

# Dominique Germain

## List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/6393535/publications.pdf>

Version: 2024-02-01

146  
papers

12,642  
citations

31902

53  
h-index

24915

109  
g-index

174  
all docs

174  
docs citations

174  
times ranked

6407  
citing authors

#	ARTICLE	IF	CITATIONS
1	Safety and Efficacy of Recombinant Human $\alpha$ -Galactosidase A Replacement Therapy in Fabry's Disease. <i>New England Journal of Medicine</i> , 2001, 345, 9-16.	13.9	1,433
2	Fabry disease. <i>Orphanet Journal of Rare Diseases</i> , 2010, 5, 30.	1.2	885
3	Fabry Disease, an Under-Recognized Multisystemic Disorder: Expert Recommendations for Diagnosis, Management, and Enzyme Replacement Therapy. <i>Annals of Internal Medicine</i> , 2003, 138, 338.	2.0	619
4	Females with Fabry disease frequently have major organ involvement: Lessons from the Fabry Registry. <i>Molecular Genetics and Metabolism</i> , 2008, 93, 112-128.	0.5	442
5	Sustained, Long-Term Renal Stabilization After 54 Months of Agalsidase $\beta$ Therapy in Patients with Fabry Disease. <i>Journal of the American Society of Nephrology: JASN</i> , 2007, 18, 1547-1557.	3.0	396
6	Long-Term Safety and Efficacy of Enzyme Replacement Therapy for Fabry Disease. <i>American Journal of Human Genetics</i> , 2004, 75, 65-74.	2.6	394
7	Fabry disease revisited: Management and treatment recommendations for adult patients. <i>Molecular Genetics and Metabolism</i> , 2018, 123, 416-427.	0.5	391
8	Treatment of Fabry's Disease with the Pharmacologic Chaperone Migalastat. <i>New England Journal of Medicine</i> , 2016, 375, 545-555.	13.9	390
9	Fabry disease: Guidelines for the evaluation and management of multi-organ system involvement. <i>Genetics in Medicine</i> , 2006, 8, 539-548.	1.1	347
10	Effect of celiprolol on prevention of cardiovascular events in vascular Ehlers-Danlos syndrome: a prospective randomised, open, blinded-endpoints trial. <i>Lancet</i> , The, 2010, 376, 1476-1484.	6.3	330
11	X-chromosome inactivation in female patients with Fabry disease. <i>Clinical Genetics</i> , 2016, 89, 44-54.	1.0	290
12	Ehlers-Danlos syndrome type IV. <i>Orphanet Journal of Rare Diseases</i> , 2007, 2, 32.	1.2	273
13	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.	1.5	266
14	Oral pharmacological chaperone migalastat compared with enzyme replacement therapy in Fabry disease: 18-month results from the randomised phase III ATTRACT study. <i>Journal of Medical Genetics</i> , 2017, 54, 288-296.	1.5	262
15	Recommendations for initiation and cessation of enzyme replacement therapy in patients with Fabry disease: the European Fabry Working Group consensus document. <i>Orphanet Journal of Rare Diseases</i> , 2015, 10, 36.	1.2	239
16	Characterization of Fabry Disease in 352 Pediatric Patients in the Fabry Registry. <i>Pediatric Research</i> , 2008, 64, 550-555.	1.1	235
17	Fabry disease: a review of current management strategies. <i>QJM - Monthly Journal of the Association of Physicians</i> , 2010, 103, 641-659.	0.2	165
18	The validation of pharmacogenetics for the identification of Fabry patients to be treated with migalastat. <i>Genetics in Medicine</i> , 2017, 19, 430-438.	1.1	157

