

Mirella Filocamo

List of Publications by Year in descending order

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161
papers

5,481
citations

101384

36
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123241

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. <i>Nature Genetics</i> , 2005, 37, 1038-1040.	9.4	597
2	Early Visual Seizures and Progressive Myoclonus Epilepsy in Neuronopathic Gaucher Disease Due to a Rare Compound Heterozygosity (N188S/S107L). <i>Epilepsia</i> , 2004, 45, 1154-1157.	2.6	148
3	Ambroxol as a pharmacological chaperone for mutant glucocerebrosidase. <i>Blood Cells, Molecules, and Diseases</i> , 2013, 50, 141-145.	0.6	116
4	Mutation profile of the GAA gene in 40 Italian patients with late onset glycogen storage disease type II. <i>Human Mutation</i> , 2006, 27, 999-1006.	1.1	115
5	GM1 gangliosidosis and Morquio B disease: An update on genetic alterations and clinical findings. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2011, 1812, 782-790.	1.8	115
6	Pharmacological Enhancement of Mutated α -Glucosidase Activity in Fibroblasts from Patients with Pompe Disease. <i>Molecular Therapy</i> , 2007, 15, 508-514.	3.7	108
7	Pseudogene-mediated posttranscriptional silencing of HMGA1 can result in insulin resistance and type 2 diabetes. <i>Nature Communications</i> , 2010, 1, 40.	5.8	102
8	Lysosomal storage disorders: Molecular basis and laboratory testing. <i>Human Genomics</i> , 2011, 5, 156.	1.4	100
9	Mutation Update of <i>ARSA</i> and <i>PSAP</i> Genes Causing Metachromatic Leukodystrophy. <i>Human Mutation</i> , 2016, 37, 16-27.	1.1	96
10	Identification and characterization of 15 novel GALC gene mutations causing Krabbe disease. <i>Human Mutation</i> , 2010, 31, E1894-E1914.	1.1	93
11	Characterization of the ERAD process of the L444P mutant glucocerebrosidase variant. <i>Blood Cells, Molecules, and Diseases</i> , 2011, 46, 4-10.	0.6	91
12	Unfolded protein response in Gaucher disease: from human to <i>Drosophila</i> . <i>Orphanet Journal of Rare Diseases</i> , 2013, 8, 140.	1.2	88
13	Functional Variants of the <i>HMGA1</i> Gene and Type 2 Diabetes Mellitus. <i>JAMA - Journal of the American Medical Association</i> , 2011, 305, 903.	3.8	87
14	Lysosomal lipase deficiency: Molecular characterization of eleven patients with Wolman or cholesteryl ester storage disease. <i>Molecular Genetics and Metabolism</i> , 2012, 105, 450-456.	0.5	71
15	The role of a bioresource research impact factor as an incentive to share human bioresources. <i>Nature Genetics</i> , 2011, 43, 503-504.	9.4	66
16	IDUA mutational profiling of a cohort of 102 European patients with mucopolysaccharidosis type I: identification and characterization of 35 novel α -L-iduronidase (IDUA) alleles. <i>Human Mutation</i> , 2011, 32, E2189-E2210.	1.1	66
17	SMPD1 Mutation Update: Database and Comprehensive Analysis of Published and Novel Variants. <i>Human Mutation</i> , 2016, 37, 139-147.	1.1	66
18	Molecular analysis and characterization of nine novel CTSK mutations in twelve patients affected by pycnodysostosis. <i>Human Mutation</i> , 2007, 28, 524-524.	1.1	64

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19	Molecular and functional analysis ofSUMF1 mutations in multiple sulfatase deficiency. Human Mutation, 2004, 23, 576-581.	1.1	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.	1.4	63
21	Clinical and molecular findings in patients with giant axonal neuropathy (GAN). Neurology, 2004, 62, 13-16.	1.5	62
