

Nicola Brunetti-Pierri

List of Publications by Year in descending order

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Version: 2024-02-01

218
papers

16,655
citations

50276

46
h-index

16650

123
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228
all docs

228
docs citations

228
times ranked

31589
citing authors

#	ARTICLE	IF	CITATIONS
1	Diagnostic issues faced by a rare disease healthcare network during Covid-19 outbreak: data from the Campania Rare Disease Registry. <i>Journal of Public Health</i> , 2022, 44, 586-594.	1.8	12
2	Recurrent <i>de novo</i> missense variants in <i>GNB2</i> can cause syndromic intellectual disability. <i>Journal of Medical Genetics</i> , 2022, 59, 511-516.	3.2	4
3	Long-Term Efficacy of T3 Analogue Triac in Children and Adults With MCT8 Deficiency: A Real-Life Retrospective Cohort Study. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2022, 107, e1136-e1147.	3.6	15
4	Epilepsy in KAT6A syndrome: Description of two individuals and revision of the literature. <i>European Journal of Medical Genetics</i> , 2022, 65, 104380.	1.3	5
5	Alpha-1 antitrypsin deficiency: A re-surfacing adult liver disorder. <i>Journal of Hepatology</i> , 2022, 76, 946-958.	3.7	30
6	Novel diagnostic DNA methylation epigenatures expand and refine the epigenetic landscapes of Mendelian disorders. <i>Human Genetics and Genomics Advances</i> , 2022, 3, 100075.	1.7	42
7	Cholangiopathies and the noncoding revolution. <i>Current Opinion in Gastroenterology</i> , 2022, 38, 128-135.	2.3	0
8	Expanded cardiovascular phenotype of Myhre syndrome includes tetralogy of Fallot suggesting a role for <i>SMAD4</i> in human neural crest defects. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 1384-1395.	1.2	2
9	De Novo <i>ATP1A1</i> Variants in an Early-Onset Complex Neurodevelopmental Syndrome. <i>Neurology</i> , 2022, 98, 440-445.	1.1	5
10	Expanding the phenotype of <i>HNRNPU</i> -related neurodevelopmental disorder with emphasis on seizure phenotype and review of literature. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 1497-1514.	1.2	6
11	Epigenetic Alterations in Inborn Errors of Immunity. <i>Journal of Clinical Medicine</i> , 2022, 11, 1261.	2.4	8
12	Disease burden and management of <i>Crigler-Najjar</i> syndrome: Report of a world registry. <i>Liver International</i> , 2022, 42, 1593-1604.	3.9	8
13	Mild neurological phenotype in a family carrying a novel N-terminal null <i>GRIN2A</i> variant. <i>European Journal of Medical Genetics</i> , 2022, 65, 104500.	1.3	1
14	Biallelic variants in <i>CENPF</i> causing a phenotype distinct from Strømme syndrome. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2022, , .	1.6	3
15	Liver-Directed Adeno-Associated Virus-Mediated Gene Therapy for Mucopolysaccharidosis Type VI. , 2022, 1, .		5
16	Liver gene therapy: The magic bullet for the sick lung. <i>Molecular Therapy - Methods and Clinical Development</i> , 2022, 26, 72-73.	4.1	0
17	The evolving landscape of gene therapy for congenital haemophilia: An unprecedented, problematic but promising opportunity for worldwide clinical studies. <i>Blood Reviews</i> , 2021, 46, 100737.	5.7	7
18	Clinical and Functional Consequences of C-Terminal Variants in MCT8: A Case Series. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2021, 106, 539-553.	3.6	4