#	ARTICLE	IF	CITATIONS
19	Clinical and Genetic Features of Vascular Ehlers-Danlos Syndrome. <i>Annals of Vascular Surgery</i> , 2002, 16, 391-397.	0.4	149
20	Vascular Ehlers-Danlos syndrome. <i>Annales De G�n�tologie</i> , 2004, 47, 1-9.	0.4	135
21	Renal outcomes of agalsidase beta treatment for Fabry disease: role of proteinuria and timing of treatment initiation. <i>Nephrology Dialysis Transplantation</i> , 2012, 27, 1042-1049.	0.4	132
22	Safety and Efficacy of Enzyme Replacement Therapy with Agalsidase Beta: An International, Open-label Study in Pediatric Patients with Fabry Disease. <i>Journal of Pediatrics</i> , 2008, 152, 563-570.e1.	0.9	126
23	Prognostic Indicators of Renal Disease Progression in Adults with Fabry Disease. <i>Clinical Journal of the American Society of Nephrology: CJASN</i> , 2010, 5, 2220-2228.	2.2	122
24	European expert consensus statement on therapeutic goals in Fabry disease. <i>Molecular Genetics and Metabolism</i> , 2018, 124, 189-203.	0.5	122
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. <i>Molecular Genetics and Metabolism Reports</i> , 2019, 19, 100454.	0.4	120
26	Pseudoxanthoma elasticum. <i>Orphanet Journal of Rare Diseases</i> , 2017, 12, 85.	1.2	118
27	Patients affected with Fabry disease have an increased incidence of progressive hearing loss and sudden deafness: an investigation of twenty-two hemizygous male patients. <i>BMC Medical Genetics</i> , 2002, 3, 10.	2.1	108
28	Increased Carotid Wall Stress in Vascular Ehlers-Danlos Syndrome. <i>Circulation</i> , 2004, 109, 1530-1535.	1.6	104
29	Time to treatment benefit for adult patients with Fabry disease receiving agalsidase �: data from the Fabry Registry. <i>Journal of Medical Genetics</i> , 2016, 53, 495-502.	1.5	101
30	MALDI-TOF and cluster-TOF-SIMS imaging of Fabry disease biomarkers. <i>International Journal of Mass Spectrometry</i> , 2007, 260, 158-165.	0.7	97
31	End-stage renal disease in patients with Fabry disease: natural history data from the Fabry Registry. <i>Nephrology Dialysis Transplantation</i> , 2010, 25, 769-775.	0.4	97
32	An expert consensus document on the management of cardiovascular manifestations of Fabry disease. <i>European Journal of Heart Failure</i> , 2020, 22, 1076-1096.	2.9	96
33	Safety and pharmacodynamic effects of a pharmacological chaperone on �-galactosidase A activity and globotriaosylceramide clearance in Fabry disease: report from two phase 2 clinical studies. <i>Orphanet Journal of Rare Diseases</i> , 2012, 7, 91.	1.2	95
34	Urinary globotriaosylceramide excretion correlates with the genotype in children and adults with Fabry disease. <i>Molecular Genetics and Metabolism</i> , 2008, 93, 331-340.	0.5	88
35	Consensus recommendations for diagnosis, management and treatment of Fabry disease in paediatric patients. <i>Clinical Genetics</i> , 2019, 96, 107-117.	1.0	87
36	Ocular manifestations in Fabry disease: a survey of 32 hemizygous male patients. <i>Ophthalmic Genetics</i> , 2003, 24, 129-139.	0.5	84

