Nicola Brunetti-Pierri

List of Publications by Year
in descending order

218 papers	16,655 citations	50170 46 h-index	17055 122 g-index
228 all docs	228 docs citations	228 times ranked	31589 citing authors

#	Article	IF	Citations
1	Guidelines for the use and interpretation of assays for monitoring autophagy (3rd edition). Autophagy, 2016, 12, 1-222.	4.3	4,701
2	Guidelines for the use and interpretation of assays for monitoring autophagy. Autophagy, 2012, 8, 445-544.	4.3	3,122
3	Discovery of drug mode of action and drug repositioning from transcriptional responses. Proceedings of the National Academy of Sciences of the United States of America, 2010, 107, 14621-14626.	3.3	813
4	Recurrent reciprocal 1q21.1 deletions and duplications associated with microcephaly or macrocephaly and developmental and behavioral abnormalities. Nature Genetics, 2008, 40, 1466-1471.	9.4	535
5	Defective CFTR induces aggresome formation and lung inflammation in cystic fibrosis through ROS-mediated autophagy inhibition. Nature Cell Biology, 2010, 12, 863-875.	4.6	420
6	GM1 gangliosidosis: Review of clinical, molecular, and therapeutic aspects. Molecular Genetics and Metabolism, 2008, 94, 391-396.	0.5	359
7	Molecular and clinical genetics of mitochondrial diseases due toPOLGmutations. Human Mutation, 2008, 29, E150-E172.	1.1	256
8	Acute Toxicity After High-Dose Systemic Injection of Helper-Dependent Adenoviral Vectors into Nonhuman Primates. Human Gene Therapy, 2004, 15, 35-46.	1.4	240
9	RASA1Mutations and Associated Phenotypes in 68 Families with Capillary Malformation-Arteriovenous Malformation. Human Mutation, 2013, 34, 1632-1641.	1.1	221
10	Wilson Disease Protein ATP7B Utilizes Lysosomal Exocytosis to Maintain Copper Homeostasis. Developmental Cell, 2014, 29, 686-700.	3.1	203
11	Speech delay and autism spectrum behaviors are frequently associated with duplication of the 7q11.23 Williams-Beuren syndrome region. Genetics in Medicine, 2007, 9, 427-441.	1.1	193
12	Copy number variants at Williamsâ€“Beuren syndrome 7q11.23 region. Human Genetics, 2010, 128, 3-26.	1.8	134
13	Gene transfer of master autophagy regulator TFEB results in clearance of toxic protein and correction of hepatic disease in alphaâ€1â€antiâ€trypsin deficiency. EMBO Molecular Medicine, 2013, 5, 397-412.	3.3	134
14	Toll-like Receptor 9 Triggers an Innate Immune Response to Helper-dependent Adenoviral Vectors. Molecular Therapy, 2007, 15, 378-385.	3.7	130
15	Lathosterolosis, a Novel Multiple-Malformation/Mental Retardation Syndrome Due to Deficiency of 3Î²-Hydroxysteroid-Î”5-Desaturase. American Journal of Human Genetics, 2002, 71, 952-958.	2.6	118
16	Heterogeneous clinical presentation in ICF syndrome: correlation with underlying gene defects. European Journal of Human Genetics, 2013, 21, 1219-1225.	1.4	115
17	Inborn errors of metabolism: the flux from Mendelian to complex diseases. Nature Reviews Genetics, 2006, 7, 449-459.	7.7	113
18	DUF1220-Domain Copy Number Implicated in Human Brain-Size Pathology and Evolution. American Journal of Human Genetics, 2012, 91, 444-454.	2.6	113

