# Angela M Kaindl

List of Publications by Year
in descending order



| | | | |
|---|---|---|---|
| 139 papers | 5,333 citations | 101543 36 h-index | 95266 68 g-index |
| 147 all docs | 147 docs citations | 147 times ranked | 9065 citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | Homozygous mutation in MCM7 causes autosomal recessive primary microcephaly and intellectual disability. Journal of Medical Genetics, 2022, 59, 453-461. | 3.2 | 5 |
| 2 | In-depth analysis reveals complex molecular aetiology in a cohort of idiopathic cerebral palsy. Brain, 2022, 145, 119-141. | 7.6 | 28 |
| 3 | Cerebral Abnormalities in Spina Bifida: A Neuropathological Study. Pediatric and Developmental Pathology, 2022, 25, 107-123. | 1.0 | 3 |
| 4 | Gene replacement therapy with onasemnogene abeparvovec in children with spinal muscular atrophy aged 24 months or younger and bodyweight up to 15 kg: an observational cohort study. The Lancet Child and Adolescent Health, 2022, 6, 17-27. | 5.6 | 57 |
| 5 | Î²-Ureidopropionase deficiency due to novel and rare UPB1 mutations affecting pre-mRNA splicing and protein structural integrity and catalytic activity. Molecular Genetics and Metabolism, 2022, 136, 177-185. | 1.1 | 3 |
| 6 | Is cannabidiol worth a trial in Rasmussen encephalitis?. European Journal of Paediatric Neurology, 2022, 37, 53-55. | 1.6 | 3 |
| 7 | Cross-sectional Neuromuscular Phenotyping Study of Patients With Arhinia With SMCHD1 Variants. Neurology, 2022, 98, . | 1.1 | 3 |
| 8 | Standard values for MRI brain biometry throughout the first year of life. Pediatrics and Neonatology, 2022, 63, 255-261. | 0.9 | 1 |
| 9 | Relationship between cerebral palsy severity and cognition, aids and education. Minerva Pediatrics, 2022, , . | 0.4 | 0 |
| 10 | Epilepsy surgery in the first six months of life: A systematic review and meta-analysis. Seizure: the Journal of the British Epilepsy Association, 2022, 96, 109-117. | 2.0 | 12 |
| 11 | Modified Zipper Method, a Promising Treatment Option in Severe Pediatric Immune-Mediated Neurologic Disorders. Journal of Child Neurology, 2022, 37, 505-516. | 1.4 | 3 |
| 12 | Selenium Status in Paediatric Patients with Neurodevelopmental Diseases. Nutrients, 2022, 14, 2375. | 4.1 | 2 |
| 13 | Interferon receptor dysfunction in a child with malignant atrophic papulosis and CNS involvement. Lancet Neurology, The, 2022, 21, 682-686. | 10.2 | 2 |
| 14 | Abnormal brain structure and behavior in MyD88-deficient mice. Brain, Behavior, and Immunity, 2021, 91, 181-193. | 4.1 | 14 |
| 15 | Brain malformations and cognitive performance in spina bifida. Developmental Medicine and Child Neurology, 2021, 63, 295-302. | 2.1 | 9 |
| 16 | SIGLEC1 (CD169) as a potential diagnostical screening marker for monogenic interferonopathies. Pediatric Allergy and Immunology, 2021, 32, 621-625. | 2.6 | 15 |
| 17 | Systematic Classification of Spina Bifida. Journal of Neuropathology and Experimental Neurology, 2021, 80, 294-305. | 1.7 | 6 |
| 18 | Lumbar Puncture Opening Pressure in Patients with Spinal Muscular Atrophy. Neuropediatrics, 2021, 52, 219-223. | 0.6 | 4 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | Novel Mutation in the TSFM Gene Causes an Earlyâ€Onset Complex Chorea without Basal Ganglia Lesions. Movement Disorders Clinical Practice, 2021, 8, 453-455. | 1.5 | 1 |
