Mirella Filocamo

List of Publications by Year in descending order

161 papers	5,481 citations	101543 36 h-index	123424 61 g-index
166 all docs	166 docs citations	166 times ranked	7059 citing authors

#	Article	IF	Citations
1	Germline mutations in HRAS proto-oncogene cause Costello syndrome. Nature Genetics, 2005, 37, 1038-1040.	21.4	597
2	Early Visual Seizures and Progressive Myoclonus Epilepsy in Neuronopathic Gaucher Disease Due to a Rare Compound Heterozygosity (N188S/S107L). Epilepsia, 2004, 45, 1154-1157.	5.1	148
3	Ambroxol as a pharmacological chaperone for mutant glucocerebrosidase. Blood Cells, Molecules, and Diseases, 2013, 50, 141-145.	1.4	116
4	Mutation profile of theGAA gene in 40 Italian patients with late onset glycogen storage disease type II. Human Mutation, 2006, 27, 999-1006.	2.5	115
5	GM1 gangliosidosis and Morquio B disease: An update on genetic alterations and clinical findings. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2011, 1812, 782-790.	3.8	115
6	Pharmacological Enhancement of Mutated Î±-Glucosidase Activity in Fibroblasts from Patients with Pompe Disease. Molecular Therapy, 2007, 15, 508-514.	8.2	108
7	Pseudogene-mediated posttranscriptional silencing of HMGA1 can result in insulin resistance and type 2 diabetes. Nature Communications, 2010, 1, 40.	12.8	102
8	Lysosomal storage disorders: Molecular basis and laboratory testing. Human Genomics, 2011, 5, 156.	2.9	100
9	Mutation Update ofARSAandPSAPGenes Causing Metachromatic Leukodystrophy. Human Mutation, 2016, 37, 16-27.	2.5	96
10	Identification and characterization of 15 novel GALC gene mutations causing Krabbe disease. Human Mutation, 2010, 31, E1894-E1914.	2.5	93
11	Characterization of the ERAD process of the L444P mutant glucocerebrosidase variant. Blood Cells, Molecules, and Diseases, 2011, 46, 4-10.	1.4	91
12	Unfolded protein response in Gaucher disease: from human to Drosophila. Orphanet Journal of Rare Diseases, 2013, 8, 140.	2.7	88
13	Functional Variants of the <emph type="ital">HMGA1</emph> Gene and Type 2 Diabetes Mellitus. JAMA - Journal of the American Medical Association, 2011, 305, 903.	7.4	87
14	Lysosomal lipase deficiency: Molecular characterization of eleven patients with Wolman or cholesteryl ester storage disease. Molecular Genetics and Metabolism, 2012, 105, 450-456.	1.1	71
15	The role of a bioresource research impact factor as an incentive to share human bioresources. Nature Genetics, 2011, 43, 503-504.	21.4	66
16	IDUA mutational profiling of a cohort of 102 European patients with mucopolysaccharidosis type I: identification and characterization of 35 novel Î±-L-iduronidase (IDUA) alleles. Human Mutation, 2011, 32, E2189-E2210.	2.5	66
17	SMPD1 Mutation Update: Database and Comprehensive Analysis of Published and Novel Variants. Human Mutation, 2016, 37, 139-147.	2.5	66
18	Molecular analysis and characterization of nine novel CTSK mutations in twelve patients affected by pycnodysostosis. Human Mutation, 2007, 28, 524-524.	2.5	64

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19	Molecular and functional analysis ofSUMF1 mutations in multiple sulfatase deficiency. Human Mutation, 2004, 23, 576-581.	2.5	63
20	The EuroBioBank Network: 10 years of hands-on experience of collaborative, transnational biobanking for rare diseases. European Journal of Human Genetics, 2015, 23, 1116-1123.	2.8	63
21	Clinical and molecular findings in patients with giant axonal neuropathy (GAN). Neurology, 2004, 62, 13-16.	1.1	62