#	ARTICLE	IF	CITATIONS
37	Pain in Fabry Disease: Practical Recommendations for Diagnosis and Treatment. <i>CNS Neuroscience and Therapeutics</i> , 2016, 22, 568-576.	1.9	75
38	Analysis of left ventricular mass in untreated men and in men treated with agalsidase- $\beta$ : data from the Fabry Registry. <i>Genetics in Medicine</i> , 2013, 15, 958-965.	1.1	74
39	Familial occurrence and heritable connective tissue disorders in cervical artery dissection. <i>Neurology</i> , 2014, 83, 2023-2031.	1.5	74
40	The alpha-galactosidase A p.Arg118Cys variant does not cause a Fabry disease phenotype: Data from individual patients and family studies. <i>Molecular Genetics and Metabolism</i> , 2015, 114, 248-258.	0.5	74
41	Screening patients with hypertrophic cardiomyopathy for Fabry disease using a filter-paper test: the FOCUS study. <i>Heart</i> , 2011, 97, 131-136.	1.2	72
42	Phenotypic characteristics of the p.Asn215Ser (p.N215S) <i>G</i> mutation in male and female patients with Fabry disease: A multicenter Fabry Registry study. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2018, 6, 492-503.	0.6	70
43	Fabry Disease: Twenty Novel $\alpha$ -Galactosidase A Mutations and Genotype-Phenotype Correlations in Classical and Variant Phenotypes. <i>Molecular Medicine</i> , 2002, 8, 306-312.	1.9	69
44	The use of dried blood spot samples in the diagnosis of lysosomal storage disorders – Current status and perspectives. <i>Molecular Genetics and Metabolism</i> , 2011, 104, 144-148.	0.5	69
45	A Phase 2 study of migalastat hydrochloride in females with Fabry disease: Selection of population, safety and pharmacodynamic effects. <i>Molecular Genetics and Metabolism</i> , 2013, 109, 86-92.	0.5	69
46	Fabry disease in cardiology practice: Literature review and expert point of view. <i>Archives of Cardiovascular Diseases</i> , 2019, 112, 278-287.	0.7	69
47	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. <i>Genetics in Medicine</i> , 2019, 21, 1987-1997.	1.1	66
48	The effect of enzyme replacement therapy on clinical outcomes in female patients with Fabry disease – A systematic literature review by a European panel of experts. <i>Molecular Genetics and Metabolism</i> , 2019, 126, 224-235.	0.5	65
49	Urinary biomarker investigation in children with Fabry disease using tandem mass spectrometry. <i>Clinica Chimica Acta</i> , 2015, 438, 195-204.	0.5	62
50	Gaucher's disease: a paradigm for interventional genetics. <i>Clinical Genetics</i> , 2004, 65, 77-86.	1.0	58
51	Anti- $\alpha$ -galactosidase A antibody response to agalsidase beta treatment: Data from the Fabry Registry. <i>Molecular Genetics and Metabolism</i> , 2012, 105, 443-449.	0.5	58
52	A validated disease severity scoring system for Fabry disease. <i>Molecular Genetics and Metabolism</i> , 2010, 99, 283-290.	0.5	56
53	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. <i>PLoS ONE</i> , 2017, 12, e0181700.	1.1	55
54	Mutation spectrum in the ABCC6 gene and genotype-phenotype correlations in a French cohort with pseudoxanthoma elasticum. <i>Genetics in Medicine</i> , 2017, 19, 909-917.	1.1	54