| 20 | Zebrafish modeling mimics developmental phenotype of patients with <scp>RAPGEF1</scp> mutation. Clinical Genetics, 2021, 100, 144-155. | 2.0 | 1 |
| 21 | Arhgef2 regulates neural differentiation in the cerebral cortex through mRNA m6A-methylation of Npdc1 and Cend1. IScience, 2021, 24, 102645. | 4.1 | 4 |
| 22 | Assessment of myelination in infants and young children by T1 relaxation time measurements using the magnetization-prepared 2 rapid acquisition gradient echoes sequence. Pediatric Radiology, 2021, 51, 2058-2068. | 2.0 | 9 |
| 23 | Infratentorial MRI Findings in Rasmussen Encephalitis Suggest Primary Cerebellar Involvement. Neurology: Neuroimmunology and NeuroInflammation, 2021, 8, . | 6.0 | 4 |
| 24 | Maintenance of Elective Patient Care at Berlin University Children's Hospital During the COVID-19 Pandemic. Frontiers in Pediatrics, 2021, 9, 694963. | 1.9 | 1 |
| 25 | Motor and functional outcome of selective dorsal rhizotomy in children with spastic diplegia at 12 and 24Âmonths of follow-up. Acta Neurochirurgica, 2021, 163, 2837-2844. | 1.7 | 5 |
| 26 | Lessons learned from drug trials in neurofibromatosis: A systematic review. European Journal of Medical Genetics, 2021, 64, 104281. | 1.3 | 5 |
| 27 | OUP accepted manuscript. Human Molecular Genetics, 2021, 30, 2068-2081. | 2.9 | 7 |
| 28 | Advantages of Botulinum Toxin A Treatment in Combination with Controlled Dynamic Stretching Orthotics for the Treatment of Contractures. , 2021, 52, . | | 0 |
| 29 | First Report of Glioblastoma and Associated PNKP Mutation. Neuropediatrics, 2021, 52, . | 0.6 | 0 |
| 30 | Expanding the Phenotype of NUP85 Mutations beyond Nephrotic Syndrome to Primary Autosomal Recessive Microcephaly and Seckel Syndrome Spectrum Disorders. , 2021, 52, . | | 1 |
| 31 | Case Report: Behavioral Disorder Following Hemispherotomy: A Valproate Effect?. Frontiers in Neurology, 2021, 12, 764376. | 2.4 | 1 |
| 32 | Beware of Neurotoxins in Common Plants: Water Hemlock Poisoning Presenting as Convulsive Status Epilepticus. Pediatric Neurology, 2021, 127, 39-40. | 2.1 | 0 |
| 33 | Modified Zipper Method: A Promising Treatment Option in Severe Pediatric Cases of Immune-Mediated Neurological Disorders. Neuropediatrics, 2021, 52, . | 0.6 | 0 |
| 34 | Pilot Study to Observe Changes in Independence in Everyday Life through Intensive Wheelchair Mobility Training in Wheelchair-Dependent Children. Neuropediatrics, 2021, 52, . | 0.6 | 0 |
| 35 | Autosomal Recessive Primary Microcephaly: Not Just a Small Brain. Frontiers in Cell and Developmental Biology, 2021, 9, 784700. | 3.7 | 28 |
| 36 | Case Report: Hemispherotomy in the First Days of Life to Treat Drug-Resistant Lesional Epilepsy. Frontiers in Neurology, 2021, 12, 818972. | 2.4 | 6 |

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|---|---|---|---|
| 37 | Encephalitis patient-derived monoclonal GABAA receptor antibodies cause epileptic seizures. Journal of Experimental Medicine, 2021, 218, . | 8.5 | 19 |
| 38 | Congenital microcephalyâ€linked CDK5RAP2 affects eye development. Annals of Human Genetics, 2020, 84, 87-91. | 0.8 | 4 |
| 39 | Just Expect It: Compound Heterozygous Variants of POMT1 in a Consanguineous Familyâ€"The Role of Next Generation Sequencing in Neuromuscular Disorders. Neuropediatrics, 2020, 51, 072-075. | 0.6 | 1 |