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19	Milder presentation of TELO2-related syndrome in two sisters homozygous for the p.Arg609His pathogenic variant. <i>European Journal of Medical Genetics</i> , 2021, 64, 104116.	1.3	5
20	<i>FBXO28</i> causes developmental and epileptic encephalopathy with profound intellectual disability. <i>Epilepsia</i> , 2021, 62, e13-e21.	5.1	8
21	Dual diagnosis in a child with familial SCN8A-related encephalopathy complicated by a 1p13.2 deletion involving NRAS gene. <i>Neurological Sciences</i> , 2021, 42, 2115-2117.	1.9	1
22	Liver-directed gene-based therapies for inborn errors of metabolism. <i>Expert Opinion on Biological Therapy</i> , 2021, 21, 229-240.	3.1	11
23	Lack of resemblance between Myhre syndrome and other â€œsegmental progeroidâ€ syndromes warrants restraint in applying this classification. <i>GeroScience</i> , 2021, 43, 459-461.	4.6	0
24	Up-regulation of miR-34b/c by JNK and FOXO3 protects from liver fibrosis. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2021, 118, .	7.1	16
25	Peculiar footprints in a child with agenesis of corpus callosum. <i>Journal of Paediatrics and Child Health</i> , 2021, 57, 450-451.	0.8	0
26	Biallelic <i>KARS1</i> pathogenic variants affecting functions of cytosolic and mitochondrial isoforms are associated with a progressive and multisystem disease. <i>Human Mutation</i> , 2021, 42, 745-761.	2.5	7
27	Variants in the degron of AFF3 are associated with intellectual disability, mesomelic dysplasia, horseshoe kidney, and epileptic encephalopathy. <i>American Journal of Human Genetics</i> , 2021, 108, 857-873.	6.2	19
28	Truncating SRCAP variants outside the Floating-Harbor syndrome locus cause a distinct neurodevelopmental disorder with a specific DNA methylation signature. <i>American Journal of Human Genetics</i> , 2021, 108, 1053-1068.	6.2	31
29	Heterozygous ANKRD17 loss-of-function variants cause a syndrome with intellectual disability, speech delay, and dysmorphism. <i>American Journal of Human Genetics</i> , 2021, 108, 1138-1150.	6.2	17
30	ZTTK syndrome: Clinical and molecular findings of 15 cases and a review of the literature. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 3740-3753.	1.2	11
31	Mild Clinical Presentation of Joubert Syndrome in a Male Adult Carrying Biallelic MKS1 Truncating Variants. <i>Diagnostics</i> , 2021, 11, 1218.	2.6	4
32	Rare and de novo coding variants in chromodomain genes in Chiari I malformation. <i>American Journal of Human Genetics</i> , 2021, 108, 100-114.	6.2	17
33	Beclin-1-mediated activation of autophagy improves proximal and distal urea cycle disorders. <i>EMBO Molecular Medicine</i> , 2021, 13, e13158.	6.9	16
34	A pilot clinical trial with losartan in Myhre syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 702-709.	1.2	6
35	<i>RARS1</i> -related hypomyelinating leukodystrophy: Expanding the spectrum. <i>Annals of Clinical and Translational Neurology</i> , 2020, 7, 83-93.	3.7	18
36	Rubinstein-Taybi syndrome in diverse populations. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 2939-2950.	1.2	16