22	Mutations in MFSD8/CLN7 are a frequent cause of variant-late infantile neuronal ceroid lipofuscinosis. Human Mutation, 2009, 30, E530-E540.	1.1	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.	1.0	56
24	Analysis of the glucocerebrosidase gene and mutation profile in 144 Italian gaucher patients. Human Mutation, 2002, 20, 234-235.	1.1	51
25	Acid sphingomyelinase: Identification of nine novel mutations among Italian Niemann Pick type B patients and characterization of in vivo functional in-frame start codon. Human Mutation, 2004, 24, 186-187.	1.1	51
26	Mutational analysis of theHGSNATgene in Italian patients with mucopolysaccharidosis IIIC (Sanfilippo) Tj ETQq0 0 0 rgBT /Overlock 10 T	1.1	51
27	Molecular and functional characterization of eight novel GAA mutations in Italian infants with Pompe disease. Human Mutation, 2008, 29, E27-E36.	1.1	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.	0.5	50
29	Pulmonary Manifestations of Gaucher Disease. American Journal of Respiratory and Critical Care Medicine, 1998, 157, 985-989.	2.5	48
30	Glucocerebrosidase deficiency in zebrafish affects primary bone ossification through increased oxidative stress and reduced Wnt/ β -catenin signaling. Human Molecular Genetics, 2015, 24, 1280-1294.	1.4	46
31	Functional analysis of 11 novel GBA alleles. European Journal of Human Genetics, 2014, 22, 511-516.	1.4	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.	1.1	43
33	Molecular epidemiology of childhood neuronal ceroid-lipofuscinosis in Italy. Orphanet Journal of Rare Diseases, 2013, 8, 19.	1.2	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.	0.3	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.	1.2	40
36	Molecular analysis of NPC1 and NPC2 gene in 34 Niemann-Pick C Italian Patients: identification and structural modeling of novel mutations. Neurogenetics, 2009, 10, 229-239.	0.7	39

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37	Telethon Network of Genetic Biobanks: a key service for diagnosis and research on rare diseases. <i>Orphanet Journal of Rare Diseases</i> , 2013, 8, 129.	1.2	39
38	Mucopolysaccharidosis IVA (Morquio A): Identification of novel common mutations in the N-acetylgalactosamine-6-sulfate sulfatase (GALNS) gene in Italian patients. <i>Human Mutation</i> , 2004, 24, 187-188.	1.1	38
39	Hepatic and neuromuscular forms of glycogenosis type III: nine mutations in AGL. <i>Human Mutation</i> , 2006, 27, 600-601.	1.1	38
40	Molecular characterization of 22 novel UDP-N-acetylglucosamine-1-phosphate transferase β - and β -subunit (<i>GNPTAB</i>) gene mutations causing mucopolipidosis types III β and III β in 46 patients. <i>Human Mutation</i> , 2009, 30, E956-E973.	1.1	38
41	Quantifying the use of bioresources for promoting their sharing in scientific research. <i>GigaScience</i> , 2013, 2, 7.	3.3	38
42	Characterization of iduronate-2-sulfatase gene-pseudogene recombinations in eight patients with Mucopolysaccharidosis type II revealed by a rapid PCR-based method. <i>Human Mutation</i> , 2005, 25, 491-497.	1.1	37
43	Molecular analysis of the <i>HEXA</i> gene in Italian patients with infantile and late Onset Tay-Sachs disease: detection of fourteen novel alleles. <i>Human Mutation</i> , 2005, 26, 282-282.	1.1	37
44	Type II sialidosis: review of the clinical spectrum and identification of a new splicing defect with chitotriosidase assessment in two patients. <i>Journal of Neurology</i> , 2009, 256, 1911-1915.	1.8	37
45	Human iPSC-based models highlight defective glial and neuronal differentiation from neural progenitor cells in metachromatic leukodystrophy. <i>Cell Death and Disease</i> , 2018, 9, 698.	2.7	37