| 40 | Age-specific occurrence of pathological fractures in patients with spina bifida. European Journal of Pediatrics, 2020, 179, 773-779. | 2.7 | 4 |
| 41 | MN1 C-terminal truncation syndrome is a novel neurodevelopmental and craniofacial disorder with partial rhombencephalosynapsis. Brain, 2020, 143, 55-68. | 7.6 | 38 |
| 42 | Early Onset, Long Illness Duration, Epilepsy Type, and Polypharmacy Have an Adverse Effect on Psychosocial Outcome in Children with Epilepsy. Neuropediatrics, 2020, 51, 164-169. | 0.6 | 6 |
| 43 | Presence of anti-neuronal antibodies in children with neurological disorders beyond encephalitis. European Journal of Paediatric Neurology, 2020, 28, 159-166. | 1.6 | 4 |
| 44 | Immunofluorescence Staining of Paraffin Sections Step by Step. Frontiers in Neuroanatomy, 2020, 14, 582218. | 1.7 | 77 |
| 45 | Clinical Outcome of Children With Corpus Callosum Agenesis. Pediatric Neurology, 2020, 112, 47-52. | 2.1 | 7 |
| 46 | The clinical-phenotype continuum in DYNC1H1-related disordersâ€"genomic profiling and proposal for a novel classification. Journal of Human Genetics, 2020, 65, 1003-1017. | 2.3 | 30 |
| 47 | Intravenous Nimodipine Treatment for Severe Episode of ATP1A2 Hemiplegic Migraine. Pediatric Neurology, 2020, 112, 71-72. | 2.1 | 6 |
| 48 | The clinical, histologic, and genotypic spectrum of SEPN1 -related myopathy. Neurology, 2020, 95, e1512-e1527. | 1.1 | 44 |
| 49 | Insight Into the Ontogeny of GnRH Neurons From Patients Born Without a Nose. Journal of Clinical Endocrinology and Metabolism, 2020, 105, 1538-1551. | 3.6 | 7 |
| 50 | Clinical and Magnetic Resonance Imaging Outcome Predictors in Pediatric <scp>Antiâ€"Nâ€Methylâ€Dâ€Aspartate</scp> Receptor Encephalitis. Annals of Neurology, 2020, 88, 148-159. | 5.3 | 26 |
| 51 | Fulminant cerebral venous thrombosis associated with the m.3243A>G MELAS mutation: A new guise for an old disease. Brain and Development, 2019, 41, 901-904. | 1.1 | 1 |
| 52 | A missense mutation in SNRPE linked to non-syndromal microcephaly interferes with U snRNP assembly and pre-mRNA splicing. PLoS Genetics, 2019, 15, e1008460. | 3.5 | 18 |
| 53 | PEDIA: prioritization of exome data by image analysis. Genetics in Medicine, 2019, 21, 2807-2814. | 2.4 | 58 |
| 54 | Structural brain anomalies in patients with <scp>FOXG</scp>1 syndrome and in Foxg1+/â^' mice. Annals of Clinical and Translational Neurology, 2019, 6, 655-668. | 3.7 | 19 |

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|---|---|---|---|
| 55 | Altered inhibition and excitation in neocortical circuits in congenital microcephaly. Neurobiology of Disease, 2019, 129, 130-143. | 4.4 | 7 |
| 56 | Mutations in the tRNA methyltransferase 1 gene TRMT1 cause congenital microcephaly, isolated inferior vermian hypoplasia and cystic leukomalacia in addition to intellectual disability. American Journal of Medical Genetics, Part A, 2018, 176, 2517-2521. | 1.2 | 15 |
| 57 | Subtelomeric methylation distinguishes between subtypes of Immunodeficiency, Centromeric instability and Facial anomalies syndrome. Human Molecular Genetics, 2018, 27, 3568-3581. | 2.9 | 26 |
| 58 | P 306. Role of Cdk5rap2 in Neocortical Development. , 2018, 49, . | | 0 |
