Rossella Parini

List of Publications by Year
in descending order

121 papers	5,355 citations	87888 38 h-index	91884 69 g-index
136 all docs	136 docs citations	136 times ranked	6480 citing authors

#	Article	IF	Citations
1	MPV17 encodes an inner mitochondrial membrane protein and is mutated in infantile hepatic mitochondrial DNA depletion. Nature Genetics, 2006, 38, 570-575.	21.4	380
2	Infantile hepatocerebral syndromes associated with mutations in the mitochondrial DNA polymerase-ÂA. Brain, 2005, 128, 723-731.	7.6	284
3	Long-term outcome of Hurler syndrome patients after hematopoietic cell transplantation: an international multicenter study. Blood, 2015, 125, 2164-2172.	1.4	262
4	Recommendations for initiation and cessation of enzyme replacement therapy in patients with Fabry disease: the European Fabry Working Group consensus document. Orphanet Journal of Rare Diseases, 2015, 10, 36.	2.7	239
5	Enzyme replacement therapy and/or hematopoietic stem cell transplantation at diagnosis in patients with mucopolysaccharidosis type I: results of a European consensus procedure. Orphanet Journal of Rare Diseases, 2011, 6, 55.	2.7	194
6	The early clinical phenotype of Fabry disease: a study on 35 European children and adolescents. European Journal of Pediatrics, 2003, 162, 767-772.	2.7	176
7	Mutations of the Mitochondrial-tRNA Modifier MTO1 Cause Hypertrophic Cardiomyopathy and Lactic Acidosis. American Journal of Human Genetics, 2012, 90, 1079-1087.	6.2	164
8	Mutations in FBXL4, Encoding a Mitochondrial Protein, Cause Early-Onset Mitochondrial Encephalomyopathy. American Journal of Human Genetics, 2013, 93, 482-495.	6.2	138
9	Enzyme replacement therapy: efficacy and limitations. Italian Journal of Pediatrics, 2018, 44, 120.	2.6	128
10	Deficiency in SLC25A1, Encoding the Mitochondrial Citrate Carrier, Causes Combined D-2- and L-2-Hydroxyglutaric Aciduria. American Journal of Human Genetics, 2013, 92, 627-631.	6.2	122
11	The Morquio A Clinical Assessment Program: Baseline results illustrating progressive, multisystemic clinical impairments in Morquio A subjects. Molecular Genetics and Metabolism, 2013, 109, 54-61.	1.1	117
12	Homozygous MTTP and APOB mutations may lead to hepatic steatosis and fibrosis despite metabolic differences in congenital hypocholesterolemia. Journal of Hepatology, 2014, 61, 891-902.	3.7	116
13	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
14	Seizure and EEG Patterns in Angelman's Syndrome. Journal of Child Neurology, 1995, 10, 467-471.	1.4	105
15	Effects of enzyme replacement therapy in Fabry diseaseâ€"A comprehensive review of the medical literature. Genetics in Medicine, 2010, 12, 668-679.	2.4	100
16	The natural course and the impact of therapies of cardiac involvement in the mucopolysaccharidoses. Cardiology in the Young, 2009, 19, 170-178.	0.8	99
17	Longâ€term observational, nonâ€randomized study of enzyme replacement therapy in lateâ€onset glycogenosis type II. Journal of Inherited Metabolic Disease, 2010, 33, 727-735.	3.6	79
18	Brain and spine MRI features of Hunter disease: frequency, natural evolution and response to therapy. Journal of Inherited Metabolic Disease, 2011, 34, 763-780.	3.6	78

