# Joseph G Gleeson

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/2821046/publications.pdf

Version: 2024-02-01

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|---|---|---|---|
| 146 papers | 11,988 citations | 38742 50 h-index | 30922 102 g-index |
| 164 all docs | 164 docs citations | 164 times ranked | 20421 citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | doublecortin, a Brain-Specific Gene Mutated in Human X-Linked Lissencephaly and Double Cortex Syndrome, Encodes a Putative Signaling Protein. Cell, 1998, 92, 63-72. | 28.9 | 1,007 |
| 2 | De novo somatic mutations in components of the PI3K-AKT3-mTOR pathway cause hemimegalencephaly. Nature Genetics, 2012, 44, 941-945. | 21.4 | 628 |
| 3 | Aberrant methylation of t<scp>RNA</scp>s links cellular stress to neuroâ€developmental disorders. EMBO Journal, 2014, 33, 2020-2039. | 7.8 | 490 |
| 4 | Functional genomic screen for modulators of ciliogenesis and cilium length. Nature, 2010, 464, 1048-1051. | 27.8 | 473 |
| 5 | Exome Sequencing Links Corticospinal Motor Neuron Disease to Common Neurodegenerative Disorders. Science, 2014, 343, 506-511. | 12.6 | 466 |
| 6 | Lis1 and doublecortin function with dynein to mediate coupling of the nucleus to the centrosome in neuronal migration. Journal of Cell Biology, 2004, 165, 709-721. | 5.2 | 390 |
| 7 | Ndel1 Operates in a Common Pathway with LIS1 and Cytoplasmic Dynein to Regulate Cortical Neuronal Positioning. Neuron, 2004, 44, 263-277. | 8.1 | 334 |
| 8 | Characterization of Greater Middle Eastern genetic variation for enhanced disease gene discovery. Nature Genetics, 2016, 48, 1071-1076. | 21.4 | 314 |
| 9 | Mutations in BCKD-kinase Lead to a Potentially Treatable Form of Autism with Epilepsy. Science, 2012, 338, 394-397. | 12.6 | 272 |
| 10 | Impaired Amino Acid Transport at the Blood Brain Barrier Is a Cause of Autism Spectrum Disorder. Cell, 2016, 167, 1481-1494.e18. | 28.9 | 265 |
| 11 | The mutation significance cutoff: gene-level thresholds for variant predictions. Nature Methods, 2016, 13, 109-110. | 19.0 | 249 |
| 12 | Primary Cilia in the Developing and Mature Brain. Neuron, 2014, 82, 511-521. | 8.1 | 243 |
| 13 | Zika Virus Infects Neural Progenitors in the Adult Mouse Brain and Alters Proliferation. Cell Stem Cell, 2016, 19, 593-598. | 11.1 | 242 |
| 14 | CLP1 Founder Mutation Links tRNA Splicing and Maturation to Cerebellar Development and Neurodegeneration. Cell, 2014, 157, 651-663. | 28.9 | 228 |
| 15 | Exome Sequencing Can Improve Diagnosis and Alter Patient Management. Science Translational Medicine, 2012, 4, 138ra78. | 12.4 | 226 |
| 16 | Primary cilia in neurodevelopmental disorders. Nature Reviews Neurology, 2014, 10, 27-36. | 10.1 | 215 |
| 17 | An siRNA-based functional genomics screen for theÂidentification of regulators of ciliogenesis and ciliopathyÂgenes. Nature Cell Biology, 2015, 17, 1074-1087. | 10.3 | 215 |
| 18 | The human gene damage index as a gene-level approach to prioritizing exome variants. Proceedings of the National Academy of Sciences of the United States of America, 2015, 112, 13615-13620. | 7.1 | 213 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | Diagnosis of genetic diseases in seriously ill children by rapid whole-genome sequencing and automated phenotyping and interpretation. Science Translational Medicine, 2019, 11, . | 12.4 | 203 |