| 59 | FV 757. Clinical, Radiological and Genetic Spectrum of a Large Pediatric Cohort with Epilepsy. Neuropediatrics, 2018, 49, . | 0.6 | 0 |
| 60 | SMCHD1 mutations associated with a rare muscular dystrophy can also cause isolated arhinia and Bosma arhinia microphthalmia syndrome. Nature Genetics, 2017, 49, 238-248. | 21.4 | 131 |
| 61 | CDK5RAP2 Is Required to Maintain the Germ Cell Pool during Embryonic Development. Stem Cell Reports, 2017, 8, 198-204. | 4.8 | 17 |
| 62 | Pontine Tegmental Cap Dysplasia in an Extremely Preterm Infant and Review of the Literature. Journal of Child Neurology, 2017, 32, 334-340. | 1.4 | 7 |
| 63 | Autosomal Recessive Primary Microcephaly (MCPH): An Update. Neuropediatrics, 2017, 48, 135-142. | 0.6 | 70 |
| 64 | Lacosamide Lowers Valproate and Levetiracetam Levels. Neuropediatrics, 2017, 48, 188-189. | 0.6 | 6 |
| 65 | Identification of a novel homozygous TRAPPC9 gene mutation causing nonâ€syndromic intellectual disability, speech disorder, and secondary microcephaly. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2017, 174, 839-845. | 1.7 | 28 |
| 66 | PTRH2 gene mutation causes progressive congenital skeletal muscle pathology. Human Molecular Genetics, 2017, 26, 1458-1464. | 2.9 | 10 |
| 67 | Recessive mutation in EXOSC3 associates with mitochondrial dysfunction and pontocerebellar hypoplasia. Mitochondrion, 2017, 37, 46-54. | 3.4 | 26 |
| 68 | Homozygous ARHGEF2 mutation causes intellectual disability and midbrain-hindbrain malformation. PLoS Genetics, 2017, 13, e1006746. | 3.5 | 27 |
| 69 | Golgi-Cox Staining Step by Step. Frontiers in Neuroanatomy, 2016, 10, 38. | 1.7 | 133 |
| 70 | Genetic causes of MCPH in consanguineous Pakistani families. Clinical Genetics, 2016, 89, 744-745. | 2.0 | 6 |
| 71 | Acute Disseminated Encephalomyelitis After Human Parechovirus Infection. Pediatric Infectious Disease Journal, 2016, 35, 35-38. | 2.0 | 12 |
| 72 | Phenotype variability of infantile-onset multisystem neurologic, endocrine, and pancreatic disease IMNEPD. Orphanet Journal of Rare Diseases, 2016, 11, 52. | 2.7 | 13 |

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|---|---|---|---|
| 73 | Homozygous YME1L1 mutation causes mitochondriopathy with optic atrophy and mitochondrial network fragmentation. ELife, 2016, 5, . | 6.0 | 88 |
| 74 | Phenotype of five patients with dopa-responsive dystonia and mutations in GCH1. Journal of Pediatric Neurology, 2015, 03, 083-087. | 0.2 | 1 |
| 75 | Neutrophil oxidative burst activates ATM to regulate cytokine production and apoptosis. Blood, 2015, 126, 2842-2851. | 1.4 | 58 |
| 76 | Loss of CDK5RAP2 affects neural but not non-neural mESC differentiation into cardiomyocytes. Cell Cycle, 2015, 14, 2044-2057. | 2.6 | 15 |
| 77 | Synaptic NMDA receptor activity is coupled to the transcriptional control of the glutathione system. Nature Communications, 2015, 6, 6761. | 12.8 | 119 |
| 78 | Redefining the MED13L syndrome. European Journal of Human Genetics, 2015, 23, 1308-1317. | 2.8 | 53 |
| 79 | What next-generation sequencing (NGS) technology has enabled us to learn about primary autosomal recessive microcephaly (MCPH). Molecular and Cellular Probes, 2015, 29, 271-281. | 2.1 | 64 |
| 80 | Novel Alternative Splice Variants of Mouse Cdk5rap2. PLoS ONE, 2015, 10, e0136684. | 2.5 | 4 |