46	The lysosomal storage disorders mucopolipidosis type II, type III alpha/beta, and type III gamma: Update on <i>GNPTAB</i> and <i>GNPTG</i> mutations. <i>Human Mutation</i> , 2019, 40, 842-864.	1.1	36
47	Molecular analysis of 30 mucopolysaccharidosis type I patients: evaluation of the mutational spectrum in Italian population and identification of 13 novel mutations. <i>Human Mutation</i> , 2002, 20, 231-231.	1.1	35
48	Functional in vitro characterization of 14 <i>SMPD1</i> mutations identified in Italian patients affected by Niemann Pick Type B disease. <i>Human Mutation</i> , 2005, 26, 164-164.	1.1	35
49	Cell surface associated glycohydrolases in normal and Gaucher disease fibroblasts. <i>Journal of Inherited Metabolic Disease</i> , 2012, 35, 1081-1091.	1.7	35
50	Mutations among Italian mucopolysaccharidosis type I patients. <i>Journal of Inherited Metabolic Disease</i> , 1997, 20, 803-806.	1.7	34
51	Different Molecular Mechanisms Leading to White Matter Hypomyelination in Infantile Onset Lysosomal Disorders. <i>Neuropediatrics</i> , 2005, 36, 265-269.	0.3	34
52	A multicentre observational study for early diagnosis of Gaucher disease in patients with Splenomegaly and/or Thrombocytopenia. <i>European Journal of Haematology</i> , 2016, 96, 352-359.	1.1	34
53	Perturbations in cell signaling elicit early cardiac defects in mucopolysaccharidosis type II. <i>Human Molecular Genetics</i> , 2017, 26, 1643-1655.	1.4	34
54	Biochemical and molecular analysis in mucopolysaccharidoses: what a paediatrician must know. <i>Italian Journal of Pediatrics</i> , 2018, 44, 129.	1.0	34

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55	Unbalanced GLA mRNAs ratio quantified by real-time PCR in Fabry patients' fibroblasts results in Fabry disease. <i>European Journal of Human Genetics</i> , 2008, 16, 1311-1317.	1.4	33
56	Mucopolysaccharidosis type IIID: 12 new patients and 15 novel mutations. <i>Human Mutation</i> , 2010, 31, n/a-n/a.	1.1	33
57	The RD-Connect Registry & Biobank Finder: a tool for sharing aggregated data and metadata among rare disease researchers. <i>European Journal of Human Genetics</i> , 2018, 26, 631-643.	1.4	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. <i>European Journal of Human Genetics</i> , 1999, 7, 125-130.	1.4	32
59	Basic Fibroblast Growth Factor Activates Endothelial Nitric-Oxide Synthase in CHO-K1 Cells via the Activation of Ceramide Synthesis. <i>Molecular Pharmacology</i> , 2003, 63, 297-310.	1.0	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. <i>Orphanet Journal of Rare Diseases</i> , 2011, 6, 40.	1.2	32
61	Biochemical and molecular findings in a patient with myoclonic epilepsy due to a mistarget of the Î²-glucosidase enzyme. <i>Molecular Genetics and Metabolism</i> , 2009, 97, 309-311.	0.5	31
62	SMAD4 mutations causing Myhre syndrome result in disorganization of extracellular matrix improved by losartan. <i>European Journal of Human Genetics</i> , 2014, 22, 988-994.	1.4	31
63	Molecular defects in the Î±-N-acetylglucosaminidase gene in Italian Sanfilippo type B patients. <i>Human Genetics</i> , 2000, 107, 568-576.	1.8	30
64	Contribution of arylsulphatase A mutations located on the same allele to enzyme activity reduction and metachromatic leukodystrophy severity. <i>Human Genetics</i> , 2002, 110, 351-355.	1.8	30
65	An Alu-mediated rearrangement as cause of exon skipping in Hunter disease. <i>Human Genetics</i> , 2003, 112, 419-425.	1.8	30