| 20 | Inactivating mutations in MFSD2A, required for omega-3 fatty acid transport in brain, cause a lethal microcephaly syndrome. Nature Genetics, 2015, 47, 809-813. | 21.4 | 180 |
| 21 | Alteration of Fatty-Acid-Metabolizing Enzymes Affects Mitochondrial Form and Function in Hereditary Spastic Paraplegia. American Journal of Human Genetics, 2012, 91, 1051-1064. | 6.2 | 179 |
| 22 | Paternally inherited cis-regulatory structural variants are associated with autism. Science, 2018, 360, 327-331. | 12.6 | 174 |
| 23 | CEP41 is mutated in Joubert syndrome and is required for tubulin glutamylation at the cilium. Nature Genetics, 2012, 44, 193-199. | 21.4 | 157 |
| 24 | Patient Mutations in Doublecortin Define a Repeated Tubulin-binding Domain. Journal of Biological Chemistry, 2000, 275, 34442-34450. | 3.4 | 138 |
| 25 | Biallelic mutations in SNX14 cause a syndromic form of cerebellar atrophy and lysosome-autophagosome dysfunction. Nature Genetics, 2015, 47, 528-534. | 21.4 | 111 |
| 26 | The Neurobiology of Zika Virus. Neuron, 2016, 92, 949-958. | 8.1 | 101 |
| 27 | Defining the phenotypic spectrum of SLC6A1 mutations. Epilepsia, 2018, 59, 389-402. | 5.1 | 99 |
| 28 | Mutations in LAMB1 Cause Cobblestone Brain Malformation without Muscular or Ocular Abnormalities. American Journal of Human Genetics, 2013, 92, 468-474. | 6.2 | 96 |
| 29 | Mutations in KATNB1 Cause Complex Cerebral Malformations by Disrupting Asymmetrically Dividing Neural Progenitors. Neuron, 2014, 84, 1226-1239. | 8.1 | 95 |
| 30 | AMPD2 Regulates GTP Synthesis and Is Mutated in a Potentially Treatable Neurodegenerative Brainstem Disorder. Cell, 2013, 154, 505-517. | 28.9 | 94 |
| 31 | A human three-dimensional neural-perivascular â€˜assembloidâ€™ promotes astrocytic development and enables modeling of SARS-CoV-2 neuropathology. Nature Medicine, 2021, 27, 1600-1606. | 30.7 | 94 |
| 32 | Analysis of 17 genes detects mutations in 81% of 811 patients with lissencephaly. Genetics in Medicine, 2018, 20, 1354-1364. | 2.4 | 92 |
| 33 | Cytosine-5 RNA methylation links protein synthesis to cell metabolism. PLoS Biology, 2019, 17, e3000297. | 5.6 | 87 |
| 34 | Loss of tubulin deglutamylase <scp>CCP</scp> 1 causes infantileâ€onset neurodegeneration. EMBO Journal, 2018, 37, . | 7.8 | 86 |
| 35 | NSUN2 introduces 5-methylcytosines in mammalian mitochondrial tRNAs. Nucleic Acids Research, 2019, 47, 8720-8733. | 14.5 | 84 |
| 36 | Off-Target Effect of doublecortin Family shRNA on Neuronal Migration Associated with Endogenous MicroRNA Dysregulation. Neuron, 2014, 82, 1255-1262. | 8.1 | 79 |

| # | Article | IF | Citations |
|---|---|---|---|
| 37 | Pathogenetic mechanisms of focal cortical dysplasia. Epilepsia, 2014, 55, 970-978. | 5.1 | 76 |
| 38 | Autism risk in offspring can be assessed through quantification of male sperm mosaicism. Nature Medicine, 2020, 26, 143-150. | 30.7 | 76 |
| 39 | Mutations in CSPP1 Lead to Classical Joubert Syndrome. American Journal of Human Genetics, 2014, 94, 80-86. | 6.2 | 75 |
