patients with Fabry disease receiving agalsidase Î²: data from the Fabry Registry. Journal of Medical Genetics, 2016, 53, 495-502.	1.5	101
46	Determinants of white matter hyperintensity burden in patients with Fabry disease. Neurology, 2016, 86, 1880-1886.	1.5	42
47	Treatment of Fabryâ€™s Disease with the Pharmacologic Chaperone Migalastat. New England Journal of Medicine, 2016, 375, 545-555.	13.9	390
48	Pain in Fabry Disease: Practical Recommendations for Diagnosis and Treatment. CNS Neuroscience and Therapeutics, 2016, 22, 568-576.	1.9	75
49	Xâ€chromosome inactivation in female patients with Fabry disease. Clinical Genetics, 2016, 89, 44-54.	1.0	290
50	Wholeâ€body muscle magnetic resonance imaging in SEPN1â€related myopathy shows a homogeneous and recognizable pattern. Muscle and Nerve, 2015, 52, 728-735.	1.0	47
51	Enzymatic diagnosis of Fabry disease using a fluorometric assay on dried blood spots: An alternative methodology. European Journal of Medical Genetics, 2015, 58, 681-684.	0.7	12
52	Raynaudâ€™s phenomenon associated with Fabry disease. Journal of Inherited Metabolic Disease, 2015, 38, 367-368.	1.7	5
53	Recommendations for initiation and cessation of enzyme replacement therapy in patients with Fabry disease: the European Fabry Working Group consensus document. Orphanet Journal of Rare Diseases, 2015, 10, 36.	1.2	239
54	Adult patients with Fabry disease: what does the cardiologist need to know?: TableÂ1. Heart, 2015, 101, 916-918.	1.2	5

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55	Ten-year outcome of enzyme replacement therapy with agalsidase beta in patients with Fabry disease. Journal of Medical Genetics, 2015, 52, 353-358.	1.5	266
56	Visual Impairment in Pseudoxanthoma Elasticum: A Survey of 40 Patients. Ophthalmic Genetics, 2015, 36, 327-332.	0.5	16
57	Coronary CT angiography for chest pain in pseudoxanthoma elasticum and cardiac intervention management. Journal of Cardiovascular Computed Tomography, 2015, 9, 238-241.	0.7	5
58	The alpha-galactosidase A p.Arg118Cys variant does not cause a Fabry disease phenotype: Data from individual patients and family studies. Molecular Genetics and Metabolism, 2015, 114, 248-258.	0.5	74
59	Urinary biomarker investigation in children with Fabry disease using tandem mass spectrometry. Clinica Chimica Acta, 2015, 438, 195-204.	0.5	62
60	Familial occurrence and heritable connective tissue disorders in cervical artery dissection. Neurology, 2014, 83, 2023-2031.	1.5	74
61	Congenital muscular dystrophy phenotype with neuromuscular spindles excess in a 5-year-old girl caused by HRAS mutation. Neuromuscular Disorders, 2014, 24, 993-998.	0.3	6
62	Neurologic manifestations of inherited disorders of connective tissue. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2014, 119, 565-576.	1.0	41
63	A Phase 2 study of migalastat hydrochloride in females with Fabry disease: Selection of population, safety and pharmacodynamic effects. Molecular Genetics and Metabolism, 2013, 109, 86-92.	0.5	69
64	Migalastat HCl Reduces Globotriaosylsphingosine (Lyso-Gb3) in Fabry Transgenic Mice and in the Plasma of Fabry Patients. PLoS ONE, 2013, 8, e57631.	1.1	40
65	Analysis of left ventricular mass in untreated men and in men treated with agalsidase-Î²: data from the Fabry Registry. Genetics in Medicine, 2013, 15, 958-965.	1.1	74
66	Long-term changes in arterial structure and function and left ventricular geometry after enzyme replacement therapy in patients affected with Fabry disease. European Journal of Preventive Cardiology, 2012, 19, 43-54.	0.8	13
67	Renal outcomes of agalsidase beta treatment for Fabry disease: role of proteinuria and timing of treatment initiation. Nephrology Dialysis Transplantation, 2012, 27, 1042-1049.	0.4	132
68	Anti-Î±-galactosidase A antibody response to agalsidase beta treatment: Data from the Fabry Registry. Molecular Genetics and Metabolism, 2012, 105, 443-449.	0.5	58
69	Safety and pharmacodynamic effects of a pharmacological chaperone on Î±-galactosidase A activity and globotriaosylceramide clearance in Fabry disease: report from two phase 2 clinical studies. Orphanet Journal of Rare Diseases, 2012, 7, 91.	1.2	95