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19	Mutations in theMPV17 gene are responsible for rapidly progressive liver failure in infancy. Hepatology, 2007, 46, 1218-1227.	3.6	111
20	Generalized metabolic bone disease in Neurofibromatosis type I. Molecular Genetics and Metabolism, 2008, 94, 105-111.	0.5	105
21	Large-scale targeted sequencing identifies risk genes for neurodevelopmental disorders. Nature Communications, 2020, 11, 4932.	5.8	105
22	Duplications of FOXG1 in 14q12 are associated with developmental epilepsy, mental retardation, and severe speech impairment. European Journal of Human Genetics, 2011, 19, 102-107.	1.4	104
23	Transcriptional gene network inference from a massive dataset elucidates transcriptome organization and gene function. Nucleic Acids Research, 2011, 39, 8677-8688.	6.5	102
24	Efficient, Long-term Hepatic Gene Transfer Using Clinically Relevant HDAd Doses by Balloon Occlusion Catheter Delivery in Nonhuman Primates. Molecular Therapy, 2009, 17, 327-333.	3.7	88
25	Supravalvular Aortic Stenosis. Circulation: Cardiovascular Genetics, 2012, 5, 692-696.	5.1	87
26	Pseudo-hydrodynamic Delivery of Helper-dependent Adenoviral Vectors into Non-human Primates for Liver-directed Gene Therapy. Molecular Therapy, 2007, 15, 732-740.	3.7	81
27	Progress and prospects: gene therapy for genetic diseases with helper-dependent adenoviral vectors. Gene Therapy, 2008, 15, 553-560.	2.3	78
28	Transgene Expression up to 7 Years in Nonhuman Primates Following Hepatic Transduction with Helper-Dependent Adenoviral Vectors. Human Gene Therapy, 2013, 24, 761-765.	1.4	78
29	Phenylbutyrate therapy for maple syrup urine disease. Human Molecular Genetics, 2011, 20, 631-640.	1.4	77
30	Clinical Consequences of Urea Cycle Enzyme Deficiencies and Potential Links to Arginine and Nitric Oxide Metabolism. Journal of Nutrition, 2004, 134, 2775S-2782S.	1.3	76
31	Cystic fibrosis: A disorder with defective autophagy. Autophagy, 2011, 7, 104-106.	4.3	75
32	Sustained Phenotypic Correction of Canine Hemophilia B After Systemic Administration of Helper-Dependent Adenoviral Vector. Human Gene Therapy, 2005, 16, 811-820.	1.4	74
33	Improved Hepatic Transduction, Reduced Systemic Vector Dissemination, and Long-Term Transgene Expression by Delivering Helper-Dependent Adenoviral Vectors into the Surgically Isolated Liver of Nonhuman Primates. Human Gene Therapy, 2006, 17, 391-404.	1.4	74
34	Nitric-Oxide Supplementation for Treatment of Long-Term Complications in Argininosuccinic Aciduria. American Journal of Human Genetics, 2012, 90, 836-846.	2.6	73
35	Increased Hepatic Transduction with Reduced Systemic Dissemination and Proinflammatory Cytokines Following Hydrodynamic Injection of Helper-Dependent Adenoviral Vectors. Molecular Therapy, 2005, 12, 99-106.	3.7	72
36	Sensory ataxic neuropathy with ophthalmoparesis caused by POLG mutations. Neuromuscular Disorders, 2008, 18, 626-632.	0.3	71

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37	Helper-dependent adenoviral vectors for liver-directed gene therapy. Human Molecular Genetics, 2011, 20, R7-R13.	1.4	71
38	Pyruvate dehydrogenase complex and lactate dehydrogenase are targets for therapy of acute liver failure. Journal of Hepatology, 2018, 69, 325-335.	1.8	65
39	gene2drug: a computational tool for pathway-based rational drug repositioning. Bioinformatics, 2018, 34, 1498-1505.	1.8	62
40	Phenylbutyrate Therapy for Pyruvate Dehydrogenase Complex Deficiency and Lactic Acidosis. Science Translational Medicine, 2013, 5, 175ra31.	5.8	59
41	Identification of small molecules enhancing autophagic function from drug network analysis. Autophagy, 2010, 6, 1204-1205.	4.3	58
42	Nutrientâ€sensitive transcription factors <scp>TFEB</scp> and <scp>TFE</scp> 3 couple autophagy and metabolism to the peripheral clock. EMBO Journal, 2019, 38, .	3.5	58
43	Novel types of COMP mutations and genotype-phenotype association in pseudoachondroplasia and multiple epiphyseal dysplasia. Human Genetics, 2003, 112, 84-90.	1.8	56
44	Paralog Studies Augment Gene Discovery: DDX and DHX Genes. American Journal of Human Genetics, 2019, 105, 302-316.	2.6	56
45	Self-Healing Collodion Membrane and Mild Nonbullous Congenital Ichthyosiform Erythroderma Due to 2 Novel Mutations in the ALOX12B Gene. Archives of Dermatology, 2008, 144, 351-6.	1.7	54
46	SR-A and SREC-I Are Kupffer and Endothelial Cell Receptors for Helper-dependent Adenoviral Vectors. Molecular Therapy, 2013, 21, 767-774.	3.7	51
47	Induction of Nitric-Oxide Metabolism in Enterocytes Alleviates Colitis and Inflammation-Associated Colon Cancer. Cell Reports, 2018, 23, 1962-1976.	2.9	51
48	Disease characteristics of MCT8 deficiency: an international, retrospective, multicentre cohort study. Lancet Diabetes and Endocrinology,the, 2020, 8, 594-605.	5.5	50
49	Loss of SMPD4 Causes a Developmental Disorder Characterized by Microcephaly and Congenital Arthrogryposis. American Journal of Human Genetics, 2019, 105, 689-705.	2.6	48
50	Retinal transduction profiles by high-capacity viral vectors. Gene Therapy, 2014, 21, 855-865.	2.3	47
51	Next-generation sequencing for disorders of low and high bone mineral density. Osteoporosis International, 2013, 24, 2253-2259.	1.3	46
52	De Novo Heterozygous POLR2A Variants Cause a Neurodevelopmental Syndrome with Profound Infantile-Onset Hypotonia. American Journal of Human Genetics, 2019, 105, 283-301.	2.6	46
53	Differential inhibition of PDKs by phenylbutyrate and enhancement of pyruvate dehydrogenase complex activity by combination with dichloroacetate. Journal of Inherited Metabolic Disease, 2015, 38, 895-904.	1.7	45
54	Autophagy master regulator TFEB induces clearance of toxic SERPINA1/Î±-1-antitrypsin polymers. Autophagy, 2013, 9, 1094-1096.	4.3	44

