# Nicola Brunetti-Pierri

List of Publications by Year
in descending order

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Version: 2024-02-01

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|---|---|---|---|
| 218 papers | 16,655 citations | 50244 46 h-index | 16636 123 g-index |
| 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. Journal of Public Health, 2022, 44, 586-594. | 1.0 | 12 |
| 2 | Recurrent de novo missense variants in GNB2 can cause syndromic intellectual disability. Journal of Medical Genetics, 2022, 59, 511-516. | 1.5 | 4 |
| 3 | 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 |
| 4 | Epilepsy in KAT6A syndrome: Description of two individuals and revision of the literature. European Journal of Medical Genetics, 2022, 65, 104380. | 0.7 | 5 |
| 5 | Alpha-1 antitrypsin deficiency: A re-surfacing adult liver disorder. Journal of Hepatology, 2022, 76, 946-958. | 1.8 | 30 |
| 6 | 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 |
| 7 | Cholangiopathies and the noncoding revolution. Current Opinion in Gastroenterology, 2022, 38, 128-135. | 1.0 | 0 |
| 8 | 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 |
| 9 | De Novo ATP1A1 Variants in an Early-Onset Complex Neurodevelopmental Syndrome. Neurology, 2022, 98, 440-445. | 1.5 | 5 |
| 10 | 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 |
| 11 | Epigenetic Alterations in Inborn Errors of Immunity. Journal of Clinical Medicine, 2022, 11, 1261. | 1.0 | 8 |
| 12 | Disease burden and management of <scp>Criglerâ€Najjar</scp> syndrome: Report of a world registry. Liver International, 2022, 42, 1593-1604. | 1.9 | 8 |
| 13 | 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 |
| 14 | 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 |
| 15 | Liver-Directed Adeno-Associated Virusâ€“Mediated Gene Therapy for Mucopolysaccharidosis Type VI. , 2022, 1, . | | 5 |
| 16 | Liver gene therapy: The magic bullet for the sick lung. Molecular Therapy - Methods and Clinical Development, 2022, 26, 72-73. | 1.8 | 0 |
| 17 | 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 |
| 18 | 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 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | 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 |
| 20 | FBXO28 causes developmental and epileptic encephalopathy with profound intellectual disability. Epilepsia, 2021, 62, e13-e21. | 2.6 | 8 |
| 21 | 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 |
| 22 | Liver-directed gene-based therapies for inborn errors of metabolism. Expert Opinion on Biological Therapy, 2021, 21, 229-240. | 1.4 | 11 |
| 23 | 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 |
| 24 | 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 |
| 25 | Peculiar footprints in a child with agenesis of corpus callosum. Journal of Paediatrics and Child Health, 2021, 57, 450-451. | 0.4 | 0 |
| 26 | 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 |
| 27 | 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 |
| 28 | 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 |
| 29 | 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 |
| 30 | 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 |
| 31 | Mild Clinical Presentation of Joubert Syndrome in a Male Adult Carrying Biallelic MKS1 Truncating Variants. Diagnostics, 2021, 11, 1218. | 1.3 | 4 |
| 32 | 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 |
| 33 | Beclinâ€1â€mediated activation of autophagy improves proximal and distal urea cycle disorders. EMBO Molecular Medicine, 2021, 13, e13158. | 3.3 | 16 |
| 34 | A pilot clinical trial with losartan in Myhre syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 702-709. | 0.7 | 6 |
| 35 | RARS1â€related hypomyelinating leukodystrophy: Expanding the spectrum. Annals of Clinical and Translational Neurology, 2020, 7, 83-93. | 1.7 | 18 |
| 36 | Rubinsteinâ€“Taybi syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2020, 182, 2939-2950. | 0.7 | 16 |