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37	Disease characteristics of MCT8 deficiency: an international, retrospective, multicentre cohort study. <i>Lancet Diabetes and Endocrinology</i> , 2020, 8, 594-605.	11.4	50
38	De novo SMARCA2 variants clustered outside the helicase domain cause a new recognizable syndrome with intellectual disability and blepharophimosis distinct from Nicolaides-Baraitser syndrome. <i>Genetics in Medicine</i> , 2020, 22, 1838-1850.	2.4	31
39	Intrafamilial variability in SPTAN1-related disorder: From benign convulsions with mild gastroenteritis to developmental encephalopathy. <i>European Journal of Paediatric Neurology</i> , 2020, 28, 237-239.	1.6	11
40	CHOP and c-JUN up-regulate the mutant Z α 1-antitrypsin, exacerbating its aggregation and liver proteotoxicity. <i>Journal of Biological Chemistry</i> , 2020, 295, 13213-13223.	3.4	16
41	Refinement of the clinical and mutational spectrum of UBE2A deficiency syndrome. <i>Clinical Genetics</i> , 2020, 98, 172-178.	2.0	5
42	Listen to Your Patients: A Diagnostic Clue. <i>Journal of Pediatrics</i> , 2020, 224, 171.	1.8	0
43	Ensuring continuity of care for children with inherited metabolic diseases at the time of COVID-19: the experience of a metabolic unit in Italy. <i>Genetics in Medicine</i> , 2020, 22, 1178-1180.	2.4	16
44	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.	2.9	14
45	A systematic cross-sectional survey of multiple sulfatase deficiency. <i>Molecular Genetics and Metabolism</i> , 2020, 130, 283-288.	1.1	10
46	Long-term follow-up of an individual with ITPR1-related disorder. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 1846-1847.	1.2	0
47	Expansion of the phenotype of lateral meningocele syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 1259-1262.	1.2	9
48	Expansion of the phenotypic spectrum of de novo missense variants in kinesin family member 1A (KIF1A). <i>Journal of Medical Genetics</i> , 2020, 57, 107-110.	2.5	16
49	Two cases of 16q12.1q21 deletions and refinement of the critical region. <i>European Journal of Medical Genetics</i> , 2020, 63, 103878.	1.3	3
50	Cavitating and tigroid-like leukoencephalopathy in a case of NDUFA2-related disorder. <i>JIMD Reports</i> , 2020, 52, 11-16.	1.5	7
51	A small 7q11.23 microduplication involving GTF2I in a family with intellectual disability. <i>Clinical Genetics</i> , 2020, 97, 940-942.	2.0	4
52	Large-scale targeted sequencing identifies risk genes for neurodevelopmental disorders. <i>Nature Communications</i> , 2020, 11, 4932.	12.8	105
53	De Novo Heterozygous POLR2A Variants Cause a Neurodevelopmental Syndrome with Profound Infantile-Onset Hypotonia. <i>American Journal of Human Genetics</i> , 2019, 105, 283-301.	6.2	46
54	Skin fibroblasts of patients with geleophysic dysplasia due to FBN1 mutations have lysosomal inclusions and losartan improves their microfibril deposition defect. <i>Molecular Genetics & Genomic Medicine</i> , 2019, 7, e844.	1.2	8

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55	Paralog Studies Augment Gene Discovery: DDX and DHX Genes. American Journal of Human Genetics, 2019, 105, 302-316.	6.2	56
56	Sphingolipid Metabolism Perturbations in Rett Syndrome. Metabolites, 2019, 9, 221.	2.9	12
57	Loss of SMPD4 Causes a Developmental Disorder Characterized by Microcephaly and Congenital Arthrogyposis. American Journal of Human Genetics, 2019, 105, 689-705.	6.2	48
58	Prevalence and Relevance of Pre-Existing Anti-Adeno-Associated Virus Immunity in the Context of Gene Therapy for Crigler-Najjar Syndrome. Human Gene Therapy, 2019, 30, 1297-1305.	2.7	39
59	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.	2.7	22
60	Geleophysic dysplasia: novel missense variants and insights into ADAMTSL2 intracellular trafficking. Molecular Genetics and Metabolism Reports, 2019, 21, 100504.	1.1	10
61	Ammonia and autophagy: An emerging relationship with implications for disorders with hyperammonemia. Journal of Inherited Metabolic Disease, 2019, 42, 1097-1104.	3.6	20
62	Nutrient-sensitive transcription factors <i>TFEB</i> and <i>TFE3</i> couple autophagy and metabolism to the peripheral clock. EMBO Journal, 2019, 38, .	7.8	58
63	Progress and challenges in development of new therapies for urea cycle disorders. Human Molecular Genetics, 2019, 28, R42-R48.	2.9	26
64	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.	2.8	13
65	Severe presentation and complex brain malformations in an individual carrying a <i>CCND2</i> variant. Molecular Genetics & Genomic Medicine, 2019, 7, e708.	1.2	7
66	Retinal dystrophy in an individual carrying a de novo missense variant of SMARCA4. Molecular Genetics & Genomic Medicine, 2019, 7, e682.	1.2	8
67	Hepatic glutamine synthetase augmentation enhances ammonia detoxification. Journal of Inherited Metabolic Disease, 2019, 42, 1128-1135.	3.6	7
68	Pain and sleep disturbances in Rett syndrome and other neurodevelopmental disorders. Acta Paediatrica, International Journal of Paediatrics, 2019, 108, 171-172.	1.5	4
69	<i>AP1S2</i> truncating variant in a patient with severe neurodevelopmental disorder and cerebral folate deficiency. Acta Paediatrica, International Journal of Paediatrics, 2019, 108, 564-565.	1.5	2
70	Mechanisms of liver disease in AATD. , 2019, , 93-104.		2
71	Complex care of individuals with multiple sulfatase deficiency: Clinical cases and consensus statement. Molecular Genetics and Metabolism, 2018, 123, 337-346.	1.1	31
72	A child with Myhre syndrome presenting with corectopia and tetralogy of Fallot. American Journal of Medical Genetics, Part A, 2018, 176, 426-430.	1.2	15