| 40 | The landscape of somatic mutation in cerebral cortex of autistic and neurotypical individuals revealed by ultra-deep whole-genome sequencing. Nature Neuroscience, 2021, 24, 176-185. | 14.8 | 73 |
| 41 | Biallelic Mutations in Citron Kinase Link Mitotic Cytokinesis to Human Primary Microcephaly. American Journal of Human Genetics, 2016, 99, 501-510. | 6.2 | 70 |
| 42 | Biallelic loss of human CTNNA2, encoding Î±N-catenin, leads to ARP2/3 complex overactivity and disordered cortical neuronal migration. Nature Genetics, 2018, 50, 1093-1101. | 21.4 | 70 |
| 43 | Mutations in MBOAT7 , Encoding Lysophosphatidylinositol Acyltransferase I, Lead to Intellectual Disability Accompanied by Epilepsy and Autistic Features. American Journal of Human Genetics, 2016, 99, 912-916. | 6.2 | 69 |
| 44 | Loss of the sphingolipid desaturase DEGS1 causes hypomyelinating leukodystrophy. Journal of Clinical Investigation, 2019, 129, 1240-1256. | 8.2 | 68 |
| 45 | Biallelic mutations in the 3â€² exonuclease TOE1 cause pontocerebellar hypoplasia and uncover a role in snRNA processing. Nature Genetics, 2017, 49, 457-464. | 21.4 | 66 |
| 46 | A phenotypic spectrum of autism is attributable to the combined effects of rare variants, polygenic risk and sex. Nature Genetics, 2022, 54, 1284-1292. | 21.4 | 66 |
| 47 | Identification of a novel recessiveRELN mutation using a homozygous balanced reciprocal translocation. American Journal of Medical Genetics, Part A, 2007, 143A, 939-944. | 1.2 | 65 |
| 48 | Functional genome-wide siRNA screen identifies KIAA0586 as mutated in Joubert syndrome. ELife, 2015, 4, e06602. | 6.0 | 64 |
| 49 | Biallelic Truncating Mutations in FMN2, Encoding the Actin-Regulatory Protein Formin 2, Cause Nonsyndromic Autosomal-Recessive Intellectual Disability. American Journal of Human Genetics, 2014, 95, 721-728. | 6.2 | 62 |
| 50 | Ten new cases further delineate the syndromic intellectual disability phenotype caused by mutations in DYRK1A. European Journal of Human Genetics, 2015, 23, 1482-1487. | 2.8 | 62 |
| 51 | Homozygous mutation inNUP107leads to microcephaly with steroid-resistant nephrotic condition similar to Galloway-Mowat syndrome. Journal of Medical Genetics, 2017, 54, 399-403. | 3.2 | 62 |
| 52 | Biallelic Mutations in ADPRHL2, Encoding ADP-Ribosylhydrolase 3, Lead to a Degenerative Pediatric Stress-Induced Epileptic Ataxia Syndrome. American Journal of Human Genetics, 2018, 103, 431-439. | 6.2 | 62 |
| 53 | Redefining the Etiologic Landscape of Cerebellar Malformations. American Journal of Human Genetics, 2019, 105, 606-615. | 6.2 | 61 |
| 54 | Mutations inCEP120cause Joubert syndrome as well as complex ciliopathy phenotypes. Journal of Medical Genetics, 2016, 53, 608-615. | 3.2 | 55 |

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|---|---|---|---|
| 55 | Zika Virus Protease Cleavage of Host Protein Septin-2 Mediates Mitotic Defects in Neural Progenitors. Neuron, 2019, 101, 1089-1098.e4. | 8.1 | 55 |
| 56 | Biallelic Variants in OTUD6B Cause an Intellectual Disability Syndrome Associated with Seizures and Dysmorphic Features. American Journal of Human Genetics, 2017, 100, 676-688. | 6.2 | 54 |
| 57 | Biallelic variants in KIF14 cause intellectual disability with microcephaly. European Journal of Human Genetics, 2018, 26, 330-339. | 2.8 | 52 |