66	Leukoencephalopathy with vanishing white matter. <i>Neurology</i> , 2003, 61, 1818-1819.	1.5	30
67	Genetic disorders affecting white matter in the pediatric age. <i>American Journal of Medical Genetics Part A</i> , 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. <i>European Journal of Human Genetics</i> , 2013, 21, 34-39.	1.4	30
69	<i>GFAP </i>mutations and polymorphisms in 13 unrelated Italian patients affected by Alexander disease. <i>Clinical Genetics</i> , 2007, 72, 427-433.	1.0	29
70	Screening of 25 Italian patients with Niemann-Pick a reveals fourteen new mutations, one common and thirteen private, in SMPD1. <i>Human Mutation</i> , 2004, 24, 105-105.	1.1	28
71	Cerebellar atrophy without cerebellar cortex hyperintensity in infantile neuroaxonal dystrophy (INAD) due to PLA2G6 mutation. <i>European Journal of Paediatric Neurology</i> , 2007, 11, 175-177.	0.7	28
72	Molecular analysis of <i>ARSA</i> and <i>PSAP</i> genes in twenty-one Italian patients with metachromatic leukodystrophy: identification and functional characterization of 11 novel <i>ARSA</i> alleles. <i>Human Mutation</i> , 2008, 29, E220-E230.	1.1	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. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2006, 1762, 478-484.	1.8	27
74	GM1 gangliosidosis: molecular analysis of nine patients and development of an RT-PCR assay for GLB1 gene expression profiling. <i>Human Mutation</i> , 2007, 28, 204-204.	1.1	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. <i>Clinical Genetics</i> , 2011, 80, 452-458.	1.0	27
76	FGF signaling deregulation is associated with early developmental skeletal defects in animal models for mucopolysaccharidosis type II (MPSII). <i>Human Molecular Genetics</i> , 2018, 27, 2262-2275.	1.4	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. <i>Prenatal Diagnosis</i> , 1985, 5, 329-336.	1.1	26
78	Identification and molecular characterization of six novel mutations in the UDP-N-acetylglucosamine-1-phosphotransferase gamma subunit (GNPTG) gene in patients with mucopolisidosis III gamma. <i>Human Mutation</i> , 2009, 30, 978-984.	1.1	26
79	Optimizing the Molecular Diagnosis of GALNS: Novel Methods to Define and Characterize Morquio-A Syndrome-Associated Mutations. <i>Human Mutation</i> , 2015, 36, 357-368.	1.1	26
80	PLP1 gene duplication causes overexpression and alteration of the PLP/DM20 splicing balance in fibroblasts from Pelizaeus's Merzbacher disease patients. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2009, 1792, 548-554.	1.8	25
81	Functional and genetic characterization of the non-lysosomal glucosylceramidase 2 as a modifier for Gaucher disease. <i>Orphanet Journal of Rare Diseases</i> , 2013, 8, 151.	1.2	24
82	Severity of bone marrow involvement in patients with Gaucher's disease evaluated by scintigraphy with 99mTc-sestamibi. <i>Journal of Nuclear Medicine</i> , 2003, 44, 1253-62.	2.8	24
83	Identification and functional characterization of five novel mutant alleles in 58 Italian patients with Gaucher disease type 1. <i>Human Mutation</i> , 2005, 25, 100-100.	1.1	22
84	An Alu insertion in compound heterozygosity with a microduplication in GNPTAB gene underlies Mucopolisidosis II. <i>Molecular Genetics and Metabolism</i> , 2008, 93, 129-133.	0.5	22
85	Molecular and functional analysis of the HEXB gene in Italian patients affected with Sandhoff disease: identification of six novel alleles. <i>Neurogenetics</i> , 2009, 10, 49-58.	0.7	22