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73	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.	7.1	39
74	gene2drug: a computational tool for pathway-based rational drug repositioning. Bioinformatics, 2018, 34, 1498-1505.	4.1	62
75	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.	1.1	17
76	Pyruvate dehydrogenase complex and lactate dehydrogenase are targets for therapy of acute liver failure. Journal of Hepatology, 2018, 69, 325-335.	3.7	65
77	Mutations in MAST1 Cause Mega-Corpus-Callosum Syndrome with Cerebellar Hypoplasia and Cortical Malformations. Neuron, 2018, 100, 1354-1368.e5.	8.1	35
78	Recent progress in gene therapies for mucopolysaccharidoses. Expert Opinion on Orphan Drugs, 2018, 6, 611-623.	0.8	1
79	Lowry-Wood syndrome: further evidence of association with RNU4ATAC, and correlation between genotype and phenotype. Human Genetics, 2018, 137, 905-909.	3.8	11
80	Induction of Nitric-Oxide Metabolism in Enterocytes Alleviates Colitis and Inflammation-Associated Colon Cancer. Cell Reports, 2018, 23, 1962-1976.	6.4	51
81	Targeting autophagy for therapy of hyperammonemia. Autophagy, 2018, 14, 1273-1275.	9.1	15
82	Activation of the c-Jun N-terminal kinase pathway aggravates proteotoxicity of hepatic mutant Z alpha1-antitrypsin. Hepatology, 2017, 65, 1865-1874.	7.3	24
83	Reply. Hepatology, 2017, 66, 677-678.	7.3	0
84	Gene therapy with helper-dependent adenoviral vectors: lessons from studies in large animal models. Virus Genes, 2017, 53, 684-691.	1.6	25
85	Mutations in the PCYT1A gene are responsible for isolated forms of retinal dystrophy. European Journal of Human Genetics, 2017, 25, 651-655.	2.8	19
86	Down-regulation of hepatocyte nuclear factor-4 and defective zonation in livers expressing mutant Z alpha1-antitrypsin. Hepatology, 2017, 66, 124-135.	7.3	25
87	An extremely severe phenotype attributed to <i>WDR81</i> nonsense mutations. Annals of Neurology, 2017, 82, 650-651.	5.3	11
88	Gait disturbance and lower limb pain in a patient with PIK3CA-related disorder. European Journal of Medical Genetics, 2017, 60, 655-657.	1.3	3
89	Expanding the phenotype of <i>DST</i> -related disorder: A case report suggesting a genotype/phenotype correlation. American Journal of Medical Genetics, Part A, 2017, 173, 2743-2746.	1.2	23
90	Biochemical phenotyping unravels novel metabolic abnormalities and potential biomarkers associated with treatment of GLUT1 deficiency with ketogenic diet. PLoS ONE, 2017, 12, e0184022.	2.5	26