| 58 | Blacklisting variants common in private cohorts but not in public databases optimizes human exome analysis. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 950-959. | 7.1 | 52 |
| 59 | Mutations in GPAA1 , Encoding a GPI Transamidase Complex Protein, Cause Developmental Delay, Epilepsy, Cerebellar Atrophy, and Osteopenia. American Journal of Human Genetics, 2017, 101, 856-865. | 6.2 | 49 |
| 60 | Identification of a novel ARL13B variant in a Joubert syndrome-affected patient with retinal impairment and obesity. European Journal of Human Genetics, 2015, 23, 621-627. | 2.8 | 48 |
| 61 | DCLK1 phosphorylates the microtubuleâ€associated protein MAP7D1 to promote axon elongation in cortical neurons. Developmental Neurobiology, 2017, 77, 493-510. | 3.0 | 48 |
| 62 | Loss of SMPD4 Causes a Developmental Disorder Characterized by Microcephaly and Congenital Arthrogryposis. American Journal of Human Genetics, 2019, 105, 689-705. | 6.2 | 48 |
| 63 | Closing in on Mechanisms of Open Neural Tube Defects. Trends in Neurosciences, 2020, 43, 519-532. | 8.6 | 47 |
| 64 | Bi-allelic GOT2 Mutations Cause a Treatable Malate-Aspartate Shuttle-Related Encephalopathy. American Journal of Human Genetics, 2019, 105, 534-548. | 6.2 | 46 |
| 65 | Novel congenital disorder of O-linked glycosylation caused by GALNT2 loss of function. Brain, 2020, 143, 1114-1126. | 7.6 | 46 |
| 66 | Hypomorphic Recessive Variants in SUFU Impair the Sonic Hedgehog Pathway and Cause Joubert Syndrome with Cranio-facial and Skeletal Defects. American Journal of Human Genetics, 2017, 101, 552-563. | 6.2 | 45 |
| 67 | Autosomal-Recessive Mutations in the tRNA Splicing Endonuclease Subunit TSEN15 Cause Pontocerebellar Hypoplasia and Progressive Microcephaly. American Journal of Human Genetics, 2016, 99, 228-235. | 6.2 | 44 |
| 68 | Loss of NARS1 impairs progenitor proliferation in cortical brain organoids and leads to microcephaly. Nature Communications, 2020, 11, 4038. | 12.8 | 44 |
| 69 | Homozygous Mutations in TBC1D23 Lead to a Non-degenerative Form of Pontocerebellar Hypoplasia. American Journal of Human Genetics, 2017, 101, 441-450. | 6.2 | 43 |
| 70 | Somatic double-hit in MTOR and RPS6 in hemimegalencephaly with intractable epilepsy. Human Molecular Genetics, 2019, 28, 3755-3765. | 2.9 | 42 |
| 71 | Genetic variants in components of the NALCNâ€“UNC80â€“UNC79 ion channel complex cause a broad clinical phenotype (NALCN channelopathies). Human Genetics, 2018, 137, 753-768. | 3.8 | 38 |
| 72 | Mutations in UNC80, Encoding Part of the UNC79-UNC80-NALCN Channel Complex, Cause Autosomal-Recessive Severe Infantile Encephalopathy. American Journal of Human Genetics, 2016, 98, 210-215. | 6.2 | 37 |

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|---|---|---|---|
| 73 | A homozygous founder mutation in TRAPPC6B associates with a neurodevelopmental disorder characterised by microcephaly, epilepsy and autistic features. Journal of Medical Genetics, 2018, 55, 48-54. | 3.2 | 37 |
| 74 | PYCR2 Mutations cause a lethal syndrome of microcephaly and failure to thrive. Annals of Neurology, 2016, 80, 59-70. | 5.3 | 35 |