| # | Article | IF | Citations |
|---|---|---|---|
| 37 | Disease characteristics of MCT8 deficiency: an international, retrospective, multicentre cohort study. Lancet Diabetes and Endocrinology,the, 2020, 8, 594-605. | 5.5 | 50 |
| 38 | 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 |
| 39 | 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 |
| 40 | 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 |
| 41 | Refinement of the clinical and mutational spectrum of <scp>UBE2A</scp> deficiency syndrome. Clinical Genetics, 2020, 98, 172-178. | 1.0 | 5 |
| 42 | Listen to Your Patients: A Diagnostic Clue. Journal of Pediatrics, 2020, 224, 171. | 0.9 | 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. Genetics in Medicine, 2020, 22, 1178-1180. | 1.1 | 16 |
| 44 | 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 |
| 45 | A systematic cross-sectional survey of multiple sulfatase deficiency. Molecular Genetics and Metabolism, 2020, 130, 283-288. | 0.5 | 10 |
| 46 | 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 |
| 47 | Expansion of the phenotype of lateral meningocele syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 1259-1262. | 0.7 | 9 |
| 48 | Expansion of the phenotypic spectrum of de novo missense variants in kinesin family member 1A () Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 | 1.1 | 16 |
| 49 | Two cases of 16q12.1q21 deletions and refinement of the critical region. European Journal of Medical Genetics, 2020, 63, 103878. | 0.7 | 3 |
| 50 | Cavitating and tigroidâ€like leukoencephalopathy in a case of NDUFA2â€related disorder. JIMD Reports, 2020, 52, 11-16. | 0.7 | 7 |
| 51 | A small 7q11.23 microduplication involving <scp>GTF2I</scp> in a family with intellectual disability. Clinical Genetics, 2020, 97, 940-942. | 1.0 | 4 |
| 52 | Large-scale targeted sequencing identifies risk genes for neurodevelopmental disorders. Nature Communications, 2020, 11, 4932. | 5.8 | 105 |
| 53 | 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 |
| 54 | 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 |