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91	A novel SHANK3 interstitial microdeletion in a family with intellectual disability and brain MRI abnormalities resembling Unidentified Bright Objects. <i>European Journal of Paediatric Neurology</i> , 2017, 21, 902-906.	1.6	5
92	Helper-Dependent Adenoviral Vectors for Gene Therapy of Inherited Diseases. , 2017, , 61-75.		0
93	Helper-Dependent Adenoviral Vectors. , 2016, , 423-450.		4
94	MIB2variants altering NOTCH signalling result in left ventricle hypertrabeculation/non-compaction and are associated with MÃ©nÃ©trier-like gastropathy. <i>Human Molecular Genetics</i> , 2016, 26, ddw365.	2.9	7
95	AAV-mediated liver-directed gene therapy for Acute Intermittent Porphyria: It is safe but is it effective?. <i>Journal of Hepatology</i> , 2016, 65, 666-667.	3.7	6
96	In Silico Modeling of Liver Metabolism in a Human Disease Reveals a Key Enzyme for Histidine and Histamine Homeostasis. <i>Cell Reports</i> , 2016, 15, 2292-2300.	6.4	28
97	Guidelines for the use and interpretation of assays for monitoring autophagy (3rd edition). <i>Autophagy</i> , 2016, 12, 1-222.	9.1	4,701
98	De novo PIK3R2 variant causes polymicrogyria, corpus callosum hyperplasia and focal cortical dysplasia. <i>European Journal of Human Genetics</i> , 2016, 24, 1359-1362.	2.8	26
99	Helper-dependent adenoviral vectors for liver-directed gene therapy of primary hyperoxaluria type 1. <i>Gene Therapy</i> , 2016, 23, 129-134.	4.5	37
100	Progress toward improved therapies for inborn errors of metabolism. <i>Human Molecular Genetics</i> , 2016, 25, R27-R35.	2.9	16
101	Xp11.2 microduplications including IQSEC2, TSPYL2 and KDM5C genes in patients with neurodevelopmental disorders. <i>European Journal of Human Genetics</i> , 2016, 24, 373-380.	2.8	43
102	Gene Therapy for Inherited Diseases of Liver Metabolism. <i>Human Gene Therapy</i> , 2015, 26, 186-192.	2.7	10
103	Differential inhibition of PDKs by phenylbutyrate and enhancement of pyruvate dehydrogenase complex activity by combination with dichloroacetate. <i>Journal of Inherited Metabolic Disease</i> , 2015, 38, 895-904.	3.6	45
104	Enhancing Autophagy with Drugs or Lung-directed Gene Therapy Reverses the Pathological Effects of Respiratory Epithelial Cell Proteinopathy. <i>Journal of Biological Chemistry</i> , 2015, 290, 29742-29757.	3.4	35
105	Prevalence of Anti-Adeno-Associated Virus Serotype 8 Neutralizing Antibodies and Arylsulfatase B Cross-Reactive Immunologic Material in Mucopolysaccharidosis VI Patient Candidates for a Gene Therapy Trial. <i>Human Gene Therapy</i> , 2015, 26, 145-152.	2.7	19
106	Helper-Dependent Adenoviral Vectors for Gene Therapy. , 2015, , 47-84.		1
107	Challenges and Prospects for Helper-Dependent Adenoviral Vector-Mediated Gene Therapy. <i>Biomedicines</i> , 2014, 2, 132-148.	3.2	9
108	Retinal transduction profiles by high-capacity viral vectors. <i>Gene Therapy</i> , 2014, 21, 855-865.	4.5	47