| 75 | Diencephalic-mesencephalic junction dysplasia: a novel recessive brain malformation. Brain, 2012, 135, 2416-2427. | 7.6 | 34 |
| 76 | Loss of Oxidation Resistance 1, OXR1, Is Associated with an Autosomal-Recessive Neurological Disease with Cerebellar Atrophy and Lysosomal Dysfunction. American Journal of Human Genetics, 2019, 105, 1237-1253. | 6.2 | 34 |
| 77 | Loss of the neural-specific BAF subunit ACTL6B relieves repression of early response genes and causes recessive autism. Proceedings of the National Academy of Sciences of the United States of America, 2020, 117, 10055-10066. | 7.1 | 34 |
| 78 | Molybdenum cofactor and isolated sulphite oxidase deficiencies: Clinical and molecular spectrum among Egyptian patients. European Journal of Paediatric Neurology, 2016, 20, 714-722. | 1.6 | 33 |
| 79 | Negative selection on human genes underlying inborn errors depends on disease outcome and both the mode and mechanism of inheritance. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, . | 7.1 | 33 |
| 80 | Non-manifesting AHI1 truncations indicate localized loss-of-function tolerance in a severe Mendelian disease gene. Human Molecular Genetics, 2015, 24, 2594-2603. | 2.9 | 32 |
| 81 | De Novo and Bi-allelic Pathogenic Variants in NARS1 Cause Neurodevelopmental Delay Due to Toxic Gain-of-Function and Partial Loss-of-Function Effects. American Journal of Human Genetics, 2020, 107, 311-324. | 6.2 | 32 |
| 82 | Mutation spectrum of Joubert syndrome and related disorders among Arabs. Human Genome Variation, 2014, 1, 14020. | 0.7 | 31 |
| 83 | Mutations in Spliceosomal Genes PPIL1 and PRP17 Cause Neurodegenerative Pontocerebellar Hypoplasia with Microcephaly. Neuron, 2021, 109, 241-256.e9. | 8.1 | 31 |
| 84 | Biallelic Mutations in TMTC3, Encoding a Transmembrane and TPR-Containing Protein, Lead to Cobblestone Lissencephaly. American Journal of Human Genetics, 2016, 99, 1181-1189. | 6.2 | 30 |
| 85 | Homozygous Missense Variants in NTNG2, Encoding a Presynaptic Netrin-G2 Adhesion Protein, Lead to a Distinct Neurodevelopmental Disorder. American Journal of Human Genetics, 2019, 105, 1048-1056. | 6.2 | 30 |
| 86 | Clinical and Genetic Aspects of the Segmental Overgrowth Spectrum DueÂto Somatic Mutations in PIK3CA. Journal of Pediatrics, 2015, 167, 957-962. | 1.8 | 29 |
| 87 | Defining the clinical, molecular and imaging spectrum of adaptor protein complex 4-associated hereditary spastic paraplegia. Brain, 2020, 143, 2929-2944. | 7.6 | 29 |
| 88 | Biallelic mutations in valyl-tRNA synthetase gene VARS are associated with a progressive neurodevelopmental epileptic encephalopathy. Nature Communications, 2019, 10, 707. | 12.8 | 28 |
| 89 | MINPP1 prevents intracellular accumulation of the chelator inositol hexakisphosphate and is mutated in Pontocerebellar Hypoplasia. Nature Communications, 2020, 11, 6087. | 12.8 | 28 |
| 90 | Loss of function mutations in GEMIN5 cause a neurodevelopmental disorder. Nature Communications, 2021, 12, 2558. | 12.8 | 28 |

| # | Article | IF | Citations |
|---|---|---|---|
| 91 | Developmental and temporal characteristics of clonal sperm mosaicism. Cell, 2021, 184, 4772-4783.e15. | 28.9 | 27 |