70	The pulvinar sign in Fabry patients: the first report in female patients. Journal of Neurology, 2012, 259, 1227-1228.	1.8	12
71	Ultrastructural scoring of skin biopsies for diagnosis of vascular Ehlersâ€“Danlos syndrome. Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin, 2012, 460, 637-649.	1.4	8
72	Expert opinion on temporary treatment recommendations for Fabry disease during the shortage of enzyme replacement therapy (ERT). Molecular Genetics and Metabolism, 2011, 102, 99-102.	0.5	43

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73	The use of dried blood spot samples in the diagnosis of lysosomal storage disorders â€" Current status and perspectives. Molecular Genetics and Metabolism, 2011, 104, 144-148.	0.5	69
74	Sudden death associated to vascular Ehlersâ€"Danlos syndrome. A case report. Legal Medicine, 2011, 13, 145-147.	0.6	5
75	Screening patients with hypertrophic cardiomyopathy for Fabry disease using a filter-paper test: the FOCUS study. Heart, 2011, 97, 131-136.	1.2	72
76	End-stage renal disease in patients with Fabry disease: natural history data from the Fabry Registry. Nephrology Dialysis Transplantation, 2010, 25, 769-775.	0.4	97
77	Prognostic Indicators of Renal Disease Progression in Adults with Fabry Disease. Clinical Journal of the American Society of Nephrology: CJASN, 2010, 5, 2220-2228.	2.2	122
78	Fabry disease. Orphanet Journal of Rare Diseases, 2010, 5, 30.	1.2	885
79	Uneventful pregnancy outcome after enzyme replacement therapy with agalsidase beta in a heterozygous female with Fabry disease: A case report. European Journal of Medical Genetics, 2010, 53, 111-112.	0.7	13
80	A validated disease severity scoring system for Fabry disease. Molecular Genetics and Metabolism, 2010, 99, 283-290.	0.5	56
81	X-linked inheritance and its implication in the diagnosis and management of female patients in Fabry disease. Revue De Medecine Interne, 2010, 31, S209-S213.	0.6	17
82	Enzyme replacement therapy for Fabry's disease. Lancet, The, 2010, 375, 1523.	6.3	8
83	Effect of celiprolol on prevention of cardiovascular events in vascular Ehlers-Danlos syndrome: a prospective randomised, open, blinded-endpoints trial. Lancet, The, 2010, 376, 1476-1484.	6.3	330
84	Fabry disease: a review of current management strategies. QJM - Monthly Journal of the Association of Physicians, 2010, 103, 641-659.	0.2	165
85	Bone and Muscle Involvement in Fabry Disease. , 2010, , 293-298.		0
86	Safety and Efficacy of Enzyme Replacement Therapy with Agalsidase Beta: An International, Open-label Study in Pediatric Patients with Fabry Disease. Journal of Pediatrics, 2008, 152, 563-570.e1.	0.9	126
87	Females with Fabry disease frequently have major organ involvement: Lessons from the Fabry Registry. Molecular Genetics and Metabolism, 2008, 93, 112-128.	0.5	442
88	Urinary globotriaosylceramide excretion correlates with the genotype in children and adults with Fabry disease. Molecular Genetics and Metabolism, 2008, 93, 331-340.	0.5	88
89	Enzyme replacement therapy in Fabry disease: Comparison of agalsidase alfa and agalsidase beta. Molecular Genetics and Metabolism, 2008, 95, 114-115.	0.5	20
90	Characterization of Fabry Disease in 352 Pediatric Patients in the Fabry Registry. Pediatric Research, 2008, 64, 550-555.	1.1	235

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91	Successful reinstitution of agalsidase beta therapy in Fabry disease patients with previous IgE-antibody or skin-test reactivity to the recombinant enzyme. Genetics in Medicine, 2008, 10, 353-358.	1.1	43
92	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.	3.0	396
93	Ehlers-Danlos syndrome type IV. Orphanet Journal of Rare Diseases, 2007, 2, 32.	1.2	273
94	Fabry disease: The need to stratify patient populations to better understand the outcome of enzyme replacement therapy. Clinical Therapeutics, 2007, 29, S17-S18.	1.1	16