86	Sequence and Copy Number Analyses of HEXB Gene in Patients Affected by Sandhoff Disease: Functional Characterization of 9 Novel Sequence Variants. <i>PLoS ONE</i> , 2012, 7, e41516.	1.1	22
87	Multiple cryptic splice sites can be activated by IDS point mutations generating misspliced transcripts. <i>Journal of Molecular Medicine</i> , 2006, 84, 692-700.	1.7	21
88	Craniosynostosis: A rare complication of pycnodysostosis. <i>European Journal of Medical Genetics</i> , 2010, 53, 89-92.	0.7	21
89	Treatment of Human Fibroblasts Carrying NPC1 Missense Mutations with MG132 Leads to an Improvement of Intracellular Cholesterol Trafficking. <i>JIMD Reports</i> , 2011, 2, 59-69.	0.7	21
90	Gene expression profile in patients with Gaucher disease indicates activation of inflammatory processes. <i>Scientific Reports</i> , 2019, 9, 6060.	1.6	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. <i>European Journal of Human Genetics</i> , 2008, 16, 462-470.	1.4	20
92	Detection of carriers and prenatal diagnosis for fucosidosis in Calabria. <i>Human Genetics</i> , 1979, 51, 195-201.	1.8	19
93	Galactosemia caused by generalized uridine diphosphate galactose-4-epimerase deficiency. <i>Journal of Pediatrics</i> , 1983, 103, 927-930.	0.9	19
94	Prenatal diagnosis of Pelizaeus-Merzbacher disease: detection of proteolipid protein gene duplication by quantitative fluorescent multiplex PCR. <i>Prenatal Diagnosis</i> , 2001, 21, 668-671.	1.1	19
95	Genotype-phenotype correlation in five Pelizaeus-Merzbacher disease patients with <i>PLP1</i> gene duplications. <i>Clinical Genetics</i> , 2008, 73, 279-287.	1.0	19
96	Critical issues for the proper diagnosis of Metachromatic Leukodystrophy. <i>Gene</i> , 2014, 537, 348-351.	1.0	19
97	Evidence for a Founder Effect in Sicilian Patients with Glycogen Storage Disease Type II. <i>Human Heredity</i> , 2000, 50, 331-333.	0.4	18
98	Diagnosis of Pelizaeus-Merzbacher disease: detection of proteolipid protein gene copy number by real-time PCR. <i>Neurogenetics</i> , 2005, 6, 73-78.	0.7	18
99	Haplotype analysis suggests a single Balkan origin for the Gaucher disease [D409H;H255Q] double mutant allele. <i>Human Mutation</i> , 2008, 29, E58-E67.	1.1	18
100	ASAH1 variant causing a mild SMA phenotype with no myoclonic epilepsy: a clinical, biochemical and molecular study. <i>European Journal of Human Genetics</i> , 2016, 24, 1578-1583.	1.4	18
101	UPR activation and CHOP mediated induction of GBA1 transcription in Gaucher disease. <i>Blood Cells, Molecules, and Diseases</i> , 2018, 68, 21-29.	0.6	18
102	Enigmatic In Vivo iduronate-2-sulfatase (IDS) mutant transcript correction to wild-type in Hunter syndrome. <i>Human Mutation</i> , 2010, 31, E1261-E1285.	1.1	17
103	A Novel DHPLC-Based Procedure for the Analysis of COL1A1 and COL1A2 Mutations in Osteogenesis Imperfecta. <i>Journal of Molecular Diagnostics</i> , 2011, 13, 648-656.	1.2	17
104	Mucopolysaccharidosis type II in a female patient with a reciprocal X;9 translocation and skewed X chromosome inactivation. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 2627-2632.	0.7	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. <i>Human Genetics</i> , 1998, 102, 50-53.	1.8	16
106	The effect of four mutations on the expression of iduronate-2-sulfatase in mucopolysaccharidosis type II. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2001, 1537, 233-238.	1.8	16
107	ITCH regulates degradation of mutant glucocerebrosidase: implications to Gaucher disease. <i>Human Molecular Genetics</i> , 2013, 22, 1316-1327.	1.4	16