#	ARTICLE	IF	CITATIONS
55	Fluorescence-assisted mismatch analysis (FAMA) for exhaustive screening of the $\beta$ -galactosidase A gene and detection of carriers in Fabry disease. <i>Human Genetics</i> , 1996, 98, 719-726.	1.8	52
56	Fabry Disease: Identification of Novel Alpha-Galactosidase A Mutations and Molecular Carrier Detection by Use of Fluorescent Chemical Cleavage of Mismatches. <i>Biochemical and Biophysical Research Communications</i> , 1999, 257, 708-713.	1.0	52
57	Osteopenia and osteoporosis: previously unrecognized manifestations of Fabry disease. <i>Clinical Genetics</i> , 2005, 68, 93-95.	1.0	51
58	The effect of enzyme replacement therapy on clinical outcomes in paediatric patients with Fabry disease – A systematic literature review by a European panel of experts. <i>Molecular Genetics and Metabolism</i> , 2019, 126, 212-223.	0.5	50
59	Fast fingerprinting by MALDI-TOF mass spectrometry of urinary sediment glycosphingolipids in Fabry disease. <i>Analytical and Bioanalytical Chemistry</i> , 2005, 382, 1209-1216.	1.9	48
60	Whole-body muscle magnetic resonance imaging in <i>SEP1</i> -related myopathy shows a homogeneous and recognizable pattern. <i>Muscle and Nerve</i> , 2015, 52, 728-735.	1.0	47
61	Fabry disease: a functional and anatomical study of cardiac manifestations in 20 hemizygous male patients. <i>Clinical Genetics</i> , 2003, 63, 46-52.	1.0	46
62	A New Phenotype of Fabry Disease with Intermediate Severity between the Classical Form and the Cardiac Variant. , 2001, 136, 234-240.		45
63	Arterial Remodeling and Stiffness in Patients With Pseudoxanthoma Elasticum. <i>Arteriosclerosis, Thrombosis, and Vascular Biology</i> , 2003, 23, 836-841.	1.1	44
64	Successful reinstitution of agalsidase beta therapy in Fabry disease patients with previous IgE-antibody or skin-test reactivity to the recombinant enzyme. <i>Genetics in Medicine</i> , 2008, 10, 353-358.	1.1	43
65	Expert opinion on temporary treatment recommendations for Fabry disease during the shortage of enzyme replacement therapy (ERT). <i>Molecular Genetics and Metabolism</i> , 2011, 102, 99-102.	0.5	43
66	Use of a rare disease registry for establishing phenotypic classification of previously unassigned <i>GLA</i> variants: a consensus classification system by a multispecialty Fabry disease genotype-phenotype workgroup. <i>Journal of Medical Genetics</i> , 2020, 57, 542-551.	1.5	43
67	Compressibility of the Carotid Artery in Patients With Pseudoxanthoma Elasticum. <i>Hypertension</i> , 2001, 38, 1181-1184.	1.3	42
68	Determinants of white matter hyperintensity burden in patients with Fabry disease. <i>Neurology</i> , 2016, 86, 1880-1886.	1.5	42
69	Neurologic manifestations of inherited disorders of connective tissue. <i>Handbook of Clinical Neurology</i> / Edited By P J Vinken and G W Bruyn, 2014, 119, 565-576.	1.0	41
70	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. <i>Human Mutation</i> , 2001, 17, 74-75.	1.1	40
71	Migalastat HCl Reduces Globotriaosylsphingosine (Lyso-Gb3) in Fabry Transgenic Mice and in the Plasma of Fabry Patients. <i>PLoS ONE</i> , 2013, 8, e57631.	1.1	40
72	Homozygosity for the R1268Q Mutation in MRP6, the Pseudoxanthoma Elasticum Gene, Is Not Disease-Causing. <i>Biochemical and Biophysical Research Communications</i> , 2000, 274, 297-301.	1.0	39

#	ARTICLE	IF	CITATIONS
73	Arterial remodelling in Fabry disease. <i>Acta Paediatrica, International Journal of Paediatrics</i> , 2002, 91, 62-66.	0.7	39
74	Basilar Artery Changes in Fabry Disease. <i>American Journal of Neuroradiology</i> , 2017, 38, 531-536.	1.2	39
75	Exhaustive Screening of the Acid $\beta$ -Glucosidase Gene, by Fluorescence-Assisted Mismatch Analysis Using Universal Primers: Mutation Profile and Genotype/Phenotype Correlations in Gaucher Disease. <i>American Journal of Human Genetics</i> , 1998, 63, 415-427.	2.6	37
76	Endothelial markers and homocysteine in patients with classic Fabry disease. <i>Acta Paediatrica, International Journal of Paediatrics</i> , 2002, 91, 57-61.	0.7	37
77	Fabry disease: recent advances in enzyme replacement therapy. <i>Expert Opinion on Investigational Drugs</i> , 2002, 11, 1467-1476.	1.9	35
78	Atmospheric pressure photoionization coupled to porous graphitic carbon liquid chromatography for the analysis of globotriaosylceramides. Application to Fabry disease. <i>Journal of Mass Spectrometry</i> , 2006, 41, 50-58.	0.7	30
79	The role of CNVs in the etiology of rare autosomal recessive disorders: the example of TRAPPC9-associated intellectual disability. <i>European Journal of Human Genetics</i> , 2018, 26, 143-148.	1.4	26
80	The benefits and challenges of family genetic testing in rare genetic diseases—lessons from Fabry disease. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2021, 9, e1666.	0.6	26
81	Challenging the traditional approach for interpreting genetic variants: Lessons from Fabry disease. <i>Clinical Genetics</i> , 2022, 101, 390-402.	1.0	26
82	Liquid chromatography on porous graphitic carbon with atmospheric pressure photoionization mass spectrometry and tandem mass spectrometry for the analysis of glycosphingolipids. <i>Journal of Chromatography A</i> , 2006, 1117, 154-162.	1.8	25
83	The vascular ehlers-danlos syndrome. <i>Current Treatment Options in Cardiovascular Medicine</i> , 2006, 8, 121-127.	0.4	24
84	Mutation analysis of the acid $\beta$ -glucosidase gene in a patient with type 3 Gaucher disease and neutralizing antibody to $\alpha$ -glucosidase. <i>Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis</i> , 2001, 483, 89-94.	0.4	23
85	Fabry disease: twenty novel alpha-galactosidase A mutations and genotype-phenotype correlations in classical and variant phenotypes. <i>Molecular Medicine</i> , 2002, 8, 306-12.	1.9	22
86	Fabry disease caused by the GLA p.Phe113Leu (p.F113L) variant: Natural history in males. <i>European Journal of Medical Genetics</i> , 2020, 63, 103703.	0.7	21
87	Optimisation of the separation of four major neutral glycosphingolipids: application to a rapid and simple detection of urinary globotriaosylceramide in Fabry disease. <i>Journal of Chromatography B: Analytical Technologies in the Biomedical and Life Sciences</i> , 2004, 805, 331-337.	1.2	20
88	Enzyme replacement therapy in Fabry disease: Comparison of agalsidase alfa and agalsidase beta. <i>Molecular Genetics and Metabolism</i> , 2008, 95, 114-115.	0.5	20
89	Newborn Screening for Fabry Disease in Northeastern Italy: Results of Five Years of Experience. <i>Biomolecules</i> , 2021, 11, 951.	1.8	20
90	Chiari type I malformation in four unrelated patients affected with Fabry disease. <i>European Journal of Medical Genetics</i> , 2006, 49, 419-425.	0.7	18