22	Mutations in MFSD8/CLN7 are a frequent cause of variant-late infantile neuronal ceroid lipofuscinosis. Human Mutation, 2009, 30, E530-E540.	2.5	59
23	Wolman disease and cholesteryl ester storage disease diagnosed by histological and ultrastructural examination of intestinal and liver biopsy. Pathology Research and Practice, 2004, 200, 231-240.	2.3	56
24	Analysis of the glucocerebrosidase gene and mutation profile in 144 Italian gaucher patients. Human Mutation, 2002, 20, 234-235.	2.5	51
25	Acid sphingomyelinase: Identification of nine novel mutations among Italian Niemann Pick type B patients and characterization ofin vivofunctional in-frame start codon. Human Mutation, 2004, 24, 186-187.	2.5	51
26	Mutational analysis of theHGSNATgene in Italian patients with mucopolysaccharidosis IIIC (Sanfilippo) Tj ETQq0 0 0 rgBT /Overlock 10 T	2.5	51
27	Molecular and functional characterization of eight novel GAA mutations in Italian infants with Pompe disease. Human Mutation, 2008, 29, E27-E36.	2.5	51
28	First pilot newborn screening for four lysosomal storage diseases in an Italian region: Identification and analysis of a putative causative mutation in the GBA gene. Clinica Chimica Acta, 2012, 413, 1827-1831.	1.1	50
29	Pulmonary Manifestations of Gaucher Disease. American Journal of Respiratory and Critical Care Medicine, 1998, 157, 985-989.	5.6	48
30	Glucocerebrosidase deficiency in zebrafish affects primary bone ossification through increased oxidative stress and reduced Wnt/Î²-catenin signaling. Human Molecular Genetics, 2015, 24, 1280-1294.	2.9	46
31	Functional analysis of 11 novel GBA alleles. European Journal of Human Genetics, 2014, 22, 511-516.	2.8	44
32	Molecular analysis of 40 Italian patients with mucopolysaccharidosis type II: New mutations in the iduronate-2-sulfatase (IDS) gene. Human Mutation, 2001, 18, 164-165.	2.5	43
33	Molecular epidemiology of childhood neuronal ceroid-lipofuscinosis in Italy. Orphanet Journal of Rare Diseases, 2013, 8, 19.	2.7	42
34	Enhancing Cranial Nerves and Cauda Equina: An Emerging Magnetic Resonance Imaging Pattern in Metachromatic Leukodystrophy and Krabbe Disease. Neuropediatrics, 2009, 40, 291-294.	0.6	40
35	The alliance between genetic biobanks and patient organisations: the experience of the telethon network of genetic biobanks. Orphanet Journal of Rare Diseases, 2016, 11, 142.	2.7	40
36	Molecular analysis of NPC1 and NPC2 gene in 34 Niemannâ€“Pick C Italian Patients: identification and structural modeling of novel mutations. Neurogenetics, 2009, 10, 229-239.	1.4	39

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37	Telethon Network of Genetic Biobanks: a key service for diagnosis and research on rare diseases. Orphanet Journal of Rare Diseases, 2013, 8, 129.	2.7	39
38	Mucopolysaccharidosis IVA (Morquio A): Identification of novel common mutations in the N-acetylgalactosamine-6-sulfate sulfatase (GALNS) gene in Italian patients. Human Mutation, 2004, 24, 187-188.	2.5	38
39	Hepatic and neuromuscular forms of glycogenosis type III: nine mutations inAGL. Human Mutation, 2006, 27, 600-601.	2.5	38
40	Molecular characterization of 22 novel UDP-N-acetylglucosamine-1-phosphate transferase Î±- and Î²-subunit (GNPTAB) gene mutations causing mucolipidosis types IIÎ±/Î² and IIIÎ±/Î² in 46 patients. Human Mutation, 2009, 30, E956-E973.	2.5	38
41	Quantifying the use of bioresources for promoting their sharing in scientific research. GigaScience, 2013, 2, 7.	6.4	38
42	Characterization of iduronate-2-sulfatase gene-pseudogene recombinations in eight patients with Mucopolysaccharidosis type II revealed by a rapid PCR-based method. Human Mutation, 2005, 25, 491-497.	2.5	37