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55	Xp11.2 microduplications including IQSEC2, TSPYL2 and KDM5C genes in patients with neurodevelopmental disorders. European Journal of Human Genetics, 2016, 24, 373-380.	1.4	43
56	Novel diagnostic DNA methylation episignatures expand and refine the epigenetic landscapes of Mendelian disorders. Human Genetics and Genomics Advances, 2022, 3, 100075.	1.0	42
57	Systemic hypertension in two patients with ASL deficiency: A result of nitric oxide deficiency?. Molecular Genetics and Metabolism, 2009, 98, 195-197.	0.5	41
58	X-linked recessive chondrodysplasia punctata: Spectrum of arylsulfatase E gene mutations and expanded clinical variability. , 2002, 117A, 164-168.		40
59	WDR35 mutation in siblings with Sensenbrenner syndrome: A ciliopathy with variable phenotype. American Journal of Medical Genetics, Part A, 2012, 158A, 2917-2924.	0.7	40
60	Identification of three novel SEDL mutations, including mutation in the rare, non-canonical splice site of exon 4. Clinical Genetics, 2003, 64, 235-242.	1.0	39
61	Enhancement of hepatic autophagy increases ureagenesis and protects against hyperammonemia. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, 391-396.	3.3	39
62	Prevalence and Relevance of Pre-Existing Anti-Adeno-Associated Virus Immunity in the Context of Gene Therapy for Criglerâ€“Najjar Syndrome. Human Gene Therapy, 2019, 30, 1297-1305.	1.4	39
63	Helper-dependent adenoviral vectors for liver-directed gene therapy of primary hyperoxaluria type 1. Gene Therapy, 2016, 23, 129-134.	2.3	37
64	Characterization of liver involvement in defects of cholesterol biosynthesis: Long-term follow-up and review. American Journal of Medical Genetics, Part A, 2005, 132A, 144-151.	0.7	36
65	Balloon Catheter Delivery of Helper-dependent Adenoviral Vector Results in Sustained, Therapeutic hFIX Expression in Rhesus Macaques. Molecular Therapy, 2012, 20, 1863-1870.	3.7	35
66	Enhancing Autophagy with Drugs or Lung-directed Gene Therapy Reverses the Pathological Effects of Respiratory Epithelial Cell Proteinopathy. Journal of Biological Chemistry, 2015, 290, 29742-29757.	1.6	35
67	Mutations in MAST1 Cause Mega-Corpus-Callosum Syndrome with Cerebellar Hypoplasia and Cortical Malformations. Neuron, 2018, 100, 1354-1368.e5.	3.8	35
68	Contiguous gene syndrome due to an interstitial deletion in Xp22.3 in a boy with ichthyosis, chondrodysplasia punctata, mental retardation and ADHD. European Journal of Medical Genetics, 2007, 50, 301-308.	0.7	34
69	MyD88-Dependent Silencing of Transgene Expression During the Innate and Adaptive Immune Response to Helper-Dependent Adenovirus. Human Gene Therapy, 2010, 21, 325-336.	1.4	31
70	SMAD4 mutations causing Myhre syndrome result in disorganization of extracellular matrix improved by losartan. European Journal of Human Genetics, 2014, 22, 988-994.	1.4	31
71	Complex care of individuals with multiple sulfatase deficiency: Clinical cases and consensus statement. Molecular Genetics and Metabolism, 2018, 123, 337-346.	0.5	31
72	De novo SMARCA2 variants clustered outside the helicase domain cause a new recognizable syndrome with intellectual disability and blepharophimosis distinct from Nicolaidesâ€“Baraitser syndrome. Genetics in Medicine, 2020, 22, 1838-1850.	1.1	31

