Ellen Sidransky

List of Publications by Year in Descending Order

180 papers	10,151 citations	54 h-index	98 g-index
189 ext. papers	11,892 ext. citations	5.8 avg, IF	6.27 L-index

#	Paper	IF	Citations
180	No Evidence That Glucosylsphingosine Is a Biomarker for Parkinson's Disease: Statistical Differences Do Not Necessarily Indicate Biological Significance.. *Movement Disorders*, **2022**,	7	2
179	Lysosomal dysfunction in neurodegeneration: emerging concepts and methods.. *Trends in Neurosciences*, **2022**,	13.3	2
178	Lyso-IP: Uncovering Pathogenic Mechanisms of Lysosomal Dysfunction. *Biomolecules*, **2022**, 12, 616	5.9	0
177	Neuropathological Features of Gaucher Disease and Gaucher Disease with Parkinsonism. *International Journal of Molecular Sciences*, **2022**, 23, 5842	6.3	0
176	Investigation of a dysmorphic facial phenotype in patients with Gaucher disease types 2 and 3. *Molecular Genetics and Metabolism*, **2021**, 134, 274-280	3.7	1
175	Current and emerging pharmacotherapy for Gaucher disease in pediatric populations. *Expert Opinion on Pharmacotherapy*, **2021**, 22, 1489-1503	4	4
174	The Role of Exosomes in Lysosomal Storage Disorders. *Biomolecules*, **2021**, 11,	5.9	5
173	Gaucher disease in the COVID-19 pandemic environment: The good, the bad and the unknown. *Molecular Genetics and Metabolism*, **2021**, 132, 213-214	3.7	3
172	Next-Generation Sequencing Analysis of : The Challenge of Detecting Complex Recombinant Alleles. *Frontiers in Genetics*, **2021**, 12, 684067	4.5	2
171	Gaucher Disease and Heart Failure of Unknown Origin. *American Journal of Medicine*, **2021**, 134, 745-748	2.4	
170	Genetics provides new individualized therapeutic targets for Parkinson's disease. *Neural Regeneration Research*, **2021**, 16, 994-995	4.5	1
169	Diagnosing neuronopathic Gaucher disease: New considerations and challenges in assigning Gaucher phenotypes. *Molecular Genetics and Metabolism*, **2021**, 132, 49-58	3.7	6
168	Genome sequencing analysis identifies new loci associated with Lewy body dementia and provides insights into its genetic architecture. *Nature Genetics*, **2021**, 53, 294-303	36.3	31
167	Progress in generating iPSC-derived dopaminergic neurons as accurate models of neurodegenerative disease **2021**, 181-203		
166	Gaucher disease and SARS-CoV-2 infection: Emerging management challenges. *Molecular Genetics and Metabolism*, **2020**, 130, 164-169	3.7	14
165	Small Molecule Chaperones for the Treatment of Gaucher Disease and -Associated Parkinson Disease. *Frontiers in Cell and Developmental Biology*, **2020**, 8, 271	5.7	14
164	Longitudinal Positron Emission Tomography of Dopamine Synthesis in Subjects with GBA1 Mutations. *Annals of Neurology*, **2020**, 87, 652-657	9.4	3

163	Glucocerebrosidase as a therapeutic target for Parkinson's disease. *Expert Opinion on Therapeutic Targets*, **2020**, 24, 287-294	6.4	12
162	Substrate reduction therapy for GBA1-associated Parkinsonism: Are we betting on the wrong mouse?. *Movement Disorders*, **2020**, 35, 228-230	7	6
161	A characterization of Gaucher iPS-derived astrocytes: Potential implications for Parkinson's disease. *Neurobiology of Disease*, **2020**, 134, 104647	7.5	32
160	Clinical evaluation of sibling pairs with gaucher disease discordant for parkinsonism. *Movement Disorders*, **2020**, 35, 359-365	7	6