95	FabrazymeÂ® therapy in pediatric patients with Fabry disease: Improvements in quality-of-life measures. Clinical Therapeutics, 2007, 29, S31-S32.	1.1	0
96	MALDI-TOF and cluster-TOF-SIMS imaging of Fabry disease biomarkers. International Journal of Mass Spectrometry, 2007, 260, 158-165.	0.7	97
97	Cystatin C as a marker of early changes of renal function in Fabry nephropathy. Journal of Nephrology, 2007, 20, 437-43.	0.9	13
98	Phenotype variations in Gaucher disease. Revue De Medecine Interne, 2006, 27, S7-S10.	0.6	3
99	Therapeutic goals in Gaucher disease. Revue De Medecine Interne, 2006, 27, S34-S38.	0.6	0
100	Fabry disease: Guidelines for the evaluation and management of multi-organ system involvement. Genetics in Medicine, 2006, 8, 539-548.	1.1	347
101	Chiari type I malformation inÂfourÂunrelated patients affected with Fabry disease. European Journal of Medical Genetics, 2006, 49, 419-425.	0.7	18
102	Liquid chromatography on porous graphitic carbon with atmospheric pressure photoionization mass spectrometry and tandem mass spectrometry for the analysis of glycosphingolipids. Journal of Chromatography A, 2006, 1117, 154-162.	1.8	25
103	The vascular ehlers-danlos syndrome. Current Treatment Options in Cardiovascular Medicine, 2006, 8, 121-127.	0.4	24
104	Atmospheric pressure photoionization coupled to porous graphitic carbon liquid chromatography for the analysis of globotriaosylceramides. Application to Fabry disease. Journal of Mass Spectrometry, 2006, 41, 50-58.	0.7	30
105	Osteopenia and osteoporosis: previously unrecognized manifestations of Fabry disease. Clinical Genetics, 2005, 68, 93-95.	1.0	51
106	Fast fingerprinting by MALDIâ€“TOF mass spectrometry of urinary sediment glycosphingolipids in Fabry disease. Analytical and Bioanalytical Chemistry, 2005, 382, 1209-1216.	1.9	48
107	AvancÃ©es rÃ©centes dans le dÃ©pistage de la maladie de Fabry pour les populations Ã risque. Medecine/Sciences, 2005, 21, 48-50.	0.0	5
108	ThÃ©rapies enzymatiques substitutives des maladies lysosomales. Medecine/Sciences, 2005, 21, 77-83.	0.0	4

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109	Manifestations neurologiques de la maladie de Fabry. Medecine/Sciences, 2005, 21, 26-29.	0.0	4
110	DÃ©veloppement clinique de lâ€™agalsidase Î² pour le traitement de la maladie de Fabry. Medecine/Sciences, 2005, 21, 57-61.	0.0	2
111	La maladie de Fabry : de la dÃ©couverte des lysosomes Ã lâ€™avÃ¨nement de la thÃ©rapeutique. Medecine/Sciences, 2005, 21, 5-7.	0.0	3
112	IntÃ©rÃªt et limites de la dÃ©termination de lâ€™activitÃ© enzymatique de lâ€™Î±-galactosidase A dans les populations Ã risque pour la maladie de Fabry. Medecine/Sciences, 2005, 21, 53-54.	0.0	4
113	Apport de lâ€™imagerie par spectromÃ©trie de masse pour lâ€™analyse directe du globotriaosylcÃ©ramide et du galabiosylcÃ©ramide tissulaires. Medecine/Sciences, 2005, 21, 55-56.	0.0	1
114	Atteinte osseuse de la maladie de Fabry. Medecine/Sciences, 2005, 21, 43-44.	0.0	1
115	Quantification et spÃ©ciation du globotriaosylcÃ©ramide. Medecine/Sciences, 2005, 21, 51-52.	0.0	1
116	Increased Carotid Wall Stress in Vascular Ehlers-Danlos Syndrome. Circulation, 2004, 109, 1530-1535.	1.6	104
117	Gaucher's disease: a paradigm for interventional genetics. Clinical Genetics, 2004, 65, 77-86.	1.0	58
118	Vascular Ehlersâ€“Danlos syndrome. Annales De GÃ©nÃ©tique, 2004, 47, 1-9.	0.4	135
119	Optimisation of the separation of four major neutral glycosphingolipids: application to a rapid and simple detection of urinary globotriaosylceramide in Fabry disease. Journal of Chromatography B: Analytical Technologies in the Biomedical and Life Sciences, 2004, 805, 331-337.	1.2	20
120	Long-Term Safety and Efficacy of Enzyme Replacement Therapyfor Fabry Disease. American Journal of Human Genetics, 2004, 75, 65-74.	2.6	394
121	Fabry disease: a functional and anatomical study of cardiac manifestations in 20 hemizygous male patients. Clinical Genetics, 2003, 63, 46-52.	1.0	46
122	Arterial Remodeling and Stiffness in Patients With Pseudoxanthoma Elasticum. Arteriosclerosis, Thrombosis, and Vascular Biology, 2003, 23, 836-841.	1.1	44