#	ARTICLE	IF	CITATIONS
91	Improvement of Fabry Disease-Related Gastrointestinal Symptoms in a Significant Proportion of Female Patients Treated with Agalsidase Beta: Data from the Fabry Registry. <i>JIMD Reports</i> , 2017, 38, 45-51.	0.7	18
92	Diagnosis and Management of Cardiovascular Involvement in Fabry Disease. <i>Heart Failure Clinics</i> , 2022, 18, 39-49.	1.0	18
93	X-linked inheritance and its implication in the diagnosis and management of female patients in Fabry disease. <i>Revue De Medecine Interne</i> , 2010, 31, S209-S213.	0.6	17
94	Fabry disease: The need to stratify patient populations to better understand the outcome of enzyme replacement therapy. <i>Clinical Therapeutics</i> , 2007, 29, S17-S18.	1.1	16
95	Visual Impairment in Pseudoxanthoma Elasticum: A Survey of 40 Patients. <i>Ophthalmic Genetics</i> , 2015, 36, 327-332.	0.5	16
96	Atypical COL3A1 variants (glutamic acid to lysine) cause vascular Ehlers-Danlos syndrome with a consistent phenotype of tissue fragility and skin hyperextensibility. <i>Genetics in Medicine</i> , 2019, 21, 2081-2091.	1.1	16
97	Uneventful pregnancy outcome after enzyme replacement therapy with agalsidase beta in a heterozygous female with Fabry disease: A case report. <i>European Journal of Medical Genetics</i> , 2010, 53, 111-112.	0.7	13
98	Long-term changes in arterial structure and function and left ventricular geometry after enzyme replacement therapy in patients affected with Fabry disease. <i>European Journal of Preventive Cardiology</i> , 2012, 19, 43-54.	0.8	13
99	Therapeutic goals in Fabry disease: Recommendations of a European expert panel, based on current clinical evidence with enzyme replacement therapy. <i>Molecular Genetics and Metabolism</i> , 2019, 126, 210-211.	0.5	13
100	Cardiomyopathy and kidney function in agalsidase beta-treated female Fabry patients: a pre-treatment vs. post-treatment analysis. <i>ESC Heart Failure</i> , 2020, 7, 825-834.	1.4	13
101	Cystatin C as a marker of early changes of renal function in Fabry nephropathy. <i>Journal of Nephrology</i> , 2007, 20, 437-43.	0.9	13
102	Identification of a novel de novo mutation (G373D) in the $\alpha$ -galactosidase A gene (GLA) in a patient affected with Fabry disease. <i>Human Mutation</i> , 2001, 17, 353-353.	1.1	12
103	Maladie de Fabry (déficit en $\alpha$ -galactosidase A) : innovations thérapeutiques récentes. <i>Société De Biologie Journal</i> , 2002, 196, 183-190.	0.3	12
104	The pulvinar sign in Fabry patients: the first report in female patients. <i>Journal of Neurology</i> , 2012, 259, 1227-1228.	1.8	12
105	Enzymatic diagnosis of Fabry disease using a fluorometric assay on dried blood spots: An alternative methodology. <i>European Journal of Medical Genetics</i> , 2015, 58, 681-684.	0.7	12
106	Why systematic literature reviews in Fabry disease should include all published evidence. <i>European Journal of Medical Genetics</i> , 2019, 62, 103702.	0.7	12
107	Fabry disease and COVID-19: international expert recommendations for management based on real-world experience. <i>CKJ: Clinical Kidney Journal</i> , 2020, 13, 913-925.	1.4	11
108	Clinical utility gene card for: Fabry disease – update 2016. <i>European Journal of Human Genetics</i> , 2017, 25, e1-e1.	1.4	10