| 92 | Extending the mutation spectrum for Gallowayâ€“Mowat syndrome to include homozygous missense mutations in the WDR73 gene. American Journal of Medical Genetics, Part A, 2016, 170, 992-998. | 1.2 | 26 |
| 93 | Genome-wide screen identifies novel machineries required for both ciliogenesis and cell cycle arrest upon serum starvation. Biochimica Et Biophysica Acta - Molecular Cell Research, 2016, 1863, 1307-1318. | 4.1 | 26 |
| 94 | Comprehensive identification of somatic nucleotide variants in human brain tissue. Genome Biology, 2021, 22, 92. | 8.8 | 26 |
| 95 | Somatic mosaicism reveals clonal distributions of neocortical development. Nature, 2022, 604, 689-696. | 27.8 | 26 |
| 96 | Homozygous mutation of STXBP5L explains an autosomal recessive infantile-onset neurodegenerative disorder. Human Molecular Genetics, 2015, 24, 2000-2010. | 2.9 | 25 |
| 97 | MAB21L1 loss of function causes a syndromic neurodevelopmental disorder with distinctivecerebellar,ocular, craniofacial andgenital features (COFG) Tj ETQq1 1 0.784314 rgBT /Overlock 1 | 3.4 | 25 |
| 98 | Pathogenic ARH3 mutations result in ADP-ribose chromatin scars during DNA strand break repair. Nature Communications, 2020, 11, 3391. | 12.8 | 25 |
| 99 | Mutations in LNPK, Encoding the Endoplasmic Reticulum Junction Stabilizer Lunapark, Cause a Recessive Neurodevelopmental Syndrome. American Journal of Human Genetics, 2018, 103, 296-304. | 6.2 | 24 |
| 100 | Bi-allelic Mutations in FAM149B1 Cause Abnormal Primary Cilium and a Range of Ciliopathy Phenotypes in Humans. American Journal of Human Genetics, 2019, 104, 731-737. | 6.2 | 23 |
| 101 | A Human Pleiotropic Multiorgan Condition Caused by Deficient Wnt Secretion. New England Journal of Medicine, 2021, 385, 1292-1301. | 27.0 | 23 |
| 102 | Pyruvate dehydrogenase complex-E2 deficiency causes paroxysmal exercise-induced dyskinesia. Neurology, 2017, 89, 2297-2298. | 1.1 | 22 |
| 103 | Bi-allelic Variants in the GPI Transamidase Subunit PIGK Cause a Neurodevelopmental Syndrome with Hypotonia, Cerebellar Atrophy, and Epilepsy. American Journal of Human Genetics, 2020, 106, 484-495. | 6.2 | 22 |
| 104 | Biallelic loss of function variants in <scp>SYT2</scp> cause a treatable congenital onset presynaptic myasthenic syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 2272-2283. | 1.2 | 20 |
| 105 | A Zika virus mutation enhances transmission potential and confers escape from protective dengue virus immunity. Cell Reports, 2022, 39, 110655. | 6.4 | 20 |
| 106 | Loss of Protocadherinâ€12<scp>L</scp>eads to <scp>D</scp>iencephalicâ€<scp>M</scp>esencephalic <scp>J</scp>unction <scp>D</scp>ysplasia <scp>S</scp>yndrome. Annals of Neurology, 2018, 84, 638-647. | 5.3 | 19 |
| 107 | Identification of a homozygous nonsense mutation in KIAA0556 in a consanguineous family displaying Joubert syndrome. Human Genetics, 2016, 135, 919-921. | 3.8 | 18 |
| 108 | Bi-allelic Loss of Human APC2, Encoding Adenomatous Polyposis Coli Protein 2, Leads to Lissencephaly, Subcortical Heterotopia, and Global Developmental Delay. American Journal of Human Genetics, 2019, 105, 844-853. | 6.2 | 17 |

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|---|---|---|---|