159	White vitreous opacities in five patients with Gaucher disease type 3. *American Journal of Medical Genetics, Part A*, **2020**, 182, 808-812	2.5	1
158	Pro-cathepsin D, Prosaposin, and Progranulin: Lysosomal Networks in Parkinsonism. *Trends in Molecular Medicine*, **2020**, 26, 913-923	11.5	6
157	The natural history of type 2 Gaucher disease in the 21st century: A retrospective study. *Neurology*, **2020**, 95, e2119-e2130	6.5	8
156	Parkinsonism in Patients with Neuronopathic (Type 3) Gaucher Disease: A Case Series. *Movement Disorders Clinical Practice*, **2020**, 7, 834-837	2.2	3
155	EEG abnormalities in patients with chronic neuronopathic Gaucher disease: A retrospective review. *Molecular Genetics and Metabolism*, **2020**, 131, 358-363	3.7	4
154	The definition of neuronopathic Gaucher disease. *Journal of Inherited Metabolic Disease*, **2020**, 43, 1056-1059	5.1	28
153	Glucocerebrosidase and its relevance to Parkinson disease. *Molecular Neurodegeneration*, **2019**, 14, 36	19	96
152	Five-parameter evaluation of dysphagia: A novel prognostic scale for assessing neurological decline in Gaucher disease type 2. *Molecular Genetics and Metabolism*, **2019**, 127, 191-199	3.7	5
151	C-terminal β-synuclein truncations are linked to cysteine cathepsin activity in Parkinson's disease. *Journal of Biological Chemistry*, **2019**, 294, 9973-9984	5.4	23
150	First Clinicogenetic Description of Parkinson's Disease Related to Mutation S107L. *Movement Disorders Clinical Practice*, **2019**, 6, 254-258	2.2	3
149	Mutations, modifiers and epigenetics in Gaucher disease: Blurred boundaries between simple and complex disorders. *Molecular Genetics and Metabolism*, **2019**, 128, 10-13	3.7	3
148	Can GBA1-Associated Parkinson Disease Be Modeled in the Mouse?. *Trends in Neurosciences*, **2019**, 42, 631-643	13.3	6
147	Ophthalmological findings in Gaucher disease. *Molecular Genetics and Metabolism*, **2019**, 127, 23-27	3.7	6
146	Variation in cognitive function over time in Gaucher disease type 3. *Neurology*, **2019**, 93, e2272-e2283	6.5	10

145	GBA1-associated parkinsonism: new insights and therapeutic opportunities. *Current Opinion in Neurology*, **2019**, 32, 589-596	7.1	31
144	Validation of anti-glucocerebrosidase antibodies for western blot analysis on protein lysates of murine and human cells. *Biochemical Journal*, **2019**, 476, 261-274	3.8	4
143	Recent advances in the diagnosis and management of Gaucher disease. *Expert Review of Endocrinology and Metabolism*, **2018**, 13, 107-118	4.1	41
142	ACE phenotyping in Gaucher disease. *Molecular Genetics and Metabolism*, **2018**, 123, 501-510	3.7	16
141	Exploring genetic modifiers of Gaucher disease: The next horizon. *Human Mutation*, **2018**, 39, 1739-1751	4.7	28
140	Glucocerebrosidase mutations and parkinsonism: how much does the mutation matter?. *Journal of Xiangya Medicine*, **2018**, 3, 1-1	0.1	3
139	Alleles with more than one mutation can complicate genotype/phenotype studies in Mendelian disorders: Lessons from Gaucher disease. *Molecular Genetics and Metabolism*, **2018**, 125, 1-3	3.7	8
138	The Complicated Relationship between Gaucher Disease and Parkinsonism: Insights from a Rare Disease. *Neuron*, **2017**, 93, 737-746	13.9	97
137	Induced pluripotent stem cell models of lysosomal storage disorders. *DMM Disease Models and Mechanisms*, **2017**, 10, 691-704	4.1	20
136	Efferocytosis is impaired in Gaucher macrophages. *Haematologica*, **2017**, 102, 656-665	6.6	8