| 81 | Mutations in PTRH2 cause novel infantileâ€onset multisystem disease with intellectual disability, microcephaly, progressive ataxia, and muscle weakness. Annals of Clinical and Translational Neurology, 2014, 1, 1024-1035. | 3.7 | 29 |
| 82 | Large homozygous RAB3GAP1 gene microdeletion causes Warburg Micro Syndrome 1. Orphanet Journal of Rare Diseases, 2014, 9, 113. | 2.7 | 5 |
| 83 | Combined immunodeficiency develops with age in Immunodeficiency-centromeric instability-facial anomalies syndrome 2 (ICF2). Orphanet Journal of Rare Diseases, 2014, 9, 116. | 2.7 | 34 |
| 84 | Previously reported new type of autosomal recessive primary microcephaly is caused by compound heterozygousASPMgene mutations. Cell Cycle, 2014, 13, 1650-1651. | 2.6 | 8 |
| 85 | Diagnostic approach to microcephaly in childhood: a twoâ€center study and review of the literature. Developmental Medicine and Child Neurology, 2014, 56, 732-741. | 2.1 | 176 |
| 86 | Growth and psychomotor development of patients with Duchenne muscular dystrophy. European Journal of Paediatric Neurology, 2014, 18, 38-44. | 1.6 | 35 |
| 87 | Interstitial 12p deletion involving more than 40 genes in a patient with postnatal microcephaly, psychomotor delay, optic nerve atrophy, and facial dysmorphism. Meta Gene, 2014, 2, 72-82. | 0.6 | 4 |
| 88 | Autosomal recessive primary microcephalies (MCPH). European Journal of Paediatric Neurology, 2014, 18, 547-548. | 1.6 | 22 |
| 89 | Is microcephaly a so-far unrecognized feature of XYY syndrome?. Meta Gene, 2014, 2, 160-163. | 0.6 | 2 |
| 90 | Clinical and cellular features in patients with primary autosomal recessive microcephaly and a novel CDK5RAP2 mutation. Orphanet Journal of Rare Diseases, 2013, 8, 59. | 2.7 | 27 |

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|---|---|---|---|
| 91 | Microcephaly. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2013, 111, 129-141. | 1.8 | 94 |
| 92 | Abnormal centrosome and spindle morphology in a patient with autosomal recessive primary microcephaly type 2 due to compound heterozygous WDR62 gene mutation. Orphanet Journal of Rare Diseases, 2013, 8, 178. | 2.7 | 38 |
| 93 | Combined deletion 18q22.2 and duplication/triplication 18q22.1 causes microcephaly, mental retardation and leukencephalopathy. Gene, 2013, 523, 92-98. | 2.2 | 4 |
| 94 | CDK5RAP2 Expression During Murine and Human Brain Development Correlates with Pathology in Primary Autosomal Recessive Microcephaly. Cerebral Cortex, 2013, 23, 2245-2260. | 2.9 | 30 |
| 95 | G proteinâ€“coupled receptor kinase 2 and group I metabotropic glutamate receptors mediate inflammationâ€induced sensitization to excitotoxic neurodegeneration. Annals of Neurology, 2013, 73, 667-678. | 5.3 | 44 |
| 96 | Angelman syndrome and severe infections in a patient with de novo 15q11.2â€“q13.1 deletion and maternally inherited 2q21.3 microdeletion. Gene, 2013, 512, 453-455. | 2.2 | 11 |
| 97 | Reference genes in the developing murine brain and in differentiating embryonic stem cells. Neurological Research, 2012, 34, 664-668. | 1.3 | 14 |
| 98 | Activation of microglial Nâ€methylâ€Dâ€aspartate receptors triggers inflammation and neuronal cell death in the developing and mature brain. Annals of Neurology, 2012, 72, 536-549. | 5.3 | 194 |
| 99 | Neuroprotective Strategies. , 2012, , 1173-1179. | | 0 |