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73	Truncating SRCAP variants outside the Floating-Harbor syndrome locus cause a distinct neurodevelopmental disorder with a specific DNA methylation signature. American Journal of Human Genetics, 2021, 108, 1053-1068.	2.6	31
74	Alpha-1 antitrypsin deficiency: A re-surfacing adult liver disorder. Journal of Hepatology, 2022, 76, 946-958.	1.8	30
75	Short-term Correction of Arginase Deficiency in a Neonatal Murine Model With a Helper-dependent Adenoviral Vector. Molecular Therapy, 2009, 17, 1155-1163.	3.7	29
76	X-linked recessive chondrodysplasia punctata due to a new point mutation of the ARSE gene. , 1997, 73, 139-143.		28
77	In Silico Modeling of Liver Metabolism in a Human Disease Reveals a Key Enzyme for Histidine and Histamine Homeostasis. Cell Reports, 2016, 15, 2292-2300.	2.9	28
78	Progress Towards Liver and Lung-Directed Gene Therapy with Helper- Dependent Adenoviral Vectors. Current Gene Therapy, 2009, 9, 329-340.	0.9	27
79	Dilation of the aortic root in mitochondrial disease patients. Molecular Genetics and Metabolism, 2011, 103, 167-170.	0.5	27
80	Assessment of bone mineral status in children with Marfan syndrome. American Journal of Medical Genetics, Part A, 2012, 158A, 2221-2224.	0.7	26
81	De novo PIK3R2 variant causes polymicrogyria, corpus callosum hyperplasia and focal cortical dysplasia. European Journal of Human Genetics, 2016, 24, 1359-1362.	1.4	26
82	Biochemical phenotyping unravels novel metabolic abnormalities and potential biomarkers associated with treatment of GLUT1 deficiency with ketogenic diet. PLoS ONE, 2017, 12, e0184022.	1.1	26
83	Progress and challenges in development of new therapies for urea cycle disorders. Human Molecular Genetics, 2019, 28, R42-R48.	1.4	26
84	Cervical spine stenosis and possible vitamin K deficiency embryopathy in an unusual case of chondrodysplasia punctata and an updated classification system. American Journal of Medical Genetics Part A, 2003, 122A, 70-75.	2.4	25
85	Immunodeficiency, centromeric instability, facial anomalies (ICF) syndrome, due to ZBTB24 mutations, presenting with large cerebral cyst. American Journal of Medical Genetics, Part A, 2012, 158A, 2043-2046.	0.7	25
86	Gene therapy with helper-dependent adenoviral vectors: lessons from studies in large animal models. Virus Genes, 2017, 53, 684-691.	0.7	25
87	Downâ€regulation of hepatocyte nuclear factorâ€4Î± and defective zonation in livers expressing mutant Z Î±1â€antitrypsin. Hepatology, 2017, 66, 124-135.	3.6	25
88	Gray matter heterotopias and brachytelephalangic chondrodysplasia punctata: A complication of hyperemesis gravidarum induced vitamin K deficiency?. American Journal of Medical Genetics, Part A, 2007, 143A, 200-204.	0.7	24
89	Activation of the câ€Jun Nâ€terminal kinase pathway aggravates proteotoxicity of hepatic mutant Z alpha1â€antitrypsin. Hepatology, 2017, 65, 1865-1874.	3.6	24
90	Expanding the phenotype of DST â€related disorder: A case report suggesting a genotype/phenotype correlation. American Journal of Medical Genetics, Part A, 2017, 173, 2743-2746.	0.7	23

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91	Phenotypic correction of ornithine transcarbamylase deficiency using low dose helperâ€dependent adenoviral vectors. Journal of Gene Medicine, 2008, 10, 890-896.	1.4	22
92	Current Status on Clinical Development of Adeno-Associated Virus-Mediated Liver-Directed Gene Therapy for Inborn Errors of Metabolism. Human Gene Therapy, 2019, 30, 1204-1210.	1.4	22
93	Maternal vitamin K deficient embryopathy: Association with hyperemesis gravidarum and Crohn disease. American Journal of Medical Genetics, Part A, 2013, 161, 417-429.	0.7	21
94	Ammonia and autophagy: An emerging relationship with implications for disorders with hyperammonemia. Journal of Inherited Metabolic Disease, 2019, 42, 1097-1104.	1.7	20
95	Robinow syndrome: Phenotypic variability in a family with a novel intragenic ROR2 mutation. American Journal of Medical Genetics, Part A, 2008, 146A, 2804-2809.	0.7	19
96	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.	1.4	19
97	Mutations in the PCYT1A gene are responsible for isolated forms of retinal dystrophy. European Journal of Human Genetics, 2017, 25, 651-655.	1.4	19
98	Variants in the degron of AFF3 are associated with intellectual disability, mesomelic dysplasia, horseshoe kidney, and epileptic encephalopathy. American Journal of Human Genetics, 2021, 108, 857-873.	2.6	19
99	Bioengineered Factor IX Molecules with Increased Catalytic Activity Improve the Therapeutic Index of Gene Therapy Vectors for Hemophilia B. Human Gene Therapy, 2009, 20, 479-485.	1.4	18
100	SR-A and SREC-I binding peptides increase HDAd-mediated liver transduction. Gene Therapy, 2014, 21, 950-957.	2.3	18
101	RARS1â€related hypomyelinating leukodystrophy: Expanding the spectrum. Annals of Clinical and Translational Neurology, 2020, 7, 83-93.	1.7	18
102	Gene therapy for inborn errors of liver metabolism. Molecular Genetics and Metabolism, 2005, 86, 13-24.	0.5	17
103	Conditional disruption of hepatic carbamoyl phosphate synthetase 1 in mice results in hyperammonemia without orotic aciduria and can be corrected by liver-directed gene therapy. Molecular Genetics and Metabolism, 2018, 124, 243-253.	0.5	17
104	Heterozygous ANKRD17 loss-of-function variants cause a syndrome with intellectual disability, speech delay, and dysmorphism. American Journal of Human Genetics, 2021, 108, 1138-1150.	2.6	17
105	Rare and de novo coding variants in chromodomain genes in Chiari I malformation. American Journal of Human Genetics, 2021, 108, 100-114.	2.6	17
106	Phenylbutyrate increases activity of pyruvate dehydrogenase complex. Oncotarget, 2013, 4, 804-805.	0.8	17
107	Correction of Hyperbilirubinemia in Gunn Rats Using Clinically Relevant Low Doses of Helper-Dependent Adenoviral Vectors. Human Gene Therapy, 2011, 22, 483-488.	1.4	16
108	A case of 14q11.2 microdeletion with autistic features, severe obesity and facial dysmorphisms suggestive of Wolfâ€“Hirschhorn syndrome. American Journal of Medical Genetics, Part A, 2014, 164, 190-193.	0.7	16