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19	Genotype/phenotype correlation in glycogen storage disease type 1b: a multicentre study and review of the literature. European Journal of Pediatrics, 2005, 164, 501-508.	2.7	72
20	Intravenous Enzyme Replacement Therapy in Mucopolysaccharidoses: Clinical Effectiveness and Limitations. International Journal of Molecular Sciences, 2020, 21, 2975.	4.1	70
21	Open issues in Mucopolysaccharidosis type I-Hurler. Orphanet Journal of Rare Diseases, 2017, 12, 112.	2.7	67
22	Structure of the SLC7A7 Gene and Mutational Analysis of Patients Affected by Lysinuric Protein Intolerance. American Journal of Human Genetics, 2000, 66, 92-99.	6.2	66
23	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
24	Multi-domain impact of elosulfase alfa in Morquio A syndrome in the pivotal phase III trial. Molecular Genetics and Metabolism, 2015, 114, 178-185.	1.1	65
25	Long term clinical history of an Italian cohort of infantile onset Pompe disease treated with enzyme replacement therapy. Orphanet Journal of Rare Diseases, 2018, 13, 32.	2.7	65
26	Molecular and functional analysis ofSLC25A20 mutations causing carnitine-acylcarnitine translocase deficiency. Human Mutation, 2004, 24, 312-320.	2.5	63
27	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
28	Enzymatic replacement therapy for Hunter disease: Up to 9years experience with 17 patients. Molecular Genetics and Metabolism Reports, 2015, 3, 65-74.	1.1	63
29	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
30	Course and management of allogeneic stem cell transplantation in patients with mitochondrial neurogastrointestinal encephalomyopathy. Journal of Neurology, 2012, 259, 2699-2706.	3.6	52
31	Clinical pattern, mutations and in vitro residual activity in 33 patients with severe 5, 10 methylenetetrahydrofolate reductase (MTHFR) deficiency. Journal of Inherited Metabolic Disease, 2016, 39, 115-124.	3.6	52
32	Mucopolysaccharidosis VI: the Italian experience. European Journal of Pediatrics, 2009, 168, 1203-1206.	2.7	49
33	Long-term endurance and safety of elosulfase alfa enzyme replacement therapy in patients with Morquio A syndrome. Molecular Genetics and Metabolism, 2016, 119, 131-143.	1.1	47
34	Mutations in the glucose-6-phosphate transporter (G6PT) gene in patients with glycogen storage diseases type 1b and 1c. FEBS Letters, 1999, 459, 255-258.	2.8	44
35	Glucose metabolism and diet-based prevention of liver dysfunction in MPV17 mutant patients. Journal of Hepatology, 2009, 50, 215-221.	3.7	44
36	Clinical efficacy of Enzyme Replacement Therapy in paediatric Hunter patients, an independent study of 3.5 years. Orphanet Journal of Rare Diseases, 2014, 9, 129.	2.7	44

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37	Functional analysis of 11 novel GBA alleles. European Journal of Human Genetics, 2014, 22, 511-516.	2.8	44
38	Role of ?-galactosidase and elastin binding protein in lysosomal and nonlysosomal complexes of patients with GM1-gangliosidosis. Human Mutation, 2005, 25, 285-292.	2.5	43
39	Comprehensive Evaluation of Plasma 7-Ketocholesterol and Cholestan-3Î²,5Î±,6Î²-Triol in an Italian Cohort of Patients Affected by Niemann-Pick Disease due to NPC1 and SMPD1 Mutations. Clinica Chimica Acta, 2016, 455, 39-45.	1.1	42
40	Improvement of Cardiomyopathy After High-Fat Diet in Two Siblings with Glycogen Storage Disease Type III. JIMD Reports, 2014, 17, 91-95.	1.5	36
41	Acid Ceramidase Deficiency is characterized by a unique plasma cytokine and ceramide profile that is altered by therapy. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2017, 1863, 386-394.	3.8	35
42	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
43	Therapeutic goals in the treatment of Fabry disease. Genetics in Medicine, 2010, 12, 713-720.	2.4	33
44	Measuring patient experiences in Fabry disease: validation of the Fabry-specific Pediatric Health and Pain Questionnaire (FPHPQ). Health and Quality of Life Outcomes, 2012, 10, 116.	2.4	33
45	Longitudinal analysis of endurance and respiratory function from a natural history study of Morquio A syndrome. Molecular Genetics and Metabolism, 2015, 114, 186-194.	1.1	33
46	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
47	Diseases of ganglioside biosynthesis: An expanding group of congenital disorders of glycosylation. Molecular Genetics and Metabolism, 2018, 124, 230-237.	1.1	33
48	Ocular manifestations in the mucopolysaccharidoses â€“ a review. Clinical and Experimental Ophthalmology, 2010, 38, 12-22.	2.6	32
49	Clinical pharmacology of netilmicin in preterm and term newborn infants. Journal of Pediatrics, 1985, 106, 664-669.	1.8	31
50	Growth Charts for Individuals with Mucopolysaccharidosis VI (Maroteauxâ€“Lamy Syndrome). JIMD Reports, 2014, 18, 1-11.	1.5	31
51	Safety and clinical activity of elosulfase alfa in pediatric patients with Morquio A syndrome (mucopolysaccharidosis IVA) less than 5 y. Pediatric Research, 2015, 78, 717-722.	2.3	30
52	First manifestation of citrullinemia type I as differential diagnosis to postpartum psychosis in the puerperal period. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2010, 149, 228-229.	1.1	28
53	Sudden unexpected fatal encephalopathy in adults with OTC gene mutations-Clues for early diagnosis and timely treatment. Orphanet Journal of Rare Diseases, 2014, 9, 105.	2.7	28
54	Hereditary fructose intolerance: functional study of two novel ALDOB natural variants and characterization of a partial gene deletion. Human Mutation, 2010, 31, 1294-1303.	2.5	27