135	The role of epigenetics in lysosomal storage disorders: Uncharted territory. *Molecular Genetics and Metabolism*, **2017**, 122, 10-18	3.7	26
134	Glucocerebrosidase haploinsufficiency in A53T □synuclein mice impacts disease onset and course. *Molecular Genetics and Metabolism*, **2017**, 122, 198-208	3.7	18
133	Type 2 Gaucher disease in an infant despite a normal maternal glucocerebrosidase gene. *American Journal of Medical Genetics, Part A*, **2017**, 173, 3211-3215	2.5	4
132	Gaucher disease: Progress and ongoing challenges. *Molecular Genetics and Metabolism*, **2017**, 120, 8-21	3.7	72
131	Excessive burden of lysosomal storage disorder gene variants in Parkinson's disease. *Brain*, **2017**, 140, 3191-3203	11.2	209
130	The Spectrum of Neurological Manifestations Associated with Gaucher Disease. *Diseases (Basel, Switzerland)*, **2017**, 5,	4.4	41
129	Ocular Implications of Gaucher Disease. *Essentials in Ophthalmology*, **2017**, 413-423	0.2	1
128	Activation of □Glucocerebrosidase Reduces Pathological □Synuclein and Restores Lysosomal Function in Parkinson's Patient Midbrain Neurons. *Journal of Neuroscience*, **2016**, 36, 7693-706	6.6	158

127	A New Glucocerebrosidase Chaperone Reduces -Synuclein and Glycolipid Levels in iPSC-Derived Dopaminergic Neurons from Patients with Gaucher Disease and Parkinsonism. *Journal of Neuroscience*, **2016**, 36, 7441-52	6.6	150
126	Varied autopsy findings in five treated patients with Gaucher disease and parkinsonism include the absence of Gaucher cells. *Molecular Genetics and Metabolism*, **2016**, 118, 55-9	3.7	4
125	The Deleterious Duo of Neurodegeneration: Lysosomes and Mitochondria **2016**, 279-300		0
124	A peptide-linked recombinant glucocerebrosidase for targeted neuronal delivery: Design, production, and assessment. *Journal of Biotechnology*, **2016**, 221, 1-12	3.7	19
123	Once again, rare diseases provide a spotlight. *Molecular Genetics and Metabolism*, **2016**, 118, 1-2	3.7	
122	Chaperoning glucocerebrosidase: a therapeutic strategy for both Gaucher disease and Parkinsonism. *Neural Regeneration Research*, **2016**, 11, 1760-1761	4.5	7
121	A new glucocerebrosidase-deficient neuronal cell model provides a tool to probe pathophysiology and therapeutics for Gaucher disease. *DMM Disease Models and Mechanisms*, **2016**, 9, 769-78	4.1	14
120	Clinical course and prognosis in patients with Gaucher disease and parkinsonism. *Neurology: Genetics*, **2016**, 2, e57	3.8	23
119	Progress and potential of non-inhibitory small molecule chaperones for the treatment of Gaucher disease and its implications for Parkinson disease. *Expert Review of Proteomics*, **2016**, 13, 471-9	4.2	42
118	GBA p.T369M substitution in Parkinson disease: Polymorphism or association? A meta-analysis. *Neurology: Genetics*, **2016**, 2, e104	3.8	51
117	Lysosomal storage and impaired autophagy lead to inflammasome activation in Gaucher macrophages. *Aging Cell*, **2016**, 15, 77-88	9.9	87
116	Gaucher Disease-Induced Pluripotent Stem Cells Display Decreased Erythroid Potential and Aberrant Myelopoiesis. *Stem Cells Translational Medicine*, **2015**, 4, 878-86	6.9	21
115	Deficient vesicular storage: A common theme in catecholaminergic neurodegeneration. *Parkinsonism and Related Disorders*, **2015**, 21, 1013-22	3.6	24
