Nancy J Mendelsohn

List of Publications by Year
in descending order

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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	Clinical genetics evaluation in identifying the etiology of autism spectrum disorders: 2013 guideline revisions. Genetics in Medicine, 2013, 15, 399-407.	2.4	402
2	Successful immune tolerance induction to enzyme replacement therapy in CRIM-negative infantile Pompe disease. Genetics in Medicine, 2012, 14, 135-142.	2.4	183
3	WNT1 Mutations in Families Affected by Moderately Severe and Progressive Recessive Osteogenesis Imperfecta. American Journal of Human Genetics, 2013, 92, 590-597.	6.2	179
4	Genetics evaluation for the etiologic diagnosis of autism spectrum disorders. Genetics in Medicine, 2008, 10, 4-12.	2.4	136
5	Elimination of Antibodies to Recombinant Enzyme in Pompe's Disease. New England Journal of Medicine, 2009, 360, 194-195.	27.0	136
6	Clinical genetics evaluation in identifying the etiology of autism spectrum disorders. Genetics in Medicine, 2008, 10, 301-305.	2.4	112
7	Rapid molecular diagnostics of severe primary immunodeficiency determined by using targeted next-generation sequencing. Journal of Allergy and Clinical Immunology, 2016, 138, 1142-1151.e2.	2.9	85
8	The new era of Pompe disease: Advances in the detection, understanding of the phenotypic spectrum, pathophysiology, and management. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2012, 160C, 1-7.	1.6	68
9	Importance of surgical history in diagnosing mucopolysaccharidosis type II (Hunter syndrome): Data from the Hunter Outcome Survey. Genetics in Medicine, 2010, 12, 816-822.	2.4	63
10	The effect of idursulfase on growth in patients with Hunter syndrome: Data from the Hunter Outcome Survey (HOS). Molecular Genetics and Metabolism, 2013, 109, 41-48.	1.1	53
11	Ten years of the Hunter Outcome Survey (HOS): insights, achievements, and lessons learned from a global patient registry. Orphanet Journal of Rare Diseases, 2017, 12, 82.	2.7	48
12	A patient with germ-line gain-of-function PDGFRB p.N666H mutation and marked clinical response to imatinib. Genetics in Medicine, 2018, 20, 142-150.	2.4	48
13	Refining the phenotype associated with GNB1 mutations: Clinical data on 18 newly identified patients and review of the literature. American Journal of Medical Genetics, Part A, 2018, 176, 2259-2275.	1.2	47
14	Sustained immune tolerance induction in enzyme replacement therapyâ€“treated CRIM-negative patients with infantile Pompe disease. JCI Insight, 2017, 2, .	5.0	47
15	Variable brain phenotype primarily affects the brainstem and cerebellum in patients with osteogenesis imperfecta caused by recessiveWNT1mutations. Journal of Medical Genetics, 2016, 53, 427-430.	3.2	41
16	Mucopolysaccharidosis IVA (Morquio A syndrome) and VI (Maroteauxâ€“Lamy syndrome): under-recognized and challenging to diagnose. Skeletal Radiology, 2014, 43, 359-369.	2.0	39
17	The natural history of growth in patients with Hunter syndrome: Data from the Hunter Outcome Survey (HOS). Molecular Genetics and Metabolism, 2016, 117, 438-446.	1.1	33
18	Genetic Evaluation of Autism. Seminars in Pediatric Neurology, 2008, 15, 27-31.	2.0	27

#	Article	IF	Citations
19	Spondyloepiphyseal Dysplasias and Bilateral Legg-CalvÃ©-Perthes Disease: Diagnostic Considerations for Mucopolysaccharidoses. JIMD Reports, 2013, 11, 125-132.	1.5	24
20	Levels of glycosaminoglycans in the cerebrospinal fluid of healthy young adults, surrogate-normal children, and Hunter syndrome patients with and without cognitive impairment. Molecular Genetics and Metabolism Reports, 2015, 5, 103-106.	1.1	24
21	Overcoming the barriers to diagnosis of Morquio A syndrome. Orphanet Journal of Rare Diseases, 2014, 9, 192.	2.7	21
22	Whole Exome Sequencing, Familial Genomic Triangulation, and Systems Biology Converge to Identify a Novel Nonsense Mutation inTAB2-encoded TGF-beta Activated Kinase 1 in a Child with Polyvalvular Syndrome. Congenital Heart Disease, 2016, 11, 452-461.	0.2	15
23	Intrafamilial variability in the clinical manifestations of mucopolysaccharidosis type II: Data from the Hunter Outcome Survey (HOS). American Journal of Medical Genetics, Part A, 2018, 176, 301-310.	1.2	15
24	Identifying the need for a multidisciplinary approach for early recognition of mucopolysaccharidosis VI (MPS VI). Molecular Genetics and Metabolism, 2015, 115, 41-47.	1.1	9
25	Retinal dystrophy in two boys with Costello syndrome due to the HRAS p.Gly13Cys mutation. , 2017, 173, 1342-1347.		7