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109	Progress toward improved therapies for inborn errors of metabolism. Human Molecular Genetics, 2016, 25, R27-R35.	1.4	16
110	Rubinsteinâ€“Taybi syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2020, 182, 2939-2950.	0.7	16
111	CHOP and c-JUN up-regulate the mutant Z Î±1-antitrypsin, exacerbating its aggregation and liver proteotoxicity. Journal of Biological Chemistry, 2020, 295, 13213-13223.	1.6	16
112	Ensuring continuity of care for children with inherited metabolic diseases at the time of COVID-19: the experience of a metabolic unit in Italy. Genetics in Medicine, 2020, 22, 1178-1180.	1.1	16
113	Expansion of the phenotypic spectrum of de novo missense variants in kinesin family member 1A () Tj ETQq1 1 0.784314 rgBT /Overlock	1.1	16
114	Up-regulation of miR-34b/c by JNK and FOXO3 protects from liver fibrosis. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, .	3.3	16
115	Beclinâ€1â€mediated activation of autophagy improves proximal and distal urea cycle disorders. EMBO Molecular Medicine, 2021, 13, e13158.	3.3	16
116	Phenylbutyrate increases pyruvate dehydrogenase complex activity in cells harboring a variety of defects. Annals of Clinical and Translational Neurology, 2014, 1, 462-470.	1.7	15
117	A child with Myhre syndrome presenting with corectopia and tetralogy of Fallot. American Journal of Medical Genetics, Part A, 2018, 176, 426-430.	0.7	15
118	Targeting autophagy for therapy of hyperammonemia. Autophagy, 2018, 14, 1273-1275.	4.3	15
119	Long-Term Efficacy of T3 Analogue Triac in Children and Adults With MCT8 Deficiency: A Real-Life Retrospective Cohort Study. Journal of Clinical Endocrinology and Metabolism, 2022, 107, e1136-e1147.	1.8	15
120	Intrauterine growth retardation and placental vacuolization as presenting features in a case of GM1 gangliosidosis. Journal of Inherited Metabolic Disease, 2007, 30, 823-823.	1.7	14
121	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.	1.4	14
122	A Severe Case of Dentatorubro-Pallidoluysian Atrophy (DRPLA) with Microcephaly, Very Early Onset of Seizures, and Cerebral White Matter Involvement. Neuropediatrics, 2006, 37, 308-311.	0.3	13
123	15q13q14 deletions: Phenotypic characterization and molecular delineation by comparative genomic hybridization. American Journal of Medical Genetics, Part A, 2008, 146A, 1933-1941.	0.7	13
124	Brain Proton Magnetic Resonance Spectroscopy and Neuromuscular Pathology in a Patient With GM1 Gangliosidosis. Journal of Child Neurology, 2008, 23, 73-78.	0.7	13
125	Correction of Hyperbilirubinemia in Gunn Rats by Surgical Delivery of Low Doses of Helper-Dependent Adenoviral Vectors. Human Gene Therapy Methods, 2014, 25, 181-186.	2.1	13
126	Microdeletion of pseudogene chr14.232.a affects LRFN5 expression in cells of a patient with autism spectrum disorder. European Journal of Human Genetics, 2019, 27, 1475-1480.	1.4	13