114	Understanding and managing parkinsonism in patients with glucocerebrosidase mutations. *Expert Opinion on Orphan Drugs*, **2015**, 3, 549-562	1.1	1
113	The clinical management of Type 2 Gaucher disease. *Molecular Genetics and Metabolism*, **2015**, 114, 110-122	3.7	71
112	Bilateral Femoral Osteolytic Lesions in a Patient with Type 3 Gaucher Disease. *Molecular Genetics and Metabolism Reports*, **2015**, 5, 107-109	1.8	3
111	Dissociation of glucocerebrosidase dimer in solution by its co-factor, saposin C. *Biochemical and Biophysical Research Communications*, **2015**, 457, 561-6	3.4	14
110	Structural features of membrane-bound glucocerebrosidase and -synuclein probed by neutron reflectometry and fluorescence spectroscopy. *Journal of Biological Chemistry*, **2015**, 290, 744-54	5.4	37

109	New macrophage models of Gaucher disease offer new tools for drug development. *Macrophage*, **2015**, 2, e712		1
108	Applications of iPSC-derived models of Gaucher disease. *Annals of Translational Medicine*, **2015**, 3, 295	3.2	2
107	Complexity of Genotype-Phenotype Correlations in Mendelian Disorders: Lessons from Gaucher Disease. *Advances in Predictive, Preventive and Personalised Medicine*, **2015**, 69-90	0.4	2
106	Glucocerebrosidase is shaking up the synucleinopathies. *Brain*, **2014**, 137, 1304-22	11.2	113
105	Reduced glucocerebrosidase is associated with increased -synuclein in sporadic Parkinson's disease. *Brain*, **2014**, 137, 834-48	11.2	311
104	Studies of glucocerebrosidase provide new therapeutic targets for parkinsonism. *Future Neurology*, **2014**, 9, 407-409	1.5	
103	Lysosomal integral membrane protein-2: a new player in lysosome-related pathology. *Molecular Genetics and Metabolism*, **2014**, 111, 84-91	3.7	46
102	Identification of miRNAs that modulate glucocerebrosidase activity in Gaucher disease cells. *RNA Biology*, **2014**, 11, 1291-300	4.8	26
101	Macrophage models of Gaucher disease for evaluating disease pathogenesis and candidate drugs. *Science Translational Medicine*, **2014**, 6, 240ra73	17.5	72
100	Saposin C protects glucocerebrosidase against -synuclein inhibition. *Biochemistry*, **2013**, 52, 7161-3	3.2	32
99	Functional and genetic characterization of the non-lysosomal glucosylceramidase 2 as a modifier for Gaucher disease. *Orphanet Journal of Rare Diseases*, **2013**, 8, 151	4.2	23
98	Membrane-bound -synuclein interacts with glucocerebrosidase and inhibits enzyme activity. *Molecular Genetics and Metabolism*, **2013**, 108, 56-64	3.7	79
97	Is Parkinson disease associated with lysosomal integral membrane protein type-2?: challenges in interpreting association data. *Molecular Genetics and Metabolism*, **2013**, 108, 269-71	3.7	12
96	Predicting parkinsonism: new opportunities from Gaucher disease. *Molecular Genetics and Metabolism*, **2013**, 109, 235-6	3.7	4
95	A germline or de novo mutation in two families with Gaucher disease: implications for recessive disorders. *European Journal of Human Genetics*, **2013**, 21, 115-7	5.3	14
94	Atp13a2-deficient mice exhibit neuronal ceroid lipofuscinosis, limited -synuclein accumulation and age-dependent sensorimotor deficits. *Human Molecular Genetics*, **2013**, 22, 2067-82	5.6	104
93	Relationship between Gaucher disease and parkinsonism **2013**, 62-77		
92	A multicenter study of glucocerebrosidase mutations in dementia with Lewy bodies. *JAMA Neurology*, **2013**, 70, 727-35	17.2	285