108	Validity of β -d-glucosidase activity measured in dried blood samples for detection of potential Gaucher disease patients. <i>Clinical Biochemistry</i> , 2014, 47, 1293-1296.	0.8	16

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109	Origin and spread of a common deletion causing mucopolipidosis type II: insights from patterns of haplotypic diversity. <i>Clinical Genetics</i> , 2011, 80, 273-280.	1.0	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 Niemann-Pick C disease. <i>Neurogenetics</i> , 2020, 21, 105-119.	0.7	15
111	Expression studies of two novel in CIS-mutations identified in an intermediate case of Hunter syndrome. <i>American Journal of Medical Genetics Part A</i> , 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. <i>Neurogenetics</i> , 2005, 6, 195-199.	0.7	14
113	Segregation analysis in a family at risk for the Maroteaux-Lamy syndrome conclusively reveals c.1151G>A (p.S384N) as to be a polymorphism. <i>European Journal of Human Genetics</i> , 2009, 17, 1160-1164.	1.4	14
114	A Novel Polymorphic AP-1 Binding Element of the GFAP Promoter is Associated with Different Allelic Transcriptional Activities. <i>Annals of Human Genetics</i> , 2010, 74, 506-515.	0.3	14
115	An Alu-mediated duplication in NMNAT1, involved in NAD biosynthesis, causes a novel syndrome, SHILCA, affecting multiple tissues and organs. <i>Human Molecular Genetics</i> , 2020, 29, 2250-2260.	1.4	14
116	A deletion involving exons 2-4 in the iduronate-2-sulfatase gene of a patient with intermediate Hunter syndrome. <i>Clinical Genetics</i> , 1998, 53, 474-477.	1.0	13
117	Carbon nanotubes as nanovectors for intracellular delivery of laronidase in Mucopolysaccharidosis type I. <i>Nanoscale</i> , 2018, 10, 657-665.	2.8	13
118	Prenatal diagnosis of mucopolysaccharidosis I: A special difficulty arising from an unusually low enzyme activity in mother's cells. <i>Prenatal Diagnosis</i> , 1985, 5, 149-154.	1.1	12
119	Deletion of exons 11-17 and novel mutations of the galactocerebrosidase gene in adult- and early-onset patients with Krabbe disease. <i>Journal of Neurology</i> , 2000, 247, 875-877.	1.8	12
120	Identification of a Novel Recombinant Allele in Three Unrelated Italian Gaucher Patients: Implications for Prognosis and Genetic Counseling. <i>Blood Cells, Molecules, and Diseases</i> , 2000, 26, 307-311.	0.6	12
121	Nitric Oxide Production Stimulated by the Basic Fibroblast Growth Factor Requires the Synthesis of Ceramide. <i>Annals of the New York Academy of Sciences</i> , 2002, 973, 94-104.	1.8	12
122	An unusual arylsulfatase A pseudodeficiency allele carrying a splice site mutation in a metachromatic leukodystrophy patient. <i>European Journal of Human Genetics</i> , 2004, 12, 150-154.	1.4	12
123	Restoration of the Normal Splicing Pattern of the PLP1 Gene by Means of an Antisense Oligonucleotide Directed against an Exonic Mutation. <i>PLoS ONE</i> , 2013, 8, e73633.	1.1	12
124	A new case report of severe mucopolysaccharidosis type VII: diagnosis, treatment with haematopoietic cell transplantation and prenatal diagnosis in a second pregnancy. <i>Italian Journal of Pediatrics</i> , 2018, 44, 128.	1.0	12
125	Predicting the probability of Gaucher disease in subjects with splenomegaly and thrombocytopenia. <i>Scientific Reports</i> , 2021, 11, 2594.	1.6	12
126	An AT-deletion causing a frameshift in the arylsulfatase A gene of a late infantile metachromatic leukodystrophy patient. <i>Human Genetics</i> , 1995, 96, 233-235.	1.8	11