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127	Sphingolipid Metabolism Perturbations in Rett Syndrome. Metabolites, 2019, 9, 221.	1.3	12
128	Diagnostic issues faced by a rare disease healthcare network during Covid-19 outbreak: data from the Campania Rare Disease Registry. Journal of Public Health, 2022, 44, 586-594.	1.0	12
129	Characterization of de novo microdeletions involving 17q11.2q12 identified through chromosomal comparative genomic hybridization. Clinical Genetics, 2007, 72, 411-419.	1.0	11
130	Vasoactive Intestinal Peptide Increases Hepatic Transduction and Reduces Innate Immune Response Following Administration of Helper-dependent Ad. Molecular Therapy, 2010, 18, 1339-1345.	3.7	11
131	Focal congenital lipoatrophy and vascular malformation: A mild form of inverse Klippelâ€“Trenaunay syndrome?. European Journal of Medical Genetics, 2012, 55, 705-707.	0.7	11
132	An extremely severe phenotype attributed to WDR81 nonsense mutations. Annals of Neurology, 2017, 82, 650-651.	2.8	11
133	Lowry-Wood syndrome: further evidence of association with RNU4ATAC, and correlation between genotype and phenotype. Human Genetics, 2018, 137, 905-909.	1.8	11
134	Intrafamilial variability in SPTAN1-related disorder: From benign convulsions with mild gastroenteritis to developmental encephalopathy. European Journal of Paediatric Neurology, 2020, 28, 237-239.	0.7	11
135	Liver-directed gene-based therapies for inborn errors of metabolism. Expert Opinion on Biological Therapy, 2021, 21, 229-240.	1.4	11
136	ZTTK syndrome: Clinical and molecular findings ofÂ15 cases and a review of the literature. American Journal of Medical Genetics, Part A, 2021, 185, 3740-3753.	0.7	11
137	Rapidly Progressive Neurological Deterioration in a Child with Alpers Syndrome Exhibiting a Previously Unremarkable Brain MRI. Neuropediatrics, 2008, 39, 179-183.	0.3	10
138	Intrathecal Injection of Helper-Dependent Adenoviral Vectors Results in Long-Term Transgene Expression in Neuroependymal Cells and Neurons. Human Gene Therapy, 2011, 22, 745-751.	1.4	10
139	30-year follow-up of a patient with classic citrullinemia. Molecular Genetics and Metabolism, 2012, 106, 248-250.	0.5	10
140	Improved Efficacy and Reduced Toxicity by Ultrasound-Guided Intrahepatic Injections of Helper-Dependent Adenoviral Vector in Gunn Rats. Human Gene Therapy Methods, 2013, 24, 321-327.	2.1	10
141	Gene Therapy for Inherited Diseases of Liver Metabolism. Human Gene Therapy, 2015, 26, 186-192.	1.4	10
142	Geleophysic dysplasia: novel missense variants and insights into ADAMTSL2 intracellular trafficking. Molecular Genetics and Metabolism Reports, 2019, 21, 100504.	0.4	10
143	A systematic cross-sectional survey of multiple sulfatase deficiency. Molecular Genetics and Metabolism, 2020, 130, 283-288.	0.5	10
144	Progress towards the clinical application of helper-dependent adenoviral vectors for liver and lung gene therapy. Current Opinion in Molecular Therapeutics, 2006, 8, 446-54.	2.8	10

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145	Terminal osseous dysplasia with pigmentary defects (TODPD): Followâ€up of the first reported family, characterization of the radiological phenotype, and refinement of the linkage region. American Journal of Medical Genetics, Part A, 2010, 152A, 1825-1831.	0.7	9
146	Autosomal Dominant MÃ©nÃ©trierâ€like Disease. Journal of Pediatric Gastroenterology and Nutrition, 2012, 55, 717-720.	0.9	9
147	Challenges and Prospects for Helper-Dependent Adenoviral Vector-Mediated Gene Therapy. Biomedicines, 2014, 2, 132-148.	1.4	9
148	Expansion of the phenotype of lateral meningocele syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 1259-1262.	0.7	9
149	Child Neurology: Recurrent rhabdomyolysis due to a fatty acid oxidation disorder. Neurology, 2014, 82, e1-4.	1.5	8
150	Skin fibroblasts of patients with geleophysic dysplasia due to FBN1 mutations have lysosomal inclusions and losartan improves their microfibril deposition defect. Molecular Genetics & Genomic Medicine, 2019, 7, e844.	0.6	8
151	Retinal dystrophy in an individual carrying a de novo missense variant of SMARCA4. Molecular Genetics & Genomic Medicine, 2019, 7, e682.	0.6	8
152	FBXO28 causes developmental and epileptic encephalopathy with profound intellectual disability. Epilepsia, 2021, 62, e13-e21.	2.6	8
153	Epigenetic Alterations in Inborn Errors of Immunity. Journal of Clinical Medicine, 2022, 11, 1261.	1.0	8
154	Disease burden and management of <scp>Criglerâ€Najjar</scp> syndrome: Report of a world registry. Liver International, 2022, 42, 1593-1604.	1.9	8
155	Sustained Reduction of Hyperbilirubinemia in Gunn Rats After Adeno-Associated Virus-Mediated Gene Transfer of Bilirubin UDP-Glucuronosyltransferase Isozyme 1A1 to Skeletal Muscle. Human Gene Therapy, 2012, 23, 1082-1089.	1.4	7
156	MIB2variants altering NOTCH signalling result in left ventricle hypertrabeculation/non-compaction and are associated with MÃ©nÃ©trier-like gastropathy. Human Molecular Genetics, 2016, 26, ddw365.	1.4	7
157	Severe presentation and complex brain malformations in an individual carrying a CCND2 variant. Molecular Genetics & Genomic Medicine, 2019, 7, e708.	0.6	7
158	Hepatic glutamine synthetase augmentation enhances ammonia detoxification. Journal of Inherited Metabolic Disease, 2019, 42, 1128-1135.	1.7	7
159	Cavitating and tigroidâ€like leukoencephalopathy in a case of NDUFA2â€related disorder. JIMD Reports, 2020, 52, 11-16.	0.7	7
160	The evolving landscape of gene therapy for congenital haemophilia: An unprecedented, problematic but promising opportunity for worldwide clinical studies. Blood Reviews, 2021, 46, 100737.	2.8	7
161	Biâ€allelic KARS1 pathogenic variants affecting functions of cytosolic and mitochondrial isoforms are associated with a progressive and multisystem disease. Human Mutation, 2021, 42, 745-761.	1.1	7
162	A new patient with Lowry-Wood syndrome with mild phenotype. American Journal of Medical Genetics Part A, 2003, 118A, 68-70.	2.4	6