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55	Effect of alglucosidase alfa dosage on survival and walking ability in patients with classic infantile Pompe disease: a multicentre observational cohort study from the European Pompe Consortium. The Lancet Child and Adolescent Health, 2022, 6, 28-37.	5.6	27
56	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
57	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
58	Fabry disease: beyond men. Acta Paediatrica, International Journal of Paediatrics, 2008, 97, 31-32.	1.5	25
59	Impact of long-term elosulfase alfa on activities of daily living in patients with Morquio A syndrome in an open-label, multi-center, phase 3 extension study. Molecular Genetics and Metabolism, 2018, 123, 127-134.	1.1	25
60	Analysis of Renal and Cardiac Outcomes in Male Participants in the Fabry Outcome Survey Starting Agalsidase Alfa Enzyme Replacement Therapy Before and After 18 Years of Age . Drug Design, Development and Therapy, 2020, Volume 14, 2149-2158.	4.3	25
61	Impact of longâ€term elosulfase alfa treatment on respiratory function in patients with Morquio A syndrome. Journal of Inherited Metabolic Disease, 2016, 39, 839-847.	3.6	24
62	Ototoxicity of aminoglycoside antibiotics in infants and children. Pediatric Infectious Disease Journal, 1982, 1, 357-365.	2.0	23
63	Combined liver-kidney transplantation in glycogen storage disease Ia: A case beyond the guidelines. Liver Transplantation, 2007, 13, 762-764.	2.4	23
64	Common and Novel TMEM70 Mutations in a Cohort of Italian Patients with Mitochondrial Encephalocardiomyopathy. JIMD Reports, 2014, 15, 71-8.	1.5	23
65	Total loss of GM3 synthase activity by a normally processed enzyme in a novel variant and in all ST3GAL5 variants reported to cause a distinct congenital disorder of glycosylation. Glycobiology, 2019, 29, 229-241.	2.5	23
66	Dietary lipids in glycogen storage disease type III: A systematic literature study, case studies, and future recommendations. Journal of Inherited Metabolic Disease, 2020, 43, 770-777.	3.6	23
67	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
68	Congenital hyperinsulinism: Clinical and molecular analysis of a large Italian cohort. Gene, 2013, 521, 160-165.	2.2	21
69	Muscle MRI of classic infantile pompe patients: Fatty substitution and edemaâ€like changes. Muscle and Nerve, 2017, 55, 841-848.	2.2	21
70	Prevalence of Antiâ€“Adeno-Associated Virus Serotype 8 Neutralizing Antibodies and Arylsulfatase B Cross-Reactive Immunologic Material in Mucopolysaccharidosis VI Patient Candidates for a Gene Therapy Trial. Human Gene Therapy, 2015, 26, 145-152.	2.7	19
71	Quality of life of Hurler syndrome patients after successful hematopoietic stem cell transplantation. Blood Advances, 2017, 1, 2236-2242.	5.2	19
72	Molecular markers for the follow-up of enzyme-replacement therapy in mucopolysaccharidosis typeÂVI disease. Biotechnology and Applied Biochemistry, 2008, 49, 219.	3.1	18