| 100 | Implanted Neurosphere-Derived Precursors Promote Recovery After Neonatal Excitotoxic Brain Injury. Stem Cells and Development, 2011, 20, 865-879. | 2.1 | 28 |
| 101 | Neuronal Death and Oxidative Stress in the Developing Brain. Antioxidants and Redox Signaling, 2011, 14, 1535-1550. | 5.4 | 207 |
| 102 | The Yin and Yang of Microglia. Developmental Neuroscience, 2011, 33, 199-209. | 2.0 | 272 |
| 103 | Whatâ€™s the hype about CDK5RAP2?. Cellular and Molecular Life Sciences, 2011, 68, 1719-1736. | 5.4 | 28 |
| 104 | Inflammation processes in perinatal brain damage. Journal of Neural Transmission, 2010, 117, 1009-1017. | 2.8 | 51 |
| 105 | Common molecular causes for congenital heart defects and microcephaly. American Journal of Obstetrics and Gynecology, 2010, 202, e7. | 1.3 | 1 |
| 106 | Microglial MyD88 signaling regulates acute neuronal toxicity of LPS-stimulated microglia in vitro. Brain, Behavior, and Immunity, 2010, 24, 776-783. | 4.1 | 71 |
| 107 | Many roads lead to primary autosomal recessive microcephaly. Progress in Neurobiology, 2010, 90, 363-383. | 5.7 | 181 |
| 108 | The Somatostatin 2A Receptor Is Enriched in Migrating Neurons during Rat and Human Brain Development and Stimulates Migration and Axonal Outgrowth. PLoS ONE, 2009, 4, e5509. | 2.5 | 28 |

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|---|---|---|---|
| 109 | Melatonin Promotes Oligodendroglial Maturation of Injured White Matter in Neonatal Rats. PLoS ONE, 2009, 4, e7128. | 2.5 | 94 |
| 110 | Erythropoietin Attenuates Hyperoxia-Induced Cell Death by Modulation of Inflammatory Mediators and Matrix Metalloproteinases. Developmental Neuroscience, 2009, 31, 394-402. | 2.0 | 35 |
| 111 | Molecular Mechanisms Involved in Injury to the Preterm Brain. Journal of Child Neurology, 2009, 24, 1112-1118. | 1.4 | 72 |
| 112 | Autosomal recessive primary microcephalies (MCPH). European Journal of Paediatric Neurology, 2009, 13, 458. | 1.6 | 4 |
| 113 | Sedative and anticonvulsant drugs suppress postnatal neurogenesis. Annals of Neurology, 2008, 64, 434-445. | 5.3 | 157 |
| 114 | Erythropoietin protects the developing brain from hyperoxiaâ€induced cell death and proteome changes. Annals of Neurology, 2008, 64, 523-534. | 5.3 | 62 |
| 115 | Novel RYR1 missense mutation causes core rod myopathy. European Journal of Neurology, 2008, 15, e31-2. | 3.3 | 21 |
| 116 | Synaptic NMDA receptor activity boosts intrinsic antioxidant defenses. Nature Neuroscience, 2008, 11, 476-487. | 14.8 | 483 |
| 117 | Brief Alteration of NMDA or GABAA Receptor-mediated Neurotransmission Has Long Term Effects on the Developing Cerebral Cortex. Molecular and Cellular Proteomics, 2008, 7, 2293-2310. | 3.8 | 60 |
| 118 | Spinal Muscular Atrophy With Respiratory Distress Type 1 (SMARD1). Journal of Child Neurology, 2008, 23, 199-204. | 1.4 | 49 |
| 119 | Dynamics of Somatostatin Type 2A Receptor Cargoes in Living Hippocampal Neurons. Journal of Neuroscience, 2008, 28, 4336-4349. | 3.6 | 32 |
| 120 | Abnormal Distribution of Calcium-Handling Proteins. Journal of Neuropathology and Experimental Neurology, 2007, 66, 57-65. | 1.7 | 25 |
| 121 | Rapid and reliable detection of exon rearrangements in various movement disorders genes by multiplex ligationâ€dependent probe amplification. Movement Disorders, 2007, 22, 1708-1714. | 3.9 | 16 |