123	Ocular manifestations in Fabry disease: a survey of 32 hemizygous male patients. Ophthalmic Genetics, 2003, 24, 129-139.	0.5	84
124	Fabry Disease, an Under-Recognized Multisystemic Disorder: Expert Recommendations for Diagnosis, Management, and Enzyme Replacement Therapy. Annals of Internal Medicine, 2003, 138, 338.	2.0	619
125	Fabry disease: recent advances in enzyme replacement therapy. Expert Opinion on Investigational Drugs, 2002, 11, 1467-1476.	1.9	35
126	Lysosomes et Maladies de Surcharge Lysosomale. SociÃ©tÃ© De Biologie Journal, 2002, 196, 127-134.	0.3	5

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127	Maladie de Fabry (dÃ©ficit en Î±-galactosidase A) : innovations thÃ©rapeutiques rÃ©centes. SociÃ©tÃ© De Biologie Journal, 2002, 196, 183-190.	0.3	12
128	Fabry Disease: Twenty Novel Î±-Galactosidase A Mutations and Genotype-Phenotype Correlations in Classical and Variant Phenotypes. Molecular Medicine, 2002, 8, 306-312.	1.9	69
129	Clinical and Genetic Features of Vascular Ehlers-Danlos Syndrome. Annals of Vascular Surgery, 2002, 16, 391-397.	0.4	149
130	Patients affected with Fabry disease have an increased incidence of progressive hearing loss and sudden deafness: an investigation of twenty-two hemizygous male patients. BMC Medical Genetics, 2002, 3, 10.	2.1	108
131	Endothelial markers and homocysteine in patients with classic Fabry disease. Acta Paediatrica, International Journal of Paediatrics, 2002, 91, 57-61.	0.7	37
132	Arterial remodelling in Fabry disease. Acta Paediatrica, International Journal of Paediatrics, 2002, 91, 62-66.	0.7	39
133	Fabry disease: twenty novel alpha-galactosidase A mutations and genotype-phenotype correlations in classical and variant phenotypes. Molecular Medicine, 2002, 8, 306-12.	1.9	22
134	Safety and Efficacy of Recombinant Human Î±-Galactosidase A Replacement Therapy in Fabry's Disease. New England Journal of Medicine, 2001, 345, 9-16.	13.9	1,433
135	A New Phenotype of Fabry Disease with Intermediate Severity between the Classical Form and the Cardiac Variant. , 2001, 136, 234-240.		45
136	Co-occurrence and contribution of Fabry disease and Klippel-TrÃ©naunay-Weber syndrome to a patient with atypical skin lesions. Clinical Genetics, 2001, 60, 63-67.	1.0	7
137	Identification of a novelde novo mutation (G373D) in the ?-galactosidase A gene (GLA) in a patient affected with Fabry disease. Human Mutation, 2001, 17, 353-353.	1.1	12
138	Identification of novel polymorphisms in the pM5 and MRP1 (ABCC1) genes at locus 16p13.1 and exclusion of both genes as responsible for pseudoxanthoma elasticum. Human Mutation, 2001, 17, 74-75.	1.1	40
139	Mutation analysis of the acid Î²-glucosidase gene in a patient with type 3 Gaucher disease and neutralizing antibody to alglucerase. Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis, 2001, 483, 89-94.	0.4	23
140	Compressibility of the Carotid Artery in Patients With Pseudoxanthoma Elasticum. Hypertension, 2001, 38, 1181-1184.	1.3	42
141	Homozygosity for the R1268Q Mutation in MRP6, the Pseudoxanthoma Elasticum Gene, Is Not Disease-Causing. Biochemical and Biophysical Research Communications, 2000, 274, 297-301.	1.0	39
142	Fabry Disease: Identification of Novel Alpha-Galactosidase A Mutations and Molecular Carrier Detection by Use of Fluorescent Chemical Cleavage of Mismatches. Biochemical and Biophysical Research Communications, 1999, 257, 708-713.	1.0	52
143	Exhaustive Screening of the Acid Î²-Glucosidase Gene, by Fluorescence-Assisted Mismatch Analysis Using Universal Primers: Mutation Profile and Genotype/Phenotype Correlations in Gaucher Disease. American Journal of Human Genetics, 1998, 63, 415-427.	2.6	37
144	Pathological Case of the Month. JAMA Pediatrics, 1997, 151, 97.	3.6	1

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145	Fluorescence-assisted mismatch analysis (FAMA) for exhaustive screening of the Î±-galactosidase A gene and detection of carriers in Fabry disease. Human Genetics, 1996, 98, 719-726.	1.8	52
146	Arterial remodelling in Fabry disease. , 0, 91, 62.		4