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73	Paediatric Fabry disease: prognostic significance of ocular changes for disease severity. BMC Ophthalmology, 2016, 16, 202.	1.4	18
74	Chronic liver involvement in urea cycle disorders. Journal of Inherited Metabolic Disease, 2019, 42, 1118-1127.	3.6	17
75	Clinical diagnosis of long-chain acyl-coenzyme A-dehydrogenase deficiency: Use of stress and fat-loading tests. Journal of Pediatrics, 1991, 119, 77-80.	1.8	16
76	Delphi consensus on the current clinical and therapeutic knowledge on Andersonâ€“Fabry disease. European Journal of Internal Medicine, 2014, 25, 751-756.	2.2	16
77	Determination of argininosuccinate lyase and arginase activities with an amino acid analyzer. Analytical Biochemistry, 1990, 191, 384-389.	2.4	15
78	Progression of Renal Damage in Glycogen Storage Disease Type I Is Associated to Hyperlipidemia: A Multicenter Prospective Italian Study. Journal of Pediatrics, 2015, 166, 1079-1082.	1.8	15
79	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
80	Reconstitution of lymphocyte subpopulations in children with inherited metabolic storage diseases after haematopoietic cell transplantation. British Journal of Haematology, 2005, 130, 249-255.	2.5	14
81	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
82	Mis-splicing of the GALNS gene resulting from deep intronic mutations as a cause of Morquio a disease. BMC Medical Genetics, 2018, 19, 183.	2.1	14
83	Vitamin E Improves Clinical Outcome of Patients Affected by Glycogen Storage Disease Type Ib. JIMD Reports, 2015, 25, 39-45.	1.5	13
84	Identification and characterization of five novel MAN2B1 mutations in Italian patients with alpha-mannosidosis. Human Mutation, 2005, 25, 320-320.	2.5	12
85	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
86	Integration of PCR-Sequencing Analysis with Multiplex Ligation-Dependent Probe Amplification for Diagnosis of Hereditary Fructose Intolerance. JIMD Reports, 2012, 6, 31-37.	1.5	11
87	Safety of anesthesia for children with mucopolysaccharidoses: A retrospective analysis of 54 patients. Paediatric Anaesthesia, 2018, 28, 436-442.	1.1	11
88	Easyâ€toâ€use algorithm would provide faster diagnoses for mucopolysaccharidosis type I and enable patients to receive earlier treatment. Acta Paediatrica, International Journal of Paediatrics, 2018, 107, 1402-1408.	1.5	11
89	Treatment of thoracolumbar kyphosis in patients with mucopolysaccharidosis type I: results of an international consensus procedure. Orphanet Journal of Rare Diseases, 2019, 14, 17.	2.7	11
90	Pre-diagnosing and managing patients with GM1 gangliosidosis and related disorders by the evaluation of GM1 ganglioside content. Scientific Reports, 2019, 9, 17684.	3.3	11

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91	Severe Neonatal Metabolic Decompensation in Methylmalonic Acidemia Caused by CblD Defect. JIMD Reports, 2013, 11, 133-137.	1.5	10
92	Vaccination coverage of patients with inborn errors of metabolism and the attitudes of their parents towards vaccines. Vaccine, 2015, 33, 6520-6524.	3.8	10
93	International working group identifies need for newborn screening for mucopolysaccharidosis type I but states that existing hurdles must be overcome. Acta Paediatrica, International Journal of Paediatrics, 2018, 107, 2059-2065.	1.5	10
94	Enzyme replacement therapy outcomes across the disease spectrum: Findings from the mucopolysaccharidosis VI Clinical Surveillance Program. Journal of Inherited Metabolic Disease, 2019, 42, 519-526.	3.6	10
95	Modelling the resource implications of managing adults with Fabry disease in Italy. European Journal of Clinical Investigation, 2011, 41, 710-718.	3.4	9
96	Recommendations on Reintroduction of Agalsidase Beta for Patients with Fabry Disease in Europe, Following a Period of Shortage. JIMD Reports, 2012, 8, 51-56.	1.5	9
97	Pitfalls in the detection of gross gene rearrangements using MLPA in Fabry disease. Clinica Chimica Acta, 2016, 452, 82-86.	1.1	9
98	Clinical and radiological correlates of activities of daily living in cerebellar atrophy caused by PMM2 mutations (PMM2-CDG). Cerebellum, 2021, 20, 596-605.	2.5	8
99	Enzyme replacement therapy initiated in adulthood: Findings from the mucopolysaccharidosis VI Clinical Surveillance Program. Molecular Genetics and Metabolism, 2019, 127, 355-360.	1.1	7
100	The new frame for Mucopolysaccharidoses. Italian Journal of Pediatrics, 2018, 44, 117.	2.6	6
101	Metabolic screening for the newborn. Journal of Maternal-Fetal and Neonatal Medicine, 2011, 24, 6-8.	1.5	5
102	Molecular basis, diagnosis and clinical management of mucopolysaccharidoses. Neurology International, 2013, 3, .	0.5	5
103	Mucopolysaccharidosis type VI enzyme replacement therapy initiated in adulthood: Findings from the MPS VI clinical surveillance program. Molecular Genetics and Metabolism, 2018, 123, S59.	1.1	5
104	Resting energy expenditure in argininosuccinic aciduria and in other urea cycle disorders. Journal of Inherited Metabolic Disease, 2019, 42, 1105-1117.	3.6	5
105	High-Frequency Rhythmic Cortical Myoclonus in a Long-Surviving Patient With Nonketotic Hypergylcemia. Journal of Child Neurology, 2008, 23, 321-324.	1.4	4
106	Females and children with Andersonâ€“Fabry disease: diagnosis, monitoring, benefits of enzyme replacement therapy (ERT) and considerations on timing of starting ERT. Expert Opinion on Orphan Drugs, 2013, 1, 315-330.	0.8	4
107	Characterization of Hearing Loss in Children with Mucopolysaccharidosis. , 0, , .		4
108	â€œGrowth patterns in children with mucopolysaccharidosis type I-Hurler after hematopoietic stem cell transplantation: Comparison with untreated patientsâ€• Molecular Genetics and Metabolism Reports, 2021, 28, 100787.	1.1	3