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127	Somatic Mosaicism in a Patient with Gaucher Disease Type 2: Implication for Genetic Counseling and Therapeutic Decision-Making. <i>Blood Cells, Molecules, and Diseases</i> , 2000, 26, 611-612.	0.6	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). <i>Molecular Genetics and Metabolism</i> , 2007, 90, 77-80.	0.5	11
129	Functional characterization of four novel MAN2B1 mutations causing juvenile onset alpha-mannosidosis. <i>Clinica Chimica Acta</i> , 2007, 375, 136-139.	0.5	11
130	A rare G6490 A>T substitution at the last nucleotide of exon 10 of the glucocerebrosidase gene in two unrelated Italian Gaucher patients. <i>Clinical Genetics</i> , 1995, 48, 123-127.	1.0	11
131	AIMP1/p43 Mutation and PMLD. <i>American Journal of Human Genetics</i> , 2011, 88, 391.	2.6	11
132	Sjögren-Larsson syndrome: Nuclear magnetic resonance imaging of the brain in a 4-year-old boy. <i>Journal of Inherited Metabolic Disease</i> , 1994, 17, 112-114.	1.7	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). <i>American Journal of Medical Genetics Part A</i> , 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. <i>American Journal of Medical Genetics, Part A</i> , 2005, 134A, 95-96.	0.7	10
135	Movement and mood disorder in two brothers with Gaucher disease. <i>Clinical Genetics</i> , 2007, 72, 357-361.	1.0	10
136	Functional Characterization of the Novel Mutation IVS 8 (A~11delC) (A~14T>A) in the Intron 8 of the Glucocerebrosidase Gene of Two Italian Siblings with Gaucher Disease Type I. <i>Blood Cells, Molecules, and Diseases</i> , 2000, 26, 171-176.	0.6	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. <i>Prenatal Diagnosis</i> , 2006, 26, 77-80.	1.1	8
139	A novel homozygous splicing mutation in PSAP gene causes metachromatic leukodystrophy in two Moroccan brothers. <i>Neurogenetics</i> , 2014, 15, 101-106.	0.7	8
140	A novel homozygous MCOLN1 double mutant allele leading to TRP channel domain ablation underlies Mucopolipidosis IV in an Italian Child. <i>Metabolic Brain Disease</i> , 2015, 30, 681-686.	1.4	8
141	Unusual white matter involvement in EAST syndrome associated with novel KCNJ10 mutations. <i>Journal of Neurology</i> , 2018, 265, 1419-1425.	1.8	8
142	Further genotype-phenotype correlation emerging from two families with PLP1 exon 4 skipping. <i>Clinical Genetics</i> , 2014, 85, 267-272.	1.0	7
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147	MLPA-based approach for initial and simultaneous detection of GBA deletions and recombinant alleles in patients affected by Gaucher Disease. <i>Molecular Genetics and Metabolism</i> , 2016, 119, 329-337.	0.5	5
148	Neonatal chitotriosidase activity is not predictive for Niemann-Pick disease type A/B: Implications for newborn screening for lysosomal storage disorders. <i>Molecular Genetics and Metabolism</i> , 2013, 108, 106.	0.5	4
149	^{99m}Tc -Sestamibi Scintigraphy to Monitor the Long-Term Efficacy of Enzyme Replacement Therapy on Bone Marrow Infiltration in Patients with Gaucher Disease. <i>Journal of Nuclear Medicine</i> , 2013, 54, 1717-1724.	2.8	4
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151	Somatic intragenic recombination of the arylsulfatase A gene in a metachromatic leukodystrophy patient. <i>Molecular Genetics and Metabolism</i> , 2006, 89, 150-155.	0.5	3
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158	A Multicenter Observational Study For Early Diagnosis Of Gaucher Disease In Patients With Splenomegaly and/Or Thrombocytopenia. <i>Blood</i> , 2013, 122, 4712-4712.	0.6	1
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