

Nancy J Mendelsohn

List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/10725486/publications.pdf>

Version: 2024-02-01

25
papers

1,862
citations

361413

20
h-index

610901

24
g-index

25
all docs

25
docs citations

25
times ranked

3102
citing authors

#	ARTICLE	IF	CITATIONS
1	Intrafamilial variability in the clinical manifestations of mucopolysaccharidosis type II: Data from the Hunter Outcome Survey (HOS). <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 301-310.	1.2	15
2	A patient with germ-line gain-of-function PDGFRB p.N666H mutation and marked clinical response to imatinib. <i>Genetics in Medicine</i> , 2018, 20, 142-150.	2.4	48
3	Refining the phenotype associated with <i>GNB1</i> mutations: Clinical data on 18 newly identified patients and review of the literature. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2259-2275.	1.2	47
4	Retinal dystrophy in two boys with Costello syndrome due to the HRAS p.Gly13Cys mutation. , 2017, 173, 1342-1347.		7
5	Ten years of the Hunter Outcome Survey (HOS): insights, achievements, and lessons learned from a global patient registry. <i>Orphanet Journal of Rare Diseases</i> , 2017, 12, 82.	2.7	48
6	Sustained immune tolerance induction in enzyme replacement therapy-treated CRIM-negative patients with infantile Pompe disease. <i>JCI Insight</i> , 2017, 2, .	5.0	47
7	Variable brain phenotype primarily affects the brainstem and cerebellum in patients with osteogenesis imperfecta caused by recessive <i>WNT1</i> mutations. <i>Journal of Medical Genetics</i> , 2016, 53, 427-430.	3.2	41
8	The natural history of growth in patients with Hunter syndrome: Data from the Hunter Outcome Survey (HOS). <i>Molecular Genetics and Metabolism</i> , 2016, 117, 438-446.	1.1	33
9	Rapid molecular diagnostics of severe primary immunodeficiency determined by using targeted next-generation sequencing. <i>Journal of Allergy and Clinical Immunology</i> , 2016, 138, 1142-1151.e2.	2.9	85
10	Whole Exome Sequencing, Familial Genomic Triangulation, and Systems Biology Converge to Identify a Novel Nonsense Mutation in <i>TAB2</i> -encoded TGF-beta Activated Kinase 1 in a Child with Polyvalvular Syndrome. <i>Congenital Heart Disease</i> , 2016, 11, 452-461.	0.2	15
11	Levels of glycosaminoglycans in the cerebrospinal fluid of healthy young adults, surrogate-normal children, and Hunter syndrome patients with and without cognitive impairment. <i>Molecular Genetics and Metabolism Reports</i> , 2015, 5, 103-106.	1.1	24
12	Identifying the need for a multidisciplinary approach for early recognition of mucopolysaccharidosis VI (MPS VI). <i>Molecular Genetics and Metabolism</i> , 2015, 115, 41-47.	1.1	9
13	Overcoming the barriers to diagnosis of Morquio A syndrome. <i>Orphanet Journal of Rare Diseases</i> , 2014, 9, 192.	2.7	21
14	Mucopolysaccharidosis IVA (Morquio A syndrome) and VI (Maroteaux-Lamy syndrome): under-recognized and challenging to diagnose. <i>Skeletal Radiology</i> , 2014, 43, 359-369.	2.0	39
15	<i>WNT1</i> Mutations in Families Affected by Moderately Severe and Progressive Recessive Osteogenesis Imperfecta. <i>American Journal of Human Genetics</i> , 2013, 92, 590-597.	6.2	179
16	Clinical genetics evaluation in identifying the etiology of autism spectrum disorders: 2013 guideline revisions. <i>Genetics in Medicine</i> , 2013, 15, 399-407.	2.4	402
17	The effect of idursulfase on growth in patients with Hunter syndrome: Data from the Hunter Outcome Survey (HOS). <i>Molecular Genetics and Metabolism</i> , 2013, 109, 41-48.	1.1	53
18	Spondyloepiphyseal Dysplasias and Bilateral Legg-Calv�-Perthes Disease: Diagnostic Considerations for Mucopolysaccharidoses. <i>JIMD Reports</i> , 2013, 11, 125-132.	1.5	24

#	ARTICLE	IF	CITATIONS
19	Successful immune tolerance induction to enzyme replacement therapy in CRIM-negative infantile Pompe disease. <i>Genetics in Medicine</i> , 2012, 14, 135-142.	2.4	183
20	The new era of Pompe disease: Advances in the detection, understanding of the phenotypic spectrum, pathophysiology, and management. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2012, 160C, 1-7.	1.6	68
21	Importance of surgical history in diagnosing mucopolysaccharidosis type II (Hunter syndrome): Data from the Hunter Outcome Survey. <i>Genetics in Medicine</i> , 2010, 12, 816-822.	2.4	63
22	Elimination of Antibodies to Recombinant Enzyme in Pompe's Disease. <i>New England Journal of Medicine</i> , 2009, 360, 194-195.	27.0	136
23	Genetic Evaluation of Autism. <i>Seminars in Pediatric Neurology</i> , 2008, 15, 27-31.	2.0	27
24	Clinical genetics evaluation in identifying the etiology of autism spectrum disorders. <i>Genetics in Medicine</i> , 2008, 10, 301-305.	2.4	112
25	Genetics evaluation for the etiologic diagnosis of autism spectrum disorders. <i>Genetics in Medicine</i> , 2008, 10, 4-12.	2.4	136