#	Article	IF	Citations
163	von Voss-Cherstvoy syndrome with transient thrombocytopenia and normal psychomotor development. American Journal of Medical Genetics Part A, 2004, 126A, 299-302.	2.4	6
164	Parkes Weber syndrome occurring in a family with capillary malformations. Clinical Dysmorphology, 2007, 16, 167-171.	0.1	6
165	Spondylocarpotarsal synostosis: Longâ€term followâ€up of a case due to FLNB mutations. American Journal of Medical Genetics, Part A, 2008, 146A, 1230-1233.	0.7	6
166	Infiltrating giant cell tumor in a case of Pagetâ€™s disease of bone. Archives of Osteoporosis, 2009, 4, 91-94.	1.0	6
167	Giant breast tumors in a patient with Beckwithâ€“Wiedemann syndrome. American Journal of Medical Genetics, Part A, 2014, 164, 182-185.	0.7	6
168	AAV-mediated liver-directed gene therapy for Acute Intermittent Porphyria: It is safe but is it effective?. Journal of Hepatology, 2016, 65, 666-667.	1.8	6
169	A pilot clinical trial with losartan in Myhre syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 702-709.	0.7	6
170	Expanding the phenotype of <scp>HNRNPU</scp>â€related neurodevelopmental disorder with emphasis on seizure phenotype and review of literature. American Journal of Medical Genetics, Part A, 2022, 188, 1497-1514.	0.7	6
171	Refinement of the clinical and mutational spectrum of <scp>UBE2A</scp> deficiency syndrome. Clinical Genetics, 2020, 98, 172-178.	1.0	5
172	Milder presentation of TELO2-related syndrome in two sisters homozygous for the p.Arg609His pathogenic variant. European Journal of Medical Genetics, 2021, 64, 104116.	0.7	5
173	A novel SHANK3 interstitial microdeletion in a family with intellectual disability and brain MRI abnormalities resembling Unidentified Bright Objects. European Journal of Paediatric Neurology, 2017, 21, 902-906.	0.7	5
174	Epilepsy in KAT6A syndrome: Description of two individuals and revision of the literature. European Journal of Medical Genetics, 2022, 65, 104380.	0.7	5
175	De Novo ATP1A1 Variants in an Early-Onset Complex Neurodevelopmental Syndrome. Neurology, 2022, 98, 440-445.	1.5	5
176	Case report: pathological features of aberrant pancreatic development in congenital hyperinsulinism due to ABCC8 mutations. Annals of Clinical and Laboratory Science, 2008, 38, 386-9.	0.2	5
177	Liver-Directed Adeno-Associated Virusâ€“Mediated Gene Therapy for Mucopolysaccharidosis Type VI. , 2022, 1, .		5
178	Inappropriate tall stature and renal ectopy in a male patient with X-linked congenital adrenal hypoplasia due to a novel missense mutation in theDAX-1 gene. American Journal of Medical Genetics, Part A, 2005, 135A, 72-74.	0.7	4
179	Gene therapy for inborn errors of liver metabolism: progress towards clinical applications. Italian Journal of Pediatrics, 2008, 34, 2.	1.0	4
180	Reply to Amor et al. European Journal of Human Genetics, 2012, 20, 597-597.	1.4	4

