Fabry disease â€˜The New Great Imposterâ€: results of Medicine Departments (FIMeD)

Clinical Genetics

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#	Article	IF	Citations
2	Cerebral hemodynamics and endothelial function in patients with Fabry disease. BMC Neurology, 2013, 13, 170.	1.8	16
3	Difficulties and barriers in diagnosing Fabry disease: what can be learnt from the literature?. Expert Opinion on Medical Diagnostics, 2013, 7, 589-599.	1.6	26
4	Misdiagnosis of familial Mediterranean fever in patients with Andersonâ€"Fabry disease. Clinical Genetics, 2013, 83, 576-581.	2.0	18
5	Analysis of the landscape of biologically-derived pharmaceuticals in Europe: Dominant production systems, molecule types on the rise and approval trends. European Journal of Pharmaceutical Sciences, 2013, 48, 428-441.	4.0	31
6	Mutation identification of Fabry disease in families with other lysosomal storage disorders. Clinical Genetics, 2013, 84, 281-285.	2.0	1
7	Angiokeratomas â€" When is a few too many?. International Journal of STD and AIDS, 2014, 25, 378-379.	1.1	3
8	Prevalence of Raynaud Phenomenon and Nailfold Capillaroscopic Abnormalities in Fabry Disease. Medicine (United States), 2015, 94, e780.	1.0	5
9	High Variability of Fabry Disease Manifestations in an Extended Italian Family. BioMed Research International, 2015, 2015, 1-5.	1.9	23
10	Diagnosing Fabry disease--delays and difficulties within discordant siblings. QJM - Monthly Journal of the Association of Physicians, 2015, 108, 585-590.	0.5	4
11	Diagnostic dilemma and delay in <scp>F</scp>abry disease: Insights from a case series of young female patients. Journal of Paediatrics and Child Health, 2015, 51, 369-372.	0.8	3
12	Fabry disease in infancy and early childhood: a systematic literature review. Genetics in Medicine, 2015, 17, 323-330.	2.4	82
13	Sudoscan as a noninvasive tool to assess sudomotor dysfunction in patients with Fabry disease: results from a case–control study. Therapeutics and Clinical Risk Management, 2016, 12, 135.	2.0	17
14	The impact of fever/hyperthermia in the diagnosis of Fabry: A retrospective analysis. European Journal of Internal Medicine, 2016, 32, 26-30.	2.2	9
15	Sudden death following AV node ablation in a man with Fabry disease mimicking hypertrophic cardiomyopathy. Journal of Clinical Forensic and Legal Medicine, 2016, 42, 8-10.	1.0	0
17	Molecular and clinical studies in five index cases with novel mutations in the GLA gene. Gene, 2016, 578, 100-104.	2.2	20
18	Musculoskeletal manifestations of Fabry disease: A retrospective study. Joint Bone Spine, 2016, 83, 421-426.	1.6	21
19	Manifestations rhumatologiques de la maladie de FabryÂ: Ã©tude rÃ©trospective. Revue Du Rhumatisme (Edition Francaise), 2016, 83, 56-61.	0.0	1
20	A painful diagnosis. Internal and Emergency Medicine, 2017, 12, 341-347.	2.0	0

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21	Disease Progression Modeling to Evaluate the Effects of Enzyme Replacement Therapy on Kidney Function in Adult Patients with the Classic Phenotype of Fabry Disease. Kidney and Blood Pressure Research, 2017, 42, 1-15.	2.0	12
22	Recommendations for the inclusion of Fabry disease as a rare febrile condition in existing algorithms for fever of unknown origin. Internal and Emergency Medicine, 2017, 12, 1059-1067.	2.0	7
24	Fabry Disease in Internal Medicine: The Role of Fever and Hyperthermia in Diagnosis. Giornale De Techniche Nefrologiche & Dialitiche, 2017, 29, S12-S15.	0.1	0
25	Misdiagnosis. Giornale De Techniche Nefrologiche & Dialitiche, 2017, 29, S3-S4.	0.1	0
26	Identification of a novel loss-of-function mutation of the GLA gene in a Chinese Han family with Fabry disease. BMC Medical Genetics, 2018, 19, 219.	2.1	1
27	Mutations in the GLA Gene and LysoGb3: Is It Really Anderson-Fabry Disease?. International Journal of Molecular Sciences, 2018, 19, 3726.	4.1	63
28	Neuroimaging in Fabry disease: current knowledge and future directions. Insights Into Imaging, 2018, 9, 1077-1088.	3.4	37
29	Systematic DNA Study for Fabry Disease in the End Stage Renal Disease Patients from a Southern Italy Area. Kidney and Blood Pressure Research, 2018, 43, 1344-1351.	2.0	7
30	Multiple sclerosis and Fabry disease - diagnostic â€œmixupâ€• Multiple Sclerosis and Related Disorders, 2019, 34, 112-115.	2.0	5
31	Higher rate of rheumatic manifestations and delay in diagnosis in Brazilian Fabry disease patients. Advances in Rheumatology, 2020, 60, 7.	1.7	11
32	Screening for Fabry Disease in Kidney Transplant Recipients: Experience of a Multidisciplinary Team. Biomedicines, 2020, 8, 396.	3.2	15
33	Manifestaciones osteoarticulares de las esfingolipidosis en adultos. EMC - Aparato Locomotor, 2020, 53, 1-29.	0.1	0
34	When and How to Diagnose Fabry Disease in Clinical Pratice. American Journal of the Medical Sciences, 2020, 360, 641-649.	1.1	39
35	Recurrent fever of unknown origin: An overlooked symptom of Fabry disease. Molecular Genetics & Genomic Medicine, 2020, 8, e1454.	1.2	1
36	AA amyloidosis associated with Fabry disease. International Journal of Clinical Practice, 2020, 74, e13577.	1.7	1
37	Prevalence of GLA gene mutations and polymorphisms in patients with multiple sclerosis: A cross-sectional study. Journal of the Neurological Sciences, 2020, 412, 116782.	0.6	2
38	Global warming, heat-related illnesses, and the dermatologist. International Journal of Women's Dermatology, 2021, 7, 70-84.	2.0	21
39	Corneal densitometry: a potential indicator for early diagnosis of Fabry disease. Graefe's Archive for Clinical and Experimental Ophthalmology, 2021, 259, 941-948.	1.9	4