43	Molecular analysis of theHEXAgene in Italian patients with infantile and late Onset Tay-Sachs disease: detection of fourteen novel alleles. Human Mutation, 2005, 26, 282-282.	2.5	37
44	Type II sialidosis: review of the clinical spectrum and identification of a new splicing defect with chitotriosidase assessment in two patients. Journal of Neurology, 2009, 256, 1911-1915.	3.6	37
45	Human iPSC-based models highlight defective glial and neuronal differentiation from neural progenitor cells in metachromatic leukodystrophy. Cell Death and Disease, 2018, 9, 698.	6.3	37
46	The lysosomal storage disorders mucolipidosis type II, type III alpha/beta, and type III gamma: Update onGNPTABandGNPTGmutations. Human Mutation, 2019, 40, 842-864.	2.5	36
47	Molecular analysis of 30 mucopolysaccharidosis type I patients: evaluation of the mutational spectrum in Italian population and identification of 13 novel mutations. Human Mutation, 2002, 20, 231-231.	2.5	35
48	Functionalin vitro characterization of 14SMPD1 mutations identified in Italian patients affected by Niemann Pick Type B disease. Human Mutation, 2005, 26, 164-164.	2.5	35
49	Cell surface associated glycohydrolases in normal and Gaucher disease fibroblasts. Journal of Inherited Metabolic Disease, 2012, 35, 1081-1091.	3.6	35
50	Mutations among Italian mucopolysaccharidosis type I patients. Journal of Inherited Metabolic Disease, 1997, 20, 803-806.	3.6	34
51	Different Molecular Mechanisms Leading to White Matter Hypomyelination in Infantile Onset Lysosomal Disorders. Neuropediatrics, 2005, 36, 265-269.	0.6	34
52	A multicentre observational study for early diagnosis of Gaucher disease in patients with Splenomegaly and/or Thrombocytopenia. European Journal of Haematology, 2016, 96, 352-359.	2.2	34
53	Perturbations in cell signaling elicit early cardiac defects in mucopolysaccharidosis type II. Human Molecular Genetics, 2017, 26, 1643-1655.	2.9	34
54	Biochemical and molecular analysis in mucopolysaccharidoses: what a paediatrician must know. Italian Journal of Pediatrics, 2018, 44, 129.	2.6	34

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55	Unbalanced GLA mRNAs ratio quantified by real-time PCR in Fabry patients' fibroblasts results in Fabry disease. European Journal of Human Genetics, 2008, 16, 1311-1317.	2.8	33
56	Mucopolysaccharidosis type IIID: 12 new patients and 15 novel mutations. Human Mutation, 2010, 31, n/a-n/a.	2.5	33
57	The RD-Connect Registry & Biobank Finder: a tool for sharing aggregated data and metadata among rare disease researchers. European Journal of Human Genetics, 2018, 26, 631-643.	2.8	33
58	An Asn > Lys substitution in saposin B involving a conserved amino acidic residue and leading to the loss of the single N-glycosylation site in a patient with metachromatic leukodystrophy and normal arylsulphatase A activity. European Journal of Human Genetics, 1999, 7, 125-130.	2.8	32
59	Basic Fibroblast Growth Factor Activates Endothelial Nitric-Oxide Synthase in CHO-K1 Cells via the Activation of Ceramide Synthesis. Molecular Pharmacology, 2003, 63, 297-310.	2.3	32
60	Molecular Genetic Analysis of the PLP1 Gene in 38 Families with PLP1-related disorders: Identification and Functional Characterization of 11 Novel PLP1 Mutations. Orphanet Journal of Rare Diseases, 2011, 6, 40.	2.7	32
61	Biochemical and molecular findings in a patient with myoclonic epilepsy due to a mistarget of the Î²-glucosidase enzyme. Molecular Genetics and Metabolism, 2009, 97, 309-311.	1.1	31