| 109 | Biallelic variants in HPDL, encoding 4-hydroxyphenylpyruvate dioxygenase-like protein, lead to an infantile neurodegenerative condition. Genetics in Medicine, 2021, 23, 524-533. | 2.4 | 17 |
| 110 | Alternative genomic diagnoses for individuals with a clinical diagnosis of Dubowitz syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 119-133. | 1.2 | 17 |
| 111 | Biallelic variants in SLC38A3 encoding a glutamine transporter cause epileptic encephalopathy. Brain, 2022, 145, 909-924. | 7.6 | 17 |
| 112 | UBR7 functions with UBR5 in the Notch signaling pathway and is involved in a neurodevelopmental syndrome with epilepsy, ptosis, and hypothyroidism. American Journal of Human Genetics, 2021, 108, 134-147. | 6.2 | 15 |
| 113 | Polo-like kinase 2 regulates angiogenic sprouting and blood vessel development. Developmental Biology, 2015, 404, 49-60. | 2.0 | 14 |
| 114 | When size matters: CHD8 in autism. Nature Neuroscience, 2016, 19, 1430-1432. | 14.8 | 14 |
| 115 | Regulation of human cerebral cortical development by EXOC7 and EXOC8, components of the exocyst complex, and roles in neural progenitor cell proliferation and survival. Genetics in Medicine, 2020, 22, 1040-1050. | 2.4 | 13 |
| 116 | Implication of folate deficiency in CYP2U1 loss of function. Journal of Experimental Medicine, 2021, 218, . | 8.5 | 13 |
| 117 | Sperm mosaicism: implications for genomic diversity and disease. Trends in Genetics, 2021, 37, 890-902. | 6.7 | 13 |
| 118 | Agenesis of the putamen and globus pallidus caused by recessive mutations in the homeobox gene GSX2. Brain, 2019, 142, 2965-2978. | 7.6 | 12 |
| 119 | Uner Tan syndrome caused by a homozygousTUBB2Bmutation affecting microtubule stability. Human Molecular Genetics, 2016, 26, ddw383. | 2.9 | 11 |
| 120 | Early life experience shapes neural genome. Science, 2018, 359, 1330-1331. | 12.6 | 11 |
| 121 | Expanding the phenotype of PIGSâ€associated early onset epileptic developmental encephalopathy. Epilepsia, 2021, 62, e35-e41. | 5.1 | 11 |
| 122 | Biallelic variants in KARS1 are associated with neurodevelopmental disorders and hearing loss recapitulated by the knockout zebrafish. Genetics in Medicine, 2021, 23, 1933-1943. | 2.4 | 11 |
| 123 | Loss of C2orf69 defines a fatal autoinflammatory syndrome in humans and zebrafish that evokes a glycogen-storage-associated mitochondriopathy. American Journal of Human Genetics, 2021, 108, 1301-1317. | 6.2 | 11 |
| 124 | Clinical Pertinence Metric Enables Hypothesis-Independent Genome-Phenome Analysis for Neurologic Diagnosis. Journal of Child Neurology, 2015, 30, 881-888. | 1.4 | 10 |
| 125 | Insight into developmental mechanisms of global and focal migration disorders of cortical development. Current Opinion in Neurobiology, 2021, 66, 77-84. | 4.2 | 9 |
| 126 | ABHD16A deficiency causes a complicated form of hereditary spastic paraplegia associated with intellectual disability and cerebral anomalies. American Journal of Human Genetics, 2021, 108, 2017-2023. | 6.2 | 9 |

| # | Article | IF | Citations |
|---|---|---|---|
| 127 | Biallelic FRA10AC1 variants cause a neurodevelopmental disorder with growth retardation. Brain, 2022, 145, 1551-1563. | 7.6 | 9 |