91	A high throughput glucocerebrosidase assay using the natural substrate glucosylceramide. *Analytical and Bioanalytical Chemistry*, **2012**, 402, 731-9	4.4	23
90	The link between the GBA gene and parkinsonism. *Lancet Neurology, The*, **2012**, 11, 986-98	24.1	353
89	Non-iminosugar glucocerebrosidase small molecule chaperones. *MedChemComm*, **2012**, 3, 56-60	5	20
88	The role of saposin C in Gaucher disease. *Molecular Genetics and Metabolism*, **2012**, 106, 257-63	3.7	83
87	Discovery, structure-activity relationship, and biological evaluation of noninhibitory small molecule chaperones of glucocerebrosidase. *Journal of Medicinal Chemistry*, **2012**, 55, 5734-48	8.3	93
86	The neurobiology of glucocerebrosidase-associated parkinsonism: a positron emission tomography study of dopamine synthesis and regional cerebral blood flow. *Brain*, **2012**, 135, 2440-8	11.2	67
85	Induced pluripotent stem cell model recapitulates pathologic hallmarks of Gaucher disease. *Proceedings of the National Academy of Sciences of the United States of America*, **2012**, 109, 18054-9	11.5	87
84	High throughput screening for small molecule therapy for Gaucher disease using patient tissue as the source of mutant glucocerebrosidase. *PLoS ONE*, **2012**, 7, e29861	3.7	51
83	Gaucher disease: insights from a rare Mendelian disorder. *Discovery Medicine*, **2012**, 14, 273-81	2.5	72
82	The Lysosomal Storage Diseases **2012**, 1357-1361		
81	Identification of recombinant alleles using quantitative real-time PCR implications for Gaucher disease. *Journal of Molecular Diagnostics*, **2011**, 13, 401-5	5.1	10
80	Gaucher disease glucocerebrosidase and □synuclein form a bidirectional pathogenic loop in synucleinopathies. *Cell*, **2011**, 146, 37-52	56.2	895
79	Coinheritance of Gaucher disease and □thalassemia resulting in confusion between two inherited hematologic diseases. *Blood Cells, Molecules, and Diseases*, **2011**, 46, 88-91	2.1	6
78	Gaucher disease type 2: homozygosity for the mutation F331S in two unrelated consanguineous Muslim Arab patients with Gaucher disease from the Gaza and Jenin regions. *Blood Cells, Molecules, and Diseases*, **2011**, 47, 262-3	2.1	
77	Evaluation of quinazoline analogues as glucocerebrosidase inhibitors with chaperone activity. *Journal of Medicinal Chemistry*, **2011**, 54, 1033-58	8.3	54
76	Exploring the link between glucocerebrosidase mutations and parkinsonism. *Trends in Molecular Medicine*, **2011**, 17, 485-93	11.5	126
75	Mucolipidosis type IV: an update. *Molecular Genetics and Metabolism*, **2011**, 104, 206-13	3.7	67
74	Aggregation of □synuclein in brain samples from subjects with glucocerebrosidase mutations. *Molecular Genetics and Metabolism*, **2011**, 104, 185-8	3.7	57

73	Skin ultrastructural findings in type 2 Gaucher disease: diagnostic implications. *Molecular Genetics and Metabolism*, **2011**, 104, 631-6	3.7	25
72	Bilateral symmetrical cortical osteolytic lesions in two patients with Gaucher disease. *Skeletal Radiology*, **2011**, 40, 1611-5	2.7	6
71	A mutation in SCARB2 is a modifier in Gaucher disease. *Human Mutation*, **2011**, 32, 1232-8	4.7	52
70	Alpha-synuclein interacts with Glucocerebrosidase providing a molecular link between Parkinson and Gaucher diseases. *Journal of Biological Chemistry*, **2011**, 286, 28080-8	5.4	130
69	Fabry disease - current treatment and new drug development. *Current Chemical Genomics*, **2010**, 4, 50-6		32