#	ARTICLE	IF	CITATIONS
109	Screening for Fabry disease in male patients with end-stage renal disease in western France. <i>Nephrologie Et Therapeutique</i> , 2021, 17, 180-184.	0.2	9
110	Enzyme replacement therapy for Fabry's disease. <i>Lancet, The</i> , 2010, 375, 1523.	6.3	8
111	Ultrastructural scoring of skin biopsies for diagnosis of vascular Ehlers-Danlos syndrome. <i>Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin</i> , 2012, 460, 637-649.	1.4	8
112	Co-occurrence and contribution of Fabry disease and Klippel-Tränaunay-Weber syndrome to a patient with atypical skin lesions. <i>Clinical Genetics</i> , 2001, 60, 63-67.	1.0	7
113	Congenital muscular dystrophy phenotype with neuromuscular spindles excess in a 5-year-old girl caused by HRAS mutation. <i>Neuromuscular Disorders</i> , 2014, 24, 993-998.	0.3	6
114	Treatment needs and expectations for Fabry disease in France: development of a new Patient Needs Questionnaire. <i>Orphanet Journal of Rare Diseases</i> , 2019, 14, 284.	1.2	6
115	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. <i>Molecular Genetics and Metabolism Reports</i> , 2020, 25, 100670.	0.4	6
116	Lysosomes et Maladies de Surcharge Lysosomale. <i>Société De Biologie Journal</i> , 2002, 196, 127-134.	0.3	5
117	Avancées récentes dans le dépistage de la maladie de Fabry pour les populations à risque. <i>Medecine/Sciences</i> , 2005, 21, 48-50.	0.0	5
118	Sudden death associated to vascular Ehlers-Danlos syndrome. A case report. <i>Legal Medicine</i> , 2011, 13, 145-147.	0.6	5
119	Raynaud's phenomenon associated with Fabry disease. <i>Journal of Inherited Metabolic Disease</i> , 2015, 38, 367-368.	1.7	5
120	Adult patients with Fabry disease: what does the cardiologist need to know?: Table 1. <i>Heart</i> , 2015, 101, 916-918.	1.2	5
121	Coronary CT angiography for chest pain in pseudoxanthoma elasticum and cardiac intervention management. <i>Journal of Cardiovascular Computed Tomography</i> , 2015, 9, 238-241.	0.7	5
122	Fabry disease: Four case reports of meningioma and a review of the literature on other malignancies. <i>Molecular Genetics and Metabolism Reports</i> , 2017, 11, 75-80.	0.4	5
123	Principles of Human Genetics and Mendelian Inheritance. , 2018, , 1-28.		5
124	Semen and male genital tract characteristics of patients with Fabry disease: the FERTIFABRY multicentre observational study. <i>Basic and Clinical Andrology</i> , 2019, 29, 7.	0.8	5
125	Thérapies enzymatiques substitutives des maladies lysosomales. <i>Medecine/Sciences</i> , 2005, 21, 77-83.	0.0	4
126	Manifestations neurologiques de la maladie de Fabry. <i>Medecine/Sciences</i> , 2005, 21, 26-29.	0.0	4