62	SMAD4 mutations causing Myhre syndrome result in disorganization of extracellular matrix improved by losartan. European Journal of Human Genetics, 2014, 22, 988-994.	2.8	31
63	Molecular defects in the Î±-N-acetylglucosaminidase gene in Italian Sanfilippo type B patients. Human Genetics, 2000, 107, 568-576.	3.8	30
64	Contribution of arylsulfatase A mutations located on the same allele to enzyme activity reduction and metachromatic leukodystrophy severity. Human Genetics, 2002, 110, 351-355.	3.8	30
65	An Alu-mediated rearrangement as cause of exon skipping in Hunter disease. Human Genetics, 2003, 112, 419-425.	3.8	30
66	Leukoencephalopathy with vanishing white matter:. Neurology, 2003, 61, 1818-1819.	1.1	30
67	Genetic disorders affecting white matter in the pediatric age. American Journal of Medical Genetics Part A, 2004, 129B, 85-93.	2.4	30
68	Expanded spectrum of Pelizaeusâ€“Merzbacher-like disease: literature revision and description of a novel GJC2 mutation in an unusually severe form. European Journal of Human Genetics, 2013, 21, 34-39.	2.8	30
69	GFAP mutations and polymorphisms in 13 unrelated Italian patients affected by Alexander disease. Clinical Genetics, 2007, 72, 427-433.	2.0	29
70	Screening of 25 Italian patients with Niemann-Pick a reveals fourteen new mutations, one common and thirteen private, inSMPD1. Human Mutation, 2004, 24, 105-105.	2.5	28
71	Cerebellar atrophy without cerebellar cortex hyperintensity in infantile neuroaxonal dystrophy (INAD) due to PLA2G6 mutation. European Journal of Paediatric Neurology, 2007, 11, 175-177.	1.6	28
72	Molecular analysis ofARSAandPSAPgenes in twenty-one Italian patients with metachromatic leukodystrophy: identification and functional characterization of 11 novelARSAalleles. Human Mutation, 2008, 29, E220-E230.	2.5	28

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73	Identification of nine new IDS alleles in mucopolysaccharidosis II. Quantitative evaluation by real-time RT-PCR of mRNAs sensitive to nonsense-mediated and nonstop decay mechanisms. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2006, 1762, 478-484.	3.8	27
74	GM1 gangliosidosis: molecular analysis of nine patients and development of an RT-PCR assay for GLB1 gene expression profiling. Human Mutation, 2007, 28, 204-204.	2.5	27
75	Krabbe leukodystrophy in a selected population with high rate of late onset forms: longer survival linked to c.121G>A (p.Gly41Ser) mutation. Clinical Genetics, 2011, 80, 452-458.	2.0	27
76	FGF signaling deregulation is associated with early developmental skeletal defects in animal models for mucopolysaccharidosis type II (MPSII). Human Molecular Genetics, 2018, 27, 2262-2275.	2.9	27
77	Comparative study of 15 lysosomal enzymes in chorionic villi and cultured amniotic fluid cells. Early prenatal diagnosis in seven pregnancies at risk for lysosomal storage diseases. Prenatal Diagnosis, 1985, 5, 329-336.	2.3	26
78	Identification and molecular characterization of six novel mutations in the UDP-N-acetylglucosamine-1-phosphotransferase gamma subunit (GNPTG) gene in patients with mucolipidosis III gamma. Human Mutation, 2009, 30, 978-984.	2.5	26
79	Optimizing the Molecular Diagnosis of GALNS: Novel Methods to Define and Characterize Morquio-A Syndrome-Associated Mutations. Human Mutation, 2015, 36, 357-368.	2.5	26
80	PLP1 gene duplication causes overexpression and alteration of the PLP/DM20 splicing balance in fibroblasts from Pelizaeusâ€“Merzbacher disease patients. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2009, 1792, 548-554.	3.8	25