68	Autosomal recessive mutations in the development of Parkinson's disease. *Biomarkers in Medicine*, **2010**, 4, 713-21	2.3	11
67	In silico and functional studies of the regulation of the glucocerebrosidase gene. *Molecular Genetics and Metabolism*, **2010**, 99, 275-82	3.7	9
66	False-positive results using a Gaucher diagnostic kit--RecTL and N370S. *Molecular Genetics and Metabolism*, **2010**, 100, 100-2	3.7	8
65	Glucocerebrosidase is present in □synuclein inclusions in Lewy body disorders. *Acta Neuropathologica*, **2010**, 120, 641-9	14.3	151
64	The role of glucocerebrosidase mutations in Parkinson disease and Lewy body disorders. *Current Neurology and Neuroscience Reports*, **2010**, 10, 190-8	6.6	112
63	Psychiatric and behavioral manifestations of lysosomal storage disorders. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2010**, 153B, 1253-65	3.5	25
62	Lysosomal storage disorders in the newborn. *Pediatrics*, **2009**, 123, 1191-207	7.4	121
61	Dosing enzyme replacement therapy for Gaucher disease: older, but are we wiser?. *Genetics in Medicine*, **2009**, 11, 90-1	8.1	14
60	Mutations in GBA are associated with familial Parkinson disease susceptibility and age at onset. *Neurology*, **2009**, 73, 1424-5, author reply 1425-6	6.5	45
59	The association between mutations in the lysosomal protein glucocerebrosidase and parkinsonism. *Movement Disorders*, **2009**, 24, 1571-8	7	61
58	Type 2 Gaucher disease occurs in Ashkenazi Jews but is surprisingly rare. *Blood Cells, Molecules, and Diseases*, **2009**, 43, 294-7	2.1	13
57	Complete screening for glucocerebrosidase mutations in Parkinson disease patients from Portugal. *Neurobiology of Aging*, **2009**, 30, 1515-7	5.6	79
56	Cognitive outcome in treated patients with chronic neuronopathic Gaucher disease. *Journal of Pediatrics*, **2008**, 153, 89-94	3.6	35

55	The need for appropriate genotyping strategies for glucocerebrosidase mutations in cohorts with Parkinson disease. *Archives of Neurology*, **2008**, 65, 850-1; author reply 851		9
54	Glucocerebrosidase gene mutations: a risk factor for Lewy body disorders. *Archives of Neurology*, **2008**, 65, 379-82		146
53	The spectrum of parkinsonian manifestations associated with glucocerebrosidase mutations. *Archives of Neurology*, **2008**, 65, 1353-7		142
52	Optimization and validation of two miniaturized glucocerebrosidase enzyme assays for high throughput screening. *Combinatorial Chemistry and High Throughput Screening*, **2008**, 11, 817-24	1.3	31
51	Gaucher disease: mutation and polymorphism spectrum in the glucocerebrosidase gene (GBA). *Human Mutation*, **2008**, 29, 567-83	4.7	426
50	N4-phenyl modifications of N2-(2-hydroxyl)ethyl-6-(pyrrolidin-1-yl)-1,3,5-triazine-2,4-diamines enhance glucocerebrosidase inhibition by small molecules with potential as chemical chaperones for Gaucher disease. *Bioorganic and Medicinal Chemistry Letters*, **2007**, 17, 5783-9	2.9	26
49	Three classes of glucocerebrosidase inhibitors identified by quantitative high-throughput screening are chaperone leads for Gaucher disease. *Proceedings of the National Academy of Sciences of the United States of America*, **2007**, 104, 13192-7	11.5	130
48	Therapy for Gaucher disease: don't stop thinking about tomorrow. *Molecular Genetics and Metabolism*, **2007**, 90, 122-5	3.7	13