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109	Proposal of an Algorithm to Early Detect Attenuated Type I Mucopolysaccharidosis (MPS Ia) among Children with Growth Abnormalities. Medicina (Lithuania), 2022, 58, 97.	2.0	3
110	Sudden infant death and multiple acyl-CoA dehydrogenation disorders. European Journal of Pediatrics, 1995, 154, 421-422.	2.7	2
111	Vagal Nerve Stimulation in the Treatment of Drug-Resistant Epileptic Encephalopathies in Inborn Errors of Metabolism. Child Neurology Open, 2015, 2, 2329048X1561243.	1.1	2
112	Familiar unbalanced complex rearrangements involving 13 p-arm: description of two cases. Molecular Cytogenetics, 2018, 11, 52.	0.9	1
113	Thoracolumbar kyphosis in MPS I: A natural history study and an international consensus procedure for the development of a clinical practice guideline. Molecular Genetics and Metabolism, 2019, 126, S88-S89.	1.1	1
114	Evidence of treatment benefits in patients with MPSI-Hurler in long-term follow up using a new MRI scoring system. Journal of Pediatrics, 2021, , .	1.8	1
115	Craniovertebral Junction Pathological Features and Their Management in the Mucopolysaccharidoses. Advances and Technical Standards in Neurosurgery, 2014, 40, 313-331.	0.5	1
116	Predictors of Long-Term Clinical Outcome in Hurler Syndrome Patients after Successful Hematopoietic Cell Transplantation: An International Study. Biology of Blood and Marrow Transplantation, 2014, 20, S78-S79.	2.0	0
117	372. Prevalence of Anti-AAV8 Neutralizing Antibodies and ARSB Cross-Reactive Immunologic Material in MPS VI Patients Candidates for a Gene Therapy Trial. Molecular Therapy, 2015, 23, S148.	8.2	0
118	Health-Related Quality of Life and Perception of Care of Mucopolysaccharidosis Type I - Hurler Syndrome Patients after Successful Hematopoietic Cell Transplantation: A Parents' Perspective. Biology of Blood and Marrow Transplantation, 2015, 21, S207-S208.	2.0	0
119	Quality of Life of Hurler Syndrome Patients after Successful Hematopoietic Cell Transplantation. Biology of Blood and Marrow Transplantation, 2018, 24, S29-S30.	2.0	0
120	Facing up to limits: a lesson from the Charlie Gard case. Minerva Anestesiologica, 2018, 84, 261-262.	1.0	0
121	Enzyme replacement therapy in patients with mucopolysaccharidosis type VI: Updated findings from the MPS VI clinical surveillance program. Molecular Genetics and Metabolism, 2019, 126, S68.	1.1	0