#	Article	IF	Citations
181	Terminal osseous dysplasia with pigmentary defects (TODPD) due to a recurrent filamin A (FLNA) mutation. Molecular Genetics & Genomic Medicine, 2014, 2, 467-471.	0.6	4
182	Helper-Dependent Adenoviral Vectors. , 2016, , 423-450.		4
183	Pain and sleep disturbances in Rett syndrome and other neurodevelopmental disorders. Acta Paediatrica, International Journal of Paediatrics, 2019, 108, 171-172.	0.7	4
184	A small 7q11.23 microduplication involving <scp>GTF2I</scp> in a family with intellectual disability. Clinical Genetics, 2020, 97, 940-942.	1.0	4
185	Clinical and Functional Consequences of C-Terminal Variants in MCT8: A Case Series. Journal of Clinical Endocrinology and Metabolism, 2021, 106, 539-553.	1.8	4
186	Recurrent de novo missense variants in GNB2 can cause syndromic intellectual disability. Journal of Medical Genetics, 2022, 59, 511-516.	1.5	4
187	Mild Clinical Presentation of Joubert Syndrome in a Male Adult Carrying Biallelic MKS1 Truncating Variants. Diagnostics, 2021, 11, 1218.	1.3	4
188	De novo terminal 22q12.3q13.3 duplication with pituitary hypoplasia. American Journal of Medical Genetics, Part A, 2009, 149A, 2554-2556.	0.7	3
189	Chromosomal 17p13.3 microdeletion unmasking recessive Canavan disease mutation. Molecular Genetics and Metabolism, 2011, 104, 706-707.	0.5	3
190	Low-Dose Amitriptyline-Induced Acute Dystonia in a Patient with Metachromatic Leukodystrophy. JIMD Reports, 2012, 9, 113-116.	0.7	3
191	Gait disturbance and lower limb pain in a patient with PIK3CA -related disorder. European Journal of Medical Genetics, 2017, 60, 655-657.	0.7	3
192	Two cases of 16q12.1q21 deletions and refinement of the critical region. European Journal of Medical Genetics, 2020, 63, 103878.	0.7	3
193	Biallelic variants in <scp> CENPF </scp> causing a phenotype distinct from StrÃ¸mme syndrome. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2022, , .	0.7	3
194	683. Bronchoscope-Guided, Targeted Lobar Aersolization of HDAd into the Lungs of Nonhuman Primate Results in Exceedingly High Pulmonary Transduction Uniformally throughout the Entire Lung with Negligible Toxicity. Molecular Therapy, 2006, 13, S264.	3.7	2
195	Progressive Myopathy With Multiple Symmetric Lipomatosis. Archives of Neurology, 2009, 66, 1576-7.	4.9	2
196	Cutis laxa and fatal pulmonary hypertension. Clinical Dysmorphology, 2011, 20, 77-81.	0.1	2
197	<scp>AP</scp>1S2â€truncating variant in a patient with severe neurodevelopmental disorder and cerebral folate deficiency. Acta Paediatrica, International Journal of Paediatrics, 2019, 108, 564-565.	0.7	2
198	Mechanisms of liver disease in AATD. , 2019, , 93-104.		2

#	Article	IF	Citations
199	Expanded cardiovascular phenotype of Myhre syndrome includes tetralogy of Fallot suggesting a role for <scp>SMAD4</scp> in human neural crest defects. American Journal of Medical Genetics, Part A, 2022, 188, 1384-1395.	0.7	2
200	Two familial cases of high blood galactose of unknown aetiology. Journal of Inherited Metabolic Disease, 2006, 29, 762-762.	1.7	1
201	A novel mutation in the N-terminal region of the CYP17A1 gene in a patient with 17Î±-hydroxylase/17,20-lyase deficiency. Journal of Endocrinological Investigation, 2009, 32, 322-324.	1.8	1
202	Recent progress in gene therapies for mucopolysaccharidoses. Expert Opinion on Orphan Drugs, 2018, 6, 611-623.	0.5	1
203	Dual diagnosis in a child with familial SCN8A-related encephalopathy complicated by a 1p13.2 deletion involving NRAS gene. Neurological Sciences, 2021, 42, 2115-2117.	0.9	1
204	Helper-dependent adenoviral vectors. , 2010, , 193-207.		1
205	Helper-Dependent Adenoviral Vectors for Gene Therapy. , 2015, , 47-84.		1
206	Mild neurological phenotype in a family carrying a novel N-terminal null GRIN2A variant. European Journal of Medical Genetics, 2022, 65, 104500.	0.7	1
207	Inappropriate tall stature and renal ectopy in a male patient with X-linked congenital adrenal hypoplasia due to a novel missense mutation in theDAX-1 gene. American Journal of Medical Genetics, Part A, 2005, 137A, 115-115.	0.7	0
208	213. Improving the Therapeutic Index of Helper-Dependent Adenoviral Vector for Crigler-Najjar Gene Therapy. Molecular Therapy, 2006, 13, S82.	3.7	0
209	975. TLR9 Activation Is Involved in the Immune Response to Helper Dependent Adenoviral Vectors. Molecular Therapy, 2006, 13, S375.	3.7	0
210	De novo terminal 22q12.3q13.3 duplication with pituitary hypoplasia (Am J Med Genet Part A) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 302	0.7	0
211	Reply. Hepatology, 2017, 66, 677-678.	3.6	0
212	Listen to Your Patients: A Diagnostic Clue. Journal of Pediatrics, 2020, 224, 171.	0.9	0
213	Longâ€term followâ€up of an individual with <scp>ITPR1</scp>â€related disorder. American Journal of Medical Genetics, Part A, 2020, 182, 1846-1847.	0.7	0
214	Lack of resemblance between Myhre syndrome and other â€œsegmental progeroidâ€•syndromes warrants restraint in applying this classification. GeroScience, 2021, 43, 459-461.	2.1	0
215	Peculiar footprints in a child with agenesis of corpus callosum. Journal of Paediatrics and Child Health, 2021, 57, 450-451.	0.4	0
216	Helper-Dependent Adenoviral Vectors for Gene Therapy of Inherited Diseases. , 2017, , 61-75.		0

#	Article	IF	Citations
217	Cholangiopathies and the noncoding revolution. Current Opinion in Gastroenterology, 2022, 38, 128-135.	1.0	0
218	Liver gene therapy: The magic bullet for the sick lung. Molecular Therapy - Methods and Clinical Development, 2022, 26, 72-73.	1.8	0