47	Glucocerebrosidase mutations in Chinese subjects from Taiwan with sporadic Parkinson disease. *Molecular Genetics and Metabolism*, **2007**, 91, 195-200	3.7	93
46	Glucocerebrosidase mutations are also found in subjects with early-onset parkinsonism from Venezuela. *Movement Disorders*, **2006**, 21, 282-3	7	58
45	Glucocerebrosidase mutations are not found in association with LRRK2 G2019S in subjects with parkinsonism. *Neuroscience Letters*, **2006**, 404, 163-5	3.3	13
44	Enhanced calcium release in the acute neuronopathic form of Gaucher disease. *Neurobiology of Disease*, **2005**, 18, 83-8	7.5	125
43	Gaucher disease and parkinsonism. *Molecular Genetics and Metabolism*, **2005**, 84, 302-4	3.7	66
42	Gaucher mutation N188S is associated with myoclonic epilepsy. *Human Mutation*, **2005**, 26, 271-3; author reply 274-5	4.7	17
41	The glucocerebrosidase gene and Parkinson's disease in Ashkenazi Jews. *New England Journal of Medicine*, **2005**, 352, 728-31; author reply 728-31	59.2	82
40	Perinatal lethal Gaucher disease: a distinct phenotype along the neuronopathic continuum. *Fetal and Pediatric Pathology*, **2005**, 24, 205-22	1.7	34
39	A novel alteration in metaxin 1, F202L, is associated with N370S in Gaucher disease. *Journal of Human Genetics*, **2004**, 49, 220-222	4.3	4
38	Glucocerebrosidase mutations in subjects with parkinsonism. *Molecular Genetics and Metabolism*, **2004**, 81, 70-3	3.7	305

37	Neuropathology provides clues to the pathophysiology of Gaucher disease. *Molecular Genetics and Metabolism*, **2004**, 82, 192-207	3.7	360
36	Gaucher disease: complexity in a "simple" disorder. *Molecular Genetics and Metabolism*, **2004**, 83, 6-15	3.7	295
35	Myoclonic epilepsy in Gaucher disease: genotype-phenotype insights from a rare patient subgroup. *Pediatric Research*, **2003**, 53, 387-95	3.2	85
34	Gaucher disease associated with parkinsonism: four further case reports **2003**, 116A, 348-51		47
33	Phenotypic continuum in neuronopathic Gaucher disease: an intermediate phenotype between type 2 and type 3. *Journal of Pediatrics*, **2003**, 143, 273-6	3.6	112
32	Reciprocal and nonreciprocal recombination at the glucocerebrosidase gene region: implications for complexity in Gaucher disease. *American Journal of Human Genetics*, **2003**, 72, 519-34	11	80
31	Cholelithiasis in patients with Gaucher disease. *Blood Cells, Molecules, and Diseases*, **2002**, 28, 21-7	2.1	20
30	Glucosylsphingosine accumulation in tissues from patients with Gaucher disease: correlation with phenotype and genotype. *Molecular Genetics and Metabolism*, **2002**, 76, 262-70	3.7	135
29	Life-threatening splenic hemorrhage in two patients with Gaucher disease. *American Journal of Hematology*, **2000**, 64, 140-2	7.1	10
28	Glucocerebrosidase gene mutations in patients with type 2 Gaucher disease. *Human Mutation*, **2000** , 15, 181-8	4.7	179
27	Childhood-onset schizophrenia/autistic disorder and t(1;7) reciprocal translocation: identification of a BAC contig spanning the translocation breakpoint at 7q21. *American Journal of Medical Genetics Part A*, **2000**, 96, 749-53		62
26	Gene rearranagement on 1q21 introducing a duplication of the glucocerebrosidase pseudogene and a metaxin fusion gene. *Human Genetics*, **2000**, 107, 400-3	6.3	12
25	Glucosylsphingosine accumulation in mice and patients with type 2 Gaucher disease begins early in gestation. *Pediatric Research*, **2000**, 48, 233-7	3.2	81