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|---|---|---|---|
| 55 | Paralog Studies Augment Gene Discovery: DDX and DHX Genes. American Journal of Human Genetics, 2019, 105, 302-316. | 2.6 | 56 |
| 56 | Sphingolipid Metabolism Perturbations in Rett Syndrome. Metabolites, 2019, 9, 221. | 1.3 | 12 |
| 57 | 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 |
| 58 | 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 |
| 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. | 1.4 | 22 |
| 60 | Geleophysic dysplasia: novel missense variants and insights into ADAMTSL2 intracellular trafficking. Molecular Genetics and Metabolism Reports, 2019, 21, 100504. | 0.4 | 10 |
| 61 | Ammonia and autophagy: An emerging relationship with implications for disorders with hyperammonemia. Journal of Inherited Metabolic Disease, 2019, 42, 1097-1104. | 1.7 | 20 |
| 62 | 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 |
| 63 | Progress and challenges in development of new therapies for urea cycle disorders. Human Molecular Genetics, 2019, 28, R42-R48. | 1.4 | 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. | 1.4 | 13 |
| 65 | Severe presentation and complex brain malformations in an individual carrying a CCND2 variant. Molecular Genetics & Genomic Medicine, 2019, 7, e708. | 0.6 | 7 |
| 66 | Retinal dystrophy in an individual carrying a de novo missense variant of SMARCA4. Molecular Genetics & Genomic Medicine, 2019, 7, e682. | 0.6 | 8 |
| 67 | Hepatic glutamine synthetase augmentation enhances ammonia detoxification. Journal of Inherited Metabolic Disease, 2019, 42, 1128-1135. | 1.7 | 7 |
| 68 | Pain and sleep disturbances in Rett syndrome and other neurodevelopmental disorders. Acta Paediatrica, International Journal of Paediatrics, 2019, 108, 171-172. | 0.7 | 4 |
| 69 | <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 |
| 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. | 0.5 | 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. | 0.7 | 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. | 3.3 | 39 |
| 74 | gene2drug: a computational tool for pathway-based rational drug repositioning. Bioinformatics, 2018, 34, 1498-1505. | 1.8 | 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. | 0.5 | 17 |
| 76 | Pyruvate dehydrogenase complex and lactate dehydrogenase are targets for therapy of acute liver failure. Journal of Hepatology, 2018, 69, 325-335. | 1.8 | 65 |
| 77 | Mutations in MAST1 Cause Mega-Corpus-Callosum Syndrome with Cerebellar Hypoplasia and Cortical Malformations. Neuron, 2018, 100, 1354-1368.e5. | 3.8 | 35 |
| 78 | Recent progress in gene therapies for mucopolysaccharidoses. Expert Opinion on Orphan Drugs, 2018, 6, 611-623. | 0.5 | 1 |
| 79 | Lowry-Wood syndrome: further evidence of association with RNU4ATAC, and correlation between genotype and phenotype. Human Genetics, 2018, 137, 905-909. | 1.8 | 11 |
| 80 | Induction of Nitric-Oxide Metabolism in Enterocytes Alleviates Colitis and Inflammation-Associated Colon Cancer. Cell Reports, 2018, 23, 1962-1976. | 2.9 | 51 |
| 81 | Targeting autophagy for therapy of hyperammonemia. Autophagy, 2018, 14, 1273-1275. | 4.3 | 15 |
| 82 | 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 |
| 83 | Reply. Hepatology, 2017, 66, 677-678. | 3.6 | 0 |
| 84 | Gene therapy with helper-dependent adenoviral vectors: lessons from studies in large animal models. Virus Genes, 2017, 53, 684-691. | 0.7 | 25 |
| 85 | 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 |
| 86 | 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 |
| 87 | An extremely severe phenotype attributed to WDR81 nonsense mutations. Annals of Neurology, 2017, 82, 650-651. | 2.8 | 11 |
| 88 | 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 |
| 89 | Expanding the phenotype of DSTâ€related disorder: A case report suggesting a genotype/phenotype correlation. American Journal of Medical Genetics, Part A, 2017, 173, 2743-2746. | 0.7 | 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. | 1.1 | 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. European Journal of Paediatric Neurology, 2017, 21, 902-906. | 0.7 | 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. Human Molecular Genetics, 2016, 26, ddw365. | 1.4 | 7 |
| 95 | 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 |
| 96 | 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 |
| 97 | Guidelines for the use and interpretation of assays for monitoring autophagy (3rd edition). Autophagy, 2016, 12, 1-222. | 4.3 | 4,701 |
| 98 | 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 |
| 99 | Helper-dependent adenoviral vectors for liver-directed gene therapy of primary hyperoxaluria type 1. Gene Therapy, 2016, 23, 129-134. | 2.3 | 37 |
| 100 | Progress toward improved therapies for inborn errors of metabolism. Human Molecular Genetics, 2016, 25, R27-R35. | 1.4 | 16 |
| 101 | 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 |
| 102 | Gene Therapy for Inherited Diseases of Liver Metabolism. Human Gene Therapy, 2015, 26, 186-192. | 1.4 | 10 |
| 103 | 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 |
| 104 | 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 |
| 105 | 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 |
| 106 | Helper-Dependent Adenoviral Vectors for Gene Therapy. , 2015, , 47-84. | | 1 |
| 107 | Challenges and Prospects for Helper-Dependent Adenoviral Vector-Mediated Gene Therapy. Biomedicines, 2014, 2, 132-148. | 1.4 | 9 |
| 108 | Retinal transduction profiles by high-capacity viral vectors. Gene Therapy, 2014, 21, 855-865. | 2.3 | 47 |