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40	Genetic diseases mimicking multiple sclerosis. Postgraduate Medicine, 2021, 133, 728-749.	2.0	2
41	Fabry disease and multiple sclerosis misdiagnosis: the role of family history and neurological signs. Oncotarget, 2018, 9, 7758-7762.	1.8	11
42	Fabry HastalÄ±ÄŸÄ±: Yeni Bir Mutasyon ve Cilt Bulgulariyla Seyreden Bir TÃ¼rk O. Ã‡ukurova Ãœniversitesi TÄ±p FakÃ¼ltesi Dergisi, 2015, 40, 156.	0.0	0
43	p.R301X Mutation and Variable Phenotypic Appearance of Fabry Disease. American Journal of Case Reports, 2016, 17, 315-319.	0.8	1
44	Ocular Manifestations of Inborn Errors of Metabolism. , 2017, , 359-460.		0
46	DoenÃ§a de Fabry: DiagnÃ³stico Inaugural de uma FamÃlia. Revista De MedicinÄƒ InternÄƒ, Neurologe, Psihiatrie, Neurochirurgie, Dermato-venerologie MedicinÄƒ InternÄƒ, 2021, 28, 35-38.	0.0	0
47	Understanding the ecosystem of patients with lysosomal storage diseases in Spain: a qualitative research with patients and health care professionals. Orphanet Journal of Rare Diseases, 2022, 17, 17.	2.7	1
48	A 39Âyear-old man with acroparesthesia and uncommon renal arterial lesions. What is theÂdiagnosis?. Journal of Nephrology, 2022, , 1.	2.0	0
49	Diagnosis of Fabry Disease in a Patient with a Surgically Repaired Congenital Heart Defect: When Clinical History and Genetics Make the Difference. Neurology International, 2022, 12, 102-108.	0.5	1
50	Expert opinion on the recognition, diagnosis and management of children and adults with Fabry disease: a multidisciplinary Turkey perspective. Orphanet Journal of Rare Diseases, 2022, 17, 90.	2.7	8
51	Left Ventricular Hypertrophy: Etiology-Based Therapeutic Options. Cardiology and Therapy, 2022, 11, 203-230.	2.6	4
52	Fabry disease prevalence in patients with familial Mediterranean fever: A cohort study. Journal of Surgery and Medicine, 2022, 6, 601-604.	0.1	0
53	Balance control impairments in Fabry disease. Frontiers in Neurology, 0, 13, .	2.4	3
54	Association of Fabry Disease with Hearing Loss, Tinnitus, and Sudden Hearing Loss: A Nationwide Population-Based Study. Journal of Clinical Medicine, 2022, 11, 7396.	2.4	2
56	Biochemical Mechanisms beyond Glycosphingolipid Accumulation in Fabry Disease: Might They Provide Additional Therapeutic Treatments?. Journal of Clinical Medicine, 2023, 12, 2063.	2.4	3
57	Evaluation of Hearing and Auditory Pathways in Fabry Disease Patients. Turkish Journal of Internal Medicine, 2023, 5, 84-91.	0.6	0
58	Missed diagnosis of Fabry disease: should we screen patients with multiple sclerosis?. Neurological Sciences, 0, , .	1.9	0
59	Outcomes and management of kidney transplant recipients with Fabry disease: a review. Journal of Nephrology, 0, , .	2.0	0