81	Functional and genetic characterization of the non-lysosomal glucosylceramidase 2 as a modifier for Gaucher disease. Orphanet Journal of Rare Diseases, 2013, 8, 151.	2.7	24
82	Severity of bone marrow involvement in patients with Gaucher's disease evaluated by scintigraphy with 99mTc-sestamibi. Journal of Nuclear Medicine, 2003, 44, 1253-62.	5.0	24
83	Identification and functional characterization of five novel mutant alleles in 58 Italian patients with Gaucher disease type 1. Human Mutation, 2005, 25, 100-100.	2.5	22
84	An Alu insertion in compound heterozygosity with a microduplication in GNPTAB gene underlies Mucolipidosis II. Molecular Genetics and Metabolism, 2008, 93, 129-133.	1.1	22
85	Molecular and functional analysis of the HEXB gene in Italian patients affected with Sandhoff disease: identification of six novel alleles. Neurogenetics, 2009, 10, 49-58.	1.4	22
86	Sequence and Copy Number Analyses of HEXB Gene in Patients Affected by Sandhoff Disease: Functional Characterization of 9 Novel Sequence Variants. PLoS ONE, 2012, 7, e41516.	2.5	22
87	Multiple cryptic splice sites can be activated by IDS point mutations generating misspliced transcripts. Journal of Molecular Medicine, 2006, 84, 692-700.	3.9	21
88	Craniosynostosis: A rare complication of pycnodysostosis. European Journal of Medical Genetics, 2010, 53, 89-92.	1.3	21
89	Treatment of Human Fibroblasts Carrying NPC1 Missense Mutations with MG132 Leads to an Improvement of Intracellular Cholesterol Trafficking. JIMD Reports, 2011, 2, 59-69.	1.5	21
90	Gene expression profile in patients with Gaucher disease indicates activation of inflammatory processes. Scientific Reports, 2019, 9, 6060.	3.3	21

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91	Mild functional effects of a novel GFAP mutant allele identified in a familial case of adult-onset Alexander disease. European Journal of Human Genetics, 2008, 16, 462-470.	2.8	20
92	Detection of carriers and prenatal diagnosis for fucosidosis in Calabria. Human Genetics, 1979, 51, 195-201.	3.8	19
93	Galactosemia caused by generalized uridine diphosphate galactose-4-epimerase deficiency. Journal of Pediatrics, 1983, 103, 927-930.	1.8	19
94	Prenatal diagnosis of Pelizaeus-Merzbacher disease: detection of proteolipid protein gene duplication by quantitative fluorescent multiplex PCR. Prenatal Diagnosis, 2001, 21, 668-671.	2.3	19
95	Genotypeâ€“phenotype correlation in five Pelizaeusâ€“Merzbacher disease patients with PLP1 gene duplications. Clinical Genetics, 2008, 73, 279-287.	2.0	19
96	Critical issues for the proper diagnosis of Metachromatic Leukodystrophy. Gene, 2014, 537, 348-351.	2.2	19
97	Evidence for a Founder Effect in Sicilian Patients with Glycogen Storage Disease Type II. Human Heredity, 2000, 50, 331-333.	0.8	18
98	Diagnosis of Pelizaeusâ€“Merzbacher disease: detection of proteolipid protein gene copy number by real-time PCR. Neurogenetics, 2005, 6, 73-78.	1.4	18
99	Haplotype analysis suggests a single Balkan origin for the Gaucher disease [D409H;H255Q] double mutant allele. Human Mutation, 2008, 29, E58-E67.	2.5	18
100	ASAH1 variant causing a mild SMA phenotype with no myoclonic epilepsy: a clinical, biochemical and molecular study. European Journal of Human Genetics, 2016, 24, 1578-1583.	2.8	18
101	UPR activation and CHOP mediated induction of GBA1 transcription in Gaucher disease. Blood Cells, Molecules, and Diseases, 2018, 68, 21-29.	1.4	18