| 128 | Clinicoâ€radiological features, molecular spectrum, and identification of prognostic factors in developmental and epileptic encephalopathy due to inosine triphosphate pyrophosphatase (ITPase) deficiency. Human Mutation, 2022, 43, 403-419. | 2.5 | 9 |
| 129 | Oligonucleotide correction of an intronic TIMMDC1 variant in cells of patients with severe neurodegenerative disorder. Npj Genomic Medicine, 2022, 7, 9. | 3.8 | 8 |
| 130 | <scp>Elâ€Hattabâ€Alkuraya</scp> syndrome caused by biallelic <scp>WDR45B</scp> pathogenic variants: Further delineation of the phenotype and genotype. Clinical Genetics, 2022, 101, 530-540. | 2.0 | 7 |
| 131 | Dandyâ€“Walker malformation, genitourinary abnormalities, and intellectual disability in two families. American Journal of Medical Genetics, Part A, 2015, 167, 2503-2507. | 1.2 | 6 |
| 132 | Recurrent homozygous damaging mutation in TMX2, encoding a protein disulfide isomerase, in four families with microlissencephaly. Journal of Medical Genetics, 2020, 57, 274-282. | 3.2 | 6 |
| 133 | RSRC1 loss-of-function variants cause mild to moderate autosomal recessive intellectual disability. Brain, 2020, 143, e31-e31. | 7.6 | 6 |
| 134 | Monoallelic and biallelic mutations inRELNunderlie a graded series of neurodevelopmental disorders. Brain, 2022, 145, 3274-3287. | 7.6 | 6 |
| 135 | Novel mutation in the fukutin gene in an Egyptian family with Fukuyama congenital muscular dystrophy and microcephaly. Gene, 2014, 539, 279-282. | 2.2 | 5 |
| 136 | Unbiased mosaic variant assessment in sperm: a cohort study to test predictability of transmission. ELife, 0, 11, . | 6.0 | 5 |
| 137 | The ciliary proteins Meckelin and Jouberin are required for retinoic acid-dependent neural differentiation of mouse embryonic stem cells. Differentiation, 2014, 87, 134-146. | 1.9 | 4 |
| 138 | Clinical, biomarker and genetic spectrum of Niemannâ€Pick type C in Egypt: The detection of nine novel NPC1 mutations. Clinical Genetics, 2019, 95, 537-539. | 2.0 | 4 |
| 139 | Molecular diagnosis in recessive pediatric neurogenetic disease can help reduce disease recurrence in families. BMC Medical Genomics, 2020, 13, 68. | 1.5 | 4 |
| 140 | The Neurobiology of Modern Viral Scourges: ZIKV and COVID-19. Neuroscientist, 2022, 28, 438-452. | 3.5 | 4 |
| 141 | Bi-allelic TTC5 variants cause delayed developmental milestones and intellectual disability. Journal of Medical Genetics, 2021, 58, 237-246. | 3.2 | 4 |
| 142 | Inhibition of G-protein signalling in cardiac dysfunction of intellectual developmental disorder with cardiac arrhythmia (IDDCA) syndrome. Journal of Medical Genetics, 2021, 58, 815-831. | 3.2 | 3 |
| 143 | Biallelic BICD2 variant is a novel candidate for Cohen-like syndrome. Journal of Human Genetics, 2022, 67, 553-556. | 2.3 | 3 |
| 144 | A founder mutation in PEX12 among Egyptian patients in peroxisomal biogenesis disorder. Neurological Sciences, 2020, 42, 2737-2745. | 1.9 | 1 |

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|---|---|---|---|
| 145 | Primary Cilia and Brain Wiring, Connecting the Dots. Developmental Cell, 2019, 51, 661-663. | 7.0 | 0 |
| 146 | Editorial overview: Neurodevelopment Diseases and Neurogenetics pivot towards mechanisms and therapies. Current Opinion in Genetics and Development, 2020, 65, iii-vii. | 3.3 | 0 |