| 122 | Subacute proteome changes following traumatic injury of the developing brain: Implications for a dysregulation of neuronal migration and neurite arborization. Proteomics - Clinical Applications, 2007, 1, 640-649. | 1.6 | 13 |
| 123 | Glutamate antagonists are neurotoxins for the developing brain. Neurotoxicity Research, 2007, 11, 203-218. | 2.7 | 17 |
| 124 | Comparative Proteomics in Neurodegenerative and Non-neurodegenerative Diseases Suggest Nodal Point Proteins in Regulatory Networking. Journal of Proteome Research, 2006, 5, 1948-1958. | 3.7 | 59 |
| 125 | Facing the genetic heterogeneity in neuromuscular disorders: Linkage analysis as an economic diagnostic approach towards the molecular diagnosis. Neuromuscular Disorders, 2006, 16, 4-13. | 0.6 | 19 |
| 126 | Acute and long-term proteome changes induced by oxidative stress in the developing brain. Cell Death and Differentiation, 2006, 13, 1097-1109. | 11.2 | 53 |

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|---|---|---|---|
| 127 | Limb girdle muscular dystrophy type 2I caused by a novel missense mutation in the FKRP gene presenting as acute virus-associated myositis in infancy. European Journal of Pediatrics, 2006, 165, 62-63. | 2.7 | 7 |
| 128 | Acute Ascending Motoric Paraplegia Following Intrathecal Chemotherapy for Treatment of Acute Lymphoblastic Leukemia in Children: Case Reports and Review of the Literature. Klinische Padiatrie, 2006, 218, 350-354. | 0.6 | 13 |
| 129 | Selenoprotein N Muscular Dystrophy. Journal of Child Neurology, 2006, 21, 316-320. | 1.4 | 14 |
| 130 | Mice Lacking the Nuclear Pore Complex Protein ALADIN Show Female Infertility but Fail To Develop a Phenotype Resembling Human Triple A Syndrome. Molecular and Cellular Biology, 2006, 26, 1879-1887. | 2.3 | 41 |
| 131 | Homozygous microdeletion of chromosome 4q11-q12 causes severe limb-girdle muscular dystrophy type 2E with joint hyperlaxity and contractures. Human Mutation, 2005, 26, 279-280. | 2.5 | 12 |
| 132 | Protection with estradiol in developmental models of apoptotic neurodegeneration. Annals of Neurology, 2005, 58, 266-276. | 5.3 | 71 |
| 133 | NMDA antagonist inhibits the extracellular signal-regulated kinase pathway and suppresses cancer growth. Proceedings of the National Academy of Sciences of the United States of America, 2005, 102, 15605-15610. | 7.1 | 129 |
| 134 | Sulthiame but not levetiracetam exerts neurotoxic effect in the developing rat brain. Experimental Neurology, 2005, 193, 497-503. | 4.1 | 130 |
| 135 | Missense mutations of ACTA1 cause dominant congenital myopathy with cores. Journal of Medical Genetics, 2004, 41, 842-848. | 3.2 | 110 |
| 136 | High reproducibility of large-gel two-dimensional electrophoresis. Electrophoresis, 2004, 25, 3040-3047. | 2.4 | 56 |
| 137 | Mutations in the Gene Encoding Gap Junction Protein Î±12 (Connexin 46.6) Cause Pelizaeus-Merzbacherâ€“Like Disease. American Journal of Human Genetics, 2004, 75, 251-260. | 6.2 | 257 |
| 138 | Evaluation of Metabolic Effects of Nusinersen in Patients with Spinal Muscular Atrophy. Journal of Pediatric Neurology, 0, , . | 0.2 | 0 |
| 139 | Real-World Experience Treating Pediatric Epilepsy Patients With Cenobamate. Frontiers in Neurology, 0, 13, . | 2.4 | 14 |