102	Enigmatic In Vivo iduronate-2-sulfatase (IDS) mutant transcript correction to wild-type in Hunter syndrome. Human Mutation, 2010, 31, E1261-E1285.	2.5	17
103	A Novel DHPLC-Based Procedure for the Analysis of COL1A1 and COL1A2 Mutations in Osteogenesis Imperfecta. Journal of Molecular Diagnostics, 2011, 13, 648-656.	2.8	17
104	Mucopolysaccharidosis type II in a female patient with a reciprocal X;9 translocation and skewed X chromosome inactivation. American Journal of Medical Genetics, Part A, 2014, 164, 2627-2632.	1.2	17
105	A 9-bp deletion (2320del9) on the background of the arylsulfatase A pseudodeficiency allele in a metachromatic leukodystrophy patient and in a patient with nonprogressive neurological symptoms. Human Genetics, 1998, 102, 50-53.	3.8	16
106	The effect of four mutations on the expression of iduronate-2-sulfatase in mucopolysaccharidosis type II. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2001, 1537, 233-238.	3.8	16
107	ITCH regulates degradation of mutant glucocerebrosidase: implications to Gaucher disease. Human Molecular Genetics, 2013, 22, 1316-1327.	2.9	16
108	Validity of Î²-d-glucosidase activity measured in dried blood samples for detection of potential Gaucher disease patients. Clinical Biochemistry, 2014, 47, 1293-1296.	1.9	16

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109	Origin and spread of a common deletion causing mucolipidosis type II: insights from patterns of haplotypic diversity. Clinical Genetics, 2011, 80, 273-280.	2.0	15
110	Changes in global gene expression indicate disordered autophagy, apoptosis and inflammatory processes and downregulation of cytoskeletal signalling and neuronal development in patients with Niemannâ€“Pick C disease. Neurogenetics, 2020, 21, 105-119.	1.4	15
111	Expression studies of two novel in CIS-mutations identified in an intermediate case of Hunter syndrome. American Journal of Medical Genetics Part A, 2003, 120A, 84-87.	2.4	14
112	Homozygosity for the p.K136E mutation in the SLC17A5 gene as cause of an Italian severe Salla disease. Neurogenetics, 2005, 6, 195-199.	1.4	14
113	Segregation analysis in a family at risk for the Maroteauxâ€“Lamy syndrome conclusively reveals c.1151G>A (p.S384N) as to be a polymorphism. European Journal of Human Genetics, 2009, 17, 1160-1164.	2.8	14
114	A Novel Polymorphic APâ€1 Binding Element of the GFAP Promoter is Associated with Different Allelic Transcriptional Activities. Annals of Human Genetics, 2010, 74, 506-515.	0.8	14
115	An Alu-mediated duplication in NMNAT1, involved in NAD biosynthesis, causes a novel syndrome, SHILCA, affecting multiple tissues and organs. Human Molecular Genetics, 2020, 29, 2250-2260.	2.9	14
116	A deletion involving exons 2â€“4 in the iduronateâ€2â€sulfatase gene of a patient with intermediate Hunter syndrome. Clinical Genetics, 1998, 53, 474-477.	2.0	13
117	Carbon nanotubes as nanovectors for intracellular delivery of laronidase in Mucopolysaccharidosis type I. Nanoscale, 2018, 10, 657-665.	5.6	13
118	Prenatal diagnosis of mucopolysaccharidosis I: A special difficulty arising from an unusually low enzyme activity in mother's cells. Prenatal Diagnosis, 1985, 5, 149-154.	2.3	12
119	Deletion of exons 11-17 and novel mutations of the galactocerebrosidase gene in adult- and early-onset patients with Krabbe disease. Journal of Neurology, 2000, 247, 875-877.	3.6	12
120	Identification of a Novel Recombinant Allele in Three Unrelated Italian Gaucher Patients: Implications for Prognosis and Genetic Counseling. Blood Cells, Molecules, and Diseases, 2000, 26, 307-311.	1.4	12