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109	Child Neurology: Recurrent rhabdomyolysis due to a fatty acid oxidation disorder. <i>Neurology</i> , 2014, 82, e1-4.	1.1	8
110	Correction of Hyperbilirubinemia in Gunn Rats by Surgical Delivery of Low Doses of Helper-Dependent Adenoviral Vectors. <i>Human Gene Therapy Methods</i> , 2014, 25, 181-186.	2.1	13
111	Giant breast tumors in a patient with Beckwith-Wiedemann syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 182-185.	1.2	6
112	Phenylbutyrate increases pyruvate dehydrogenase complex activity in cells harboring a variety of defects. <i>Annals of Clinical and Translational Neurology</i> , 2014, 1, 462-470.	3.7	15
113	SR-A and SREC-I binding peptides increase HDAd-mediated liver transduction. <i>Gene Therapy</i> , 2014, 21, 950-957.	4.5	18
114	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.	2.8	31
115	A case of 14q11.2 microdeletion with autistic features, severe obesity and facial dysmorphisms suggestive of Wolf-Hirschhorn syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 190-193.	1.2	16
116	Wilson Disease Protein ATP7B Utilizes Lysosomal Exocytosis to Maintain Copper Homeostasis. <i>Developmental Cell</i> , 2014, 29, 686-700.	7.0	203
117	Terminal osseous dysplasia with pigmentary defects (TODPD) due to a recurrent filamin A (FLNA) mutation. <i>Molecular Genetics & Genomic Medicine</i> , 2014, 2, 467-471.	1.2	4
118	Next-generation sequencing for disorders of low and high bone mineral density. <i>Osteoporosis International</i> , 2013, 24, 2253-2259.	3.1	46
119	Improved Efficacy and Reduced Toxicity by Ultrasound-Guided Intrahepatic Injections of Helper-Dependent Adenoviral Vector in Gunn Rats. <i>Human Gene Therapy Methods</i> , 2013, 24, 321-327.	2.1	10
120	SR-A and SREC-I Are Kupffer and Endothelial Cell Receptors for Helper-dependent Adenoviral Vectors. <i>Molecular Therapy</i> , 2013, 21, 767-774.	8.2	51
121	Phenylbutyrate Therapy for Pyruvate Dehydrogenase Complex Deficiency and Lactic Acidosis. <i>Science Translational Medicine</i> , 2013, 5, 175ra31.	12.4	59
122	Heterogeneous clinical presentation in ICF syndrome: correlation with underlying gene defects. <i>European Journal of Human Genetics</i> , 2013, 21, 1219-1225.	2.8	115
123	RASA1 Mutations and Associated Phenotypes in 68 Families with Capillary Malformation-Arteriovenous Malformation. <i>Human Mutation</i> , 2013, 34, 1632-1641.	2.5	221
124	Gene transfer of master autophagy regulator TFEB results in clearance of toxic protein and correction of hepatic disease in alpha-1-antitrypsin deficiency. <i>EMBO Molecular Medicine</i> , 2013, 5, 397-412.	6.9	134
125	Transgene Expression up to 7 Years in Nonhuman Primates Following Hepatic Transduction with Helper-Dependent Adenoviral Vectors. <i>Human Gene Therapy</i> , 2013, 24, 761-765.	2.7	78
126	Autophagy master regulator TFEB induces clearance of toxic SERPINA1-1-antitrypsin polymers. <i>Autophagy</i> , 2013, 9, 1094-1096.	9.1	44