#	ARTICLE	IF	CITATIONS
127	IN VIVO OBSERVATION OF RETINAL VASCULAR DEPOSITS USING ADAPTIVE OPTICS IMAGING IN FABRY DISEASE. <i>Retina</i> , 2020, 40, 1623-1629.	1.0	4
128	Case Report: First Two Identified Cases of Fabry Disease in Central Asia. <i>Frontiers in Genetics</i> , 2021, 12, 657824.	1.1	4
129	Intérêt et limites de la détermination de l'activité enzymatique de l'α-galactosidase A dans les populations à risque pour la maladie de Fabry. <i>Medecine/Sciences</i> , 2005, 21, 53-54.	0.0	4
130	Arterial remodelling in Fabry disease. , 0, 91, 62.		4
131	Phenotype variations in Gaucher disease. <i>Revue De Medecine Interne</i> , 2006, 27, S7-S10.	0.6	3
132	The phenotypic characteristics of the p.N215S Fabry disease genotype in male and female patients: a multi-center Fabry Registry study. <i>Molecular Genetics and Metabolism</i> , 2017, 120, S51-S52.	0.5	3
133	La maladie de Fabry : de la découverte des lysosomes à l'avènement de la thérapie. <i>Medecine/Sciences</i> , 2005, 21, 5-7.	0.0	3
134	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. <i>Vaccines</i> , 2021, 9, 1412.	2.1	3
135	Développement clinique de l'agalsidase β pour le traitement de la maladie de Fabry. <i>Medecine/Sciences</i> , 2005, 21, 57-61.	0.0	2
136	MO035HISTORICAL CONTROL ANALYSIS DEMONSTRATES SUPERIOR REDUCTION OF PLASMA GLOBOTRIAOSYL CERAMIDE BY VENGLUSTAT COMPARED WITH PLACEBO OR AGALSIDASE BETA IN CLASSIC FABRY DISEASE PATIENTS. <i>Nephrology Dialysis Transplantation</i> , 2020, 35, .	0.4	2
137	Pathological Case of the Month. <i>JAMA Pediatrics</i> , 1997, 151, 97.	3.6	1
138	Efficacy and safety of migalastat, an oral pharmacologic chaperone for Fabry disease: results from two randomized phase 3 studies, FACETS and ATTRACT. <i>Molecular Genetics and Metabolism</i> , 2017, 120, S45-S46.	0.5	1
139	Apport de l'imagerie par spectrométrie de masse pour l'analyse directe du globotriaosylcéramide et du galabiosylcéramide tissulaires. <i>Medecine/Sciences</i> , 2005, 21, 55-56.	0.0	1
140	Atteinte osseuse de la maladie de Fabry. <i>Medecine/Sciences</i> , 2005, 21, 43-44.	0.0	1
141	Quantification et spéciation du globotriaosylcéramide. <i>Medecine/Sciences</i> , 2005, 21, 51-52.	0.0	1
142	Therapeutic goals in Gaucher disease. <i>Revue De Medecine Interne</i> , 2006, 27, S34-S38.	0.6	0
143	Fabrazyme® therapy in pediatric patients with Fabry disease: Improvements in quality-of-life measures. <i>Clinical Therapeutics</i> , 2007, 29, S31-S32.	1.1	0
144	A survivor analysis for major clinical events in heterozygous female patients with Fabry disease using group consensus phenotype classifications from hemizygous male patients. <i>Molecular Genetics and Metabolism</i> , 2018, 123, S65-S66.	0.5	0

#	ARTICLE	IF	CITATIONS
145	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. <i>Molecular Genetics and Metabolism Reports</i> , 2019, 20, 100493.	0.4	0
146	Bone and Muscle Involvement in Fabry Disease. , 2010, , 293-298.		0