121	Nitric Oxide Production Stimulated by the Basic Fibroblast Growth Factor Requires the Synthesis of Ceramide. Annals of the New York Academy of Sciences, 2002, 973, 94-104.	3.8	12
122	An unusual arylsulfatase A pseudodeficiency allele carrying a splice site mutation in a metachromatic leukodystrophy patient. European Journal of Human Genetics, 2004, 12, 150-154.	2.8	12
123	Restoration of the Normal Splicing Pattern of the PLP1 Gene by Means of an Antisense Oligonucleotide Directed against an Exonic Mutation. PLoS ONE, 2013, 8, e73633.	2.5	12
124	A new case report of severe mucopolysaccharidosis type VII: diagnosis, treatment with haematopoietic cell transplantation and prenatal diagnosis in a second pregnancy. Italian Journal of Pediatrics, 2018, 44, 128.	2.6	12
125	Predicting the probability of Gaucher disease in subjects with splenomegaly and thrombocytopenia. Scientific Reports, 2021, 11, 2594.	3.3	12
126	An AT-deletion causing a frameshift in the arylsulfatase A gene of a late infantile metachromatic leukodystrophy patient. Human Genetics, 1995, 96, 233-235.	3.8	11

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127	Somatic Mosaicism in a Patient with Gaucher Disease Type 2: Implication for Genetic Counseling and Therapeutic Decision-Making. Blood Cells, Molecules, and Diseases, 2000, 26, 611-612.	1.4	11
128	Identification and characterisation of an 8.7kb deletion and a novel nonsense mutation in two Italian families with Sanfilippo syndrome type D (mucopolysaccharidosis IIID). Molecular Genetics and Metabolism, 2007, 90, 77-80.	1.1	11
129	Funtional characterization of four novel MAN2B1 mutations causing juvenile onset alpha-mannosidosis. Clinica Chimica Acta, 2007, 375, 136-139.	1.1	11
130	A rare G6490 â†' substitution at the last nucleotide of exon 10 of the glucocerebrosidase gene in two unrelated Italian Gaucher patients. Clinical Genetics, 1995, 48, 123-127.	2.0	11
131	AIMP1/p43 Mutation and PMLD. American Journal of Human Genetics, 2011, 88, 391.	6.2	11
132	SjÃ¶gren-Larsson syndrome: Nuclear magnetic resonance imaging of the brain in a 4-year-old boy. Journal of Inherited Metabolic Disease, 1994, 17, 112-114.	3.6	10
133	Aberrant splicing at catalytic site as cause of infantile onset glycogen storage disease type II (GSDII): Molecular identification of a novel IVS9 (+2GT?GC) in combination with rare IVS10 (+1GT?CT). American Journal of Medical Genetics Part A, 2001, 101, 55-58.	2.4	10
134	Homozygosity for a non-pseudogene complex glucocerebrosidase allele as cause of an atypical neuronopathic form of Gaucher disease. American Journal of Medical Genetics, Part A, 2005, 134A, 95-96.	1.2	10
135	Movement and mood disorder in two brothers with Gaucher disease. Clinical Genetics, 2007, 72, 357-361.	2.0	10
136	Functional Characterization of the Novel Mutation IVS 8 (â^'11delC) (â^'14T>A) in the Intron 8 of the Glucocerebrosidase Gene of Two Italian Siblings with Gaucher Disease Type I. Blood Cells, Molecules, and Diseases, 2000, 26, 171-176.	1.4	9
137	Prenatal diagnosis of Sanfilippo type A syndrome in a family with S66W mutant allele. , 1999, 19, 993-994.		8
138	First-trimester fetal nuchal translucency and inherited metabolic disorders. Prenatal Diagnosis, 2006, 26, 77-80.	2.3	8
139	A novel homozygous splicing mutation in PSAP gene causes metachromatic leukodystrophy in two Moroccan brothers. Neurogenetics, 2014, 15, 101-106.	1.4	8
140	A novel homozygous MCOLN1 double mutant allele leading to TRP channel domain ablation underlies Mucolipidosis IV in an Italian Child. Metabolic Brain Disease, 2015, 30, 681-686.	2.9	8