24	Analysis and classification of 304 mutant alleles in patients with type 1 and type 3 Gaucher disease. *American Journal of Human Genetics*, **2000**, 66, 1777-86	11	243
23	Epidermal sphingomyelins are precursors for selected stratum corneum ceramides. *Journal of Lipid Research*, **2000**, 41, 2071-2082	6.3	167
22	Glucocerebrosidase gene mutations in patients with type 2 Gaucher disease **2000**, 15, 181		3
21	Is the perinatal lethal form of Gaucher disease more common than classic type 2 Gaucher disease?. *European Journal of Human Genetics*, **1999**, 7, 505-9	5.3	35
20	Apolipoprotein E alleles in childhood-onset schizophrenia **1999**, 88, 211-213		17

19	Type 2 gaucher disease: an expanding phenotype. *Molecular Genetics and Metabolism*, **1999**, 68, 209-19	3.7	38
18	Apolipoprotein E alleles in childhood-onset schizophrenia **1999**, 88, 211		1
17	Chromosome 22q11.2 interstitial deletions among childhood-onset schizophrenics and multidimensionally impaired **1998**, 81, 41-43		44
16	Genotypic heterogeneity and phenotypic variation among patients with type 2 Gaucher's disease. *Pediatric Research*, **1998**, 43, 571-8	3.2	39
15	Identification of three additional genes contiguous to the glucocerebrosidase locus on chromosome 1q21: implications for Gaucher disease. *Genome Research*, **1997**, 7, 1020-6	9.7	82
14	Prenatal lethality of a homozygous null mutation in the human glucocerebrosidase gene. *American Journal of Medical Genetics Part A*, **1997**, 73, 41-7		35
13	Genotype D399N/R463C in a patient with type 3 Gaucher disease previously assigned genotype N370S/R463C. *Biochemical and Molecular Medicine*, **1996**, 57, 149-51		13
12	Epidermal abnormalities may distinguish type 2 from type 1 and type 3 of Gaucher disease. *Pediatric Research*, **1996**, 39, 134-41	3.2	70
11	Gaucher disease: A tale of two species. *Mental Retardation and Developmental Disabilities Research Reviews*, **1995**, 1, 79-86		4
10	Congenital ichthyosis preceding neurologic symptoms in two sibs with type 2 Gaucher disease. *American Journal of Medical Genetics Part A*, **1995**, 59, 356-8		33
9	Phenotypic and genotypic heterogeneity in gaucher disease: Implications for genetic counseling. *Journal of Genetic Counseling*, **1994**, 3, 13-22	2.5	10
8	DNA mutational analysis of type 1 and type 3 Gaucher patients: how well do mutations predict phenotype?. *Human Mutation*, **1994**, 3, 25-8	4.7	52
7	Pathologic fractures may develop in Gaucher patients receiving enzyme replacement therapy. *American Journal of Hematology*, **1994**, 47, 247-9	7.1	15
6	Clinical Heterogeneity Among Patients With Gaucher's Disease. *JAMA - Journal of the American Medical Association*, **1993**, 269, 1154	27.4	30
5	Gaucher patients with oculomotor abnormalities do not have a unique genotype. *Clinical Genetics*, **1992**, 41, 1-5	4	20
4	Gaucher disease in the neonate: a distinct Gaucher phenotype is analogous to a mouse model created by targeted disruption of the glucocerebrosidase gene. *Pediatric Research*, **1992**, 32, 494-8	3.2	124
3	Erythropoietin levels in Gaucher patients. *American Journal of Hematology*, **1992**, 40, 153-4	7.1	7
2	DNA mutation analysis of Gaucher patients. *American Journal of Medical Genetics Part A*, **1992**, 42, 331-6		50

1 Reply to Drs. Beutler and Gelbart. *American Journal of Medical Genetics Part A*, **1992**, 44, 390-390