| # | Article | IF | Citations |
|---|---|---|---|
| 109 | Child Neurology: Recurrent rhabdomyolysis due to a fatty acid oxidation disorder. Neurology, 2014, 82, e1-4. | 1.5 | 8 |
| 110 | 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 |
| 111 | Giant breast tumors in a patient with Beckwithâ€“Wiedemann syndrome. American Journal of Medical Genetics, Part A, 2014, 164, 182-185. | 0.7 | 6 |
| 112 | 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 |
| 113 | SR-A and SREC-I binding peptides increase HDAd-mediated liver transduction. Gene Therapy, 2014, 21, 950-957. | 2.3 | 18 |
| 114 | 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 |
| 115 | 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 |
| 116 | Wilson Disease Protein ATP7B Utilizes Lysosomal Exocytosis to Maintain Copper Homeostasis. Developmental Cell, 2014, 29, 686-700. | 3.1 | 203 |
| 117 | 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 |
| 118 | Next-generation sequencing for disorders of low and high bone mineral density. Osteoporosis International, 2013, 24, 2253-2259. | 1.3 | 46 |
| 119 | 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 |
| 120 | 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 |
| 121 | Phenylbutyrate Therapy for Pyruvate Dehydrogenase Complex Deficiency and Lactic Acidosis. Science Translational Medicine, 2013, 5, 175ra31. | 5.8 | 59 |
| 122 | Heterogeneous clinical presentation in ICF syndrome: correlation with underlying gene defects. European Journal of Human Genetics, 2013, 21, 1219-1225. | 1.4 | 115 |
| 123 | RASA1Mutations and Associated Phenotypes in 68 Families with Capillary Malformation-Arteriovenous Malformation. Human Mutation, 2013, 34, 1632-1641. | 1.1 | 221 |
| 124 | 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 |
| 125 | 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 |
| 126 | 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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|---|---|---|---|
| 127 | 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 |
| 128 | Phenylbutyrate increases activity of pyruvate dehydrogenase complex. Oncotarget, 2013, 4, 804-805. | 0.8 | 17 |
| 129 | Reply to Amor et al. European Journal of Human Genetics, 2012, 20, 597-597. | 1.4 | 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. | 3.7 | 35 |
| 131 | Low-Dose Amitriptyline-Induced Acute Dystonia in a Patient with Metachromatic Leukodystrophy. JIMD Reports, 2012, 9, 113-116. | 0.7 | 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. | 1.4 | 7 |
| 133 | Supravalvular Aortic Stenosis. Circulation: Cardiovascular Genetics, 2012, 5, 692-696. | 5.1 | 87 |
| 134 | Autosomal Dominant MÃ©nÃ©trierâ€like Disease. Journal of Pediatric Gastroenterology and Nutrition, 2012, 55, 717-720. | 0.9 | 9 |
| 135 | 30-year follow-up of a patient with classic citrullinemia. Molecular Genetics and Metabolism, 2012, 106, 248-250. | 0.5 | 10 |
| 136 | Assessment of bone mineral status in children with Marfan syndrome. American Journal of Medical Genetics, Part A, 2012, 158A, 2221-2224. | 0.7 | 26 |
| 137 | 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 |
| 138 | Guidelines for the use and interpretation of assays for monitoring autophagy. Autophagy, 2012, 8, 445-544. | 4.3 | 3,122 |
| 139 | DUF1220-Domain Copy Number Implicated in Human Brain-Size Pathology and Evolution. American Journal of Human Genetics, 2012, 91, 444-454. | 2.6 | 113 |
| 140 | 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 |
| 141 | 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 |
| 142 | Nitric-Oxide Supplementation for Treatment of Long-Term Complications in Argininosuccinic Aciduria. American Journal of Human Genetics, 2012, 90, 836-846. | 2.6 | 73 |
| 143 | Dilation of the aortic root in mitochondrial disease patients. Molecular Genetics and Metabolism, 2011, 103, 167-170. | 0.5 | 27 |
| 144 | Chromosomal 17p13.3 microdeletion unmasking recessive Canavan disease mutation. Molecular Genetics and Metabolism, 2011, 104, 706-707. | 0.5 | 3 |

| # | Article | IF | Citations |
|---|---|---|---|
| 145 | Cystic fibrosis: A disorder with defective autophagy. Autophagy, 2011, 7, 104-106. | 4.3 | 75 |
| 146 | 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 |
| 147 | Transcriptional gene network inference from a massive dataset elucidates transcriptome organization and gene function. Nucleic Acids Research, 2011, 39, 8677-8688. | 6.5 | 102 |
| 148 | Cutis laxa and fatal pulmonary hypertension. Clinical Dysmorphology, 2011, 20, 77-81. | 0.1 | 2 |
| 149 | Helper-dependent adenoviral vectors for liver-directed gene therapy. Human Molecular Genetics, 2011, 20, R7-R13. | 1.4 | 71 |
| 150 | Phenylbutyrate therapy for maple syrup urine disease. Human Molecular Genetics, 2011, 20, 631-640. | 1.4 | 77 |
| 151 | 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 |
| 152 | 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 |
| 153 | Copy number variants at Williamsâ€“Beuren syndrome 7q11.23 region. Human Genetics, 2010, 128, 3-26. | 1.8 | 134 |
| 154 | De novo terminal 22q12.3q13.3 duplication with pituitary hypoplasia (Am J Med Genet Part A) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 382 | 0.7 | 0 |
| 155 | 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 |
| 156 | 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 |
| 157 | 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 |
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