141	Unusual white matter involvement in EAST syndrome associated with novel KCNJ10 mutations. Journal of Neurology, 2018, 265, 1419-1425.	3.6	8
142	Further genotypeâ€"phenotype correlation emerging from two families with PLP1 exon 4 skipping. Clinical Genetics, 2014, 85, 267-272.	2.0	7
143	Phenotypic variability and abnormal type I collagen unstable at body temperature in a family with mild dominant osteogenesis imperfecta. Journal of Inherited Metabolic Disease, 1991, 14, 189-201.	3.6	6
144	A novel mutation, Y103X, and exon skipping in a patient with Hunter disease. , 2000, 15, 389-389.		5

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145	A T > C transition causing a Leu > Pro substitution in a conserved region of the arylsulfatase A gene in a late infantile metachromatic leukodystrophy patient. Clinical Genetics, 1997, 52, 65-67.	2.0	5
146	A novel mutation of the Î²â€glucocererebrosidase gene associated with neurologic manifestations in three sibs. Clinical Genetics, 1998, 53, 281-285.	2.0	5
147	MLPA-based approach for initial and simultaneous detection of GBA deletions and recombinant alleles in patients affected by Gaucher Disease. Molecular Genetics and Metabolism, 2016, 119, 329-337.	1.1	5
148	Neonatal chitotriosidase activity is not predictive for Niemannâ€“Pick disease type A/B: Implications for newborn screening for lysosomal storage disorders. Molecular Genetics and Metabolism, 2013, 108, 106.	1.1	4
149	99mTc-Sestamibi Scintigraphy to Monitor the Long-Term Efficacy of Enzyme Replacement Therapy on Bone Marrow Infiltration in Patients with Gaucher Disease. Journal of Nuclear Medicine, 2013, 54, 1717-1724.	5.0	4
150	A transcriptional and post-transcriptional dysregulation of Dishevelled 1 and 2 underlies the Wnt signaling impairment in type I Gaucher disease experimental models. Human Molecular Genetics, 2020, 29, 274-285.	2.9	4
151	Somatic intragenic recombination of the arylsulfatase A gene in a metachromatic leukodystrophy patient. Molecular Genetics and Metabolism, 2006, 89, 150-155.	1.1	3
152	Spontaneous regression of hypertrophic cardiomyopathy in an infant with Pompe's disease. Molecular Genetics and Metabolism, 2012, 107, 763.	1.1	3
153	Genomic Structure of the Human UDP-GlcNAc:dolichol-P GlcNAc-1-P Transferase Gene. DNA Sequence, 2002, 13, 245-250.	0.7	2
154	Gaucher disease phenotype. Journal of Pediatrics, 2004, 145, 860.	1.8	2
155	Norrbottnian clinical variant of Gaucher disease in Southern Italy. Journal of Human Genetics, 2017, 62, 507-511.	2.3	2
156	Rare compound heterozygosity for IVS2 +1G>A and R170P in an Italian patient with Gaucher disease type 1. Clinical Genetics, 2003, 64, 261-262.	2.0	1
157	Mutation identification of Fabry disease in families with other lysosomal storage disorders. Clinical Genetics, 2013, 84, 281-285.	2.0	1
158	A Multicenter Observational Study For Early Diagnosis Of Gaucher Disease In Patients With Splenomegaly and/Or Thrombocytopenia. Blood, 2013, 122, 4712-4712.	1.4	1
159	Governance of Access in Biobanking: The Case of Telethon Network of Genetic Biobanks. Biopreservation and Biobanking, 2021, 19, 483-492.	1.0	1
160	Molecular analyses in pursuit of a diagnosis of Gaucher disease. Clinical Therapeutics, 2009, 31, S177-S178.	2.5	0
161	In vitro recapitulation of the site-specific editing (to wild-type) of mutant IDS mRNA transcripts, and the characterization of IDS protein translated from the edited mRNAs. Human Mutation, 2017, 38, 849-862.	2.5	0