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127	Maternal vitamin K deficient embryopathy: Association with hyperemesis gravidarum and Crohn disease. American Journal of Medical Genetics, Part A, 2013, 161, 417-429.	1.2	21
128	Phenylbutyrate increases activity of pyruvate dehydrogenase complex. Oncotarget, 2013, 4, 804-805.	1.8	17
129	Reply to Amor et al. European Journal of Human Genetics, 2012, 20, 597-597.	2.8	4
130	Balloon Catheter Delivery of Helper-dependent Adenoviral Vector Results in Sustained, Therapeutic hFIX Expression in Rhesus Macaques. Molecular Therapy, 2012, 20, 1863-1870.	8.2	35
131	Low-Dose Amitriptyline-Induced Acute Dystonia in a Patient with Metachromatic Leukodystrophy. JIMD Reports, 2012, 9, 113-116.	1.5	3
132	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.	2.7	7
133	Supravalvular Aortic Stenosis. Circulation: Cardiovascular Genetics, 2012, 5, 692-696.	5.1	87
134	Autosomal Dominant MÃ©nÃ©trierÃ©like Disease. Journal of Pediatric Gastroenterology and Nutrition, 2012, 55, 717-720.	1.8	9
135	30-year follow-up of a patient with classic citrullinemia. Molecular Genetics and Metabolism, 2012, 106, 248-250.	1.1	10
136	Assessment of bone mineral status in children with Marfan syndrome. American Journal of Medical Genetics, Part A, 2012, 158A, 2221-2224.	1.2	26
137	<i>WDR35</i> mutation in siblings with Sensenbrenner syndrome: A ciliopathy with variable phenotype. American Journal of Medical Genetics, Part A, 2012, 158A, 2917-2924.	1.2	40
138	Guidelines for the use and interpretation of assays for monitoring autophagy. Autophagy, 2012, 8, 445-544.	9.1	3,122
139	DUF1220-Domain Copy Number Implicated in Human Brain-Size Pathology and Evolution. American Journal of Human Genetics, 2012, 91, 444-454.	6.2	113
140	Focal congenital lipoatrophy and vascular malformation: A mild form of inverse KlippelÃ©Trenaunay syndrome?. European Journal of Medical Genetics, 2012, 55, 705-707.	1.3	11
141	Immunodeficiency, centromeric instability, facial anomalies (ICF) syndrome, due to <i>ZBTB24</i> mutations, presenting with large cerebral cyst. American Journal of Medical Genetics, Part A, 2012, 158A, 2043-2046.	1.2	25
142	Nitric-Oxide Supplementation for Treatment of Long-Term Complications in Argininosuccinic Aciduria. American Journal of Human Genetics, 2012, 90, 836-846.	6.2	73
143	Dilation of the aortic root in mitochondrial disease patients. Molecular Genetics and Metabolism, 2011, 103, 167-170.	1.1	27
144	Chromosomal 17p13.3 microdeletion unmasking recessive Canavan disease mutation. Molecular Genetics and Metabolism, 2011, 104, 706-707.	1.1	3

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145	Cystic fibrosis: A disorder with defective autophagy. <i>Autophagy</i> , 2011, 7, 104-106.	9.1	75
146	Duplications of FOXP1 in 14q12 are associated with developmental epilepsy, mental retardation, and severe speech impairment. <i>European Journal of Human Genetics</i> , 2011, 19, 102-107.	2.8	104
147	Transcriptional gene network inference from a massive dataset elucidates transcriptome organization and gene function. <i>Nucleic Acids Research</i> , 2011, 39, 8677-8688.	14.5	102
148	Cutis laxa and fatal pulmonary hypertension. <i>Clinical Dysmorphology</i> , 2011, 20, 77-81.	0.3	2
149	Helper-dependent adenoviral vectors for liver-directed gene therapy. <i>Human Molecular Genetics</i> , 2011, 20, R7-R13.	2.9	71
150	Phenylbutyrate therapy for maple syrup urine disease. <i>Human Molecular Genetics</i> , 2011, 20, 631-640.	2.9	77
151	Correction of Hyperbilirubinemia in Gunn Rats Using Clinically Relevant Low Doses of Helper-Dependent Adenoviral Vectors. <i>Human Gene Therapy</i> , 2011, 22, 483-488.	2.7	16
152	Intrathecal Injection of Helper-Dependent Adenoviral Vectors Results in Long-Term Transgene Expression in Neuroependymal Cells and Neurons. <i>Human Gene Therapy</i> , 2011, 22, 745-751.	2.7	10
153	Copy number variants at Williams-Beuren syndrome 7q11.23 region. <i>Human Genetics</i> , 2010, 128, 3-26.	3.8	134
154	De novo terminal 22q12.3q13.3 duplication with pituitary hypoplasia (<i>Am J Med Genet Part A</i>) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 382	1.2	0
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