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	Synaptic NMDA receptor activity boosts intrinsic antioxidant defenses. Nature Neuroscience, 2008, 11, 476-487.	14.8	483
2	The Yin and Yang of Microglia. Developmental Neuroscience, 2011, 33, 199-209.	2.0	272
3	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
4	Neuronal Death and Oxidative Stress in the Developing Brain. Antioxidants and Redox Signaling, 2011, 14, 1535-1550.	5.4	207
5	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
6	Many roads lead to primary autosomal recessive microcephaly. Progress in Neurobiology, 2010, 90, 363-383.	5.7	181
7	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
8	Sedative and anticonvulsant drugs suppress postnatal neurogenesis. Annals of Neurology, 2008, 64, 434-445.	5.3	157
9	Golgi-Cox Staining Step by Step. Frontiers in Neuroanatomy, 2016, 10, 38.	1.7	133
10	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
11	Sulthiame but not levetiracetam exerts neurotoxic effect in the developing rat brain. Experimental Neurology, 2005, 193, 497-503.	4.1	130
12	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
13	Synaptic NMDA receptor activity is coupled to the transcriptional control of the glutathione system. Nature Communications, 2015, 6, 6761.	12.8	119
14	Missense mutations of ACTA1 cause dominant congenital myopathy with cores. Journal of Medical Genetics, 2004, 41, 842-848.	3.2	110
15	Melatonin Promotes Oligodendroglial Maturation of Injured White Matter in Neonatal Rats. PLoS ONE, 2009, 4, e7128.	2.5	94
16	Microcephaly. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2013, 111, 129-141.	1.8	94
17	Homozygous YME1L1 mutation causes mitochondriopathy with optic atrophy and mitochondrial network fragmentation. ELife, 2016, 5, .	6.0	88
18	Immunofluorescence Staining of Paraffin Sections Step by Step. Frontiers in Neuroanatomy, 2020, 14, 582218.	1.7	77

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19	Molecular Mechanisms Involved in Injury to the Preterm Brain. Journal of Child Neurology, 2009, 24, 1112-1118.	1.4	72
20	Protection with estradiol in developmental models of apoptotic neurodegeneration. Annals of Neurology, 2005, 58, 266-276.	5.3	71
21	Microglial MyD88 signaling regulates acute neuronal toxicity of LPS-stimulated microglia in vitro. Brain, Behavior, and Immunity, 2010, 24, 776-783.	4.1	71
22	Autosomal Recessive Primary Microcephaly (MCPH): An Update. Neuropediatrics, 2017, 48, 135-142.	0.6	70
23	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
24	Erythropoietin protects the developing brain from hyperoxiaâ€induced cell death and proteome changes. Annals of Neurology, 2008, 64, 523-534.	5.3	62
25	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
26	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
27	Neutrophil oxidative burst activates ATM to regulate cytokine production and apoptosis. Blood, 2015, 126, 2842-2851.	1.4	58
28	PEDIA: prioritization of exome data by image analysis. Genetics in Medicine, 2019, 21, 2807-2814.	2.4	58
29	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
30	High reproducibility of large-gel two-dimensional electrophoresis. Electrophoresis, 2004, 25, 3040-3047.	2.4	56
31	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
32	Redefining the MED13L syndrome. European Journal of Human Genetics, 2015, 23, 1308-1317.	2.8	53
33	Inflammation processes in perinatal brain damage. Journal of Neural Transmission, 2010, 117, 1009-1017.	2.8	51
34	Spinal Muscular Atrophy With Respiratory Distress Type 1 (SMARD1). Journal of Child Neurology, 2008, 23, 199-204.	1.4	49
35	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
36	The clinical, histologic, and genotypic spectrum of SEPN1 -related myopathy. Neurology, 2020, 95, e1512-e1527.	1.1	44

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37	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
38	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
39	MN1 C-terminal truncation syndrome is a novel neurodevelopmental and craniofacial disorder with partial rhombencephalosynapsis. Brain, 2020, 143, 55-68.	7.6	38
40	Erythropoietin Attenuates Hyperoxia-Induced Cell Death by Modulation of Inflammatory Mediators and Matrix Metalloproteinases. Developmental Neuroscience, 2009, 31, 394-402.	2.0	35
41	Growth and psychomotor development of patients with Duchenne muscular dystrophy. European Journal of Paediatric Neurology, 2014, 18, 38-44.	1.6	35
42	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
43	Dynamics of Somatostatin Type 2A Receptor Cargoes in Living Hippocampal Neurons. Journal of Neuroscience, 2008, 28, 4336-4349.	3.6	32
44	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
45	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
46	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
47	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
48	Implanted Neurosphere-Derived Precursors Promote Recovery After Neonatal Excitotoxic Brain Injury. Stem Cells and Development, 2011, 20, 865-879.	2.1	28
49	Whatâ€™s the hype about CDK5RAP2?. Cellular and Molecular Life Sciences, 2011, 68, 1719-1736.	5.4	28
50	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
51	In-depth analysis reveals complex molecular aetiology in a cohort of idiopathic cerebral palsy. Brain, 2022, 145, 119-141.	7.6	28
52	Autosomal Recessive Primary Microcephaly: Not Just a Small Brain. Frontiers in Cell and Developmental Biology, 2021, 9, 784700.	3.7	28
53	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
54	Homozygous ARHGEF2 mutation causes intellectual disability and midbrain-hindbrain malformation. PLoS Genetics, 2017, 13, e1006746.	3.5	27

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55	Recessive mutation in EXOSC3 associates with mitochondrial dysfunction and pontocerebellar hypoplasia. Mitochondrion, 2017, 37, 46-54.	3.4	26
56	Subtelomeric methylation distinguishes between subtypes of Immunodeficiency, Centromeric instability and Facial anomalies syndrome. Human Molecular Genetics, 2018, 27, 3568-3581.	2.9	26
57	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
58	Abnormal Distribution of Calcium-Handling Proteins. Journal of Neuropathology and Experimental Neurology, 2007, 66, 57-65.	1.7	25
59	Autosomal recessive primary microcephalies (MCPH). European Journal of Paediatric Neurology, 2014, 18, 547-548.	1.6	22
60	Novel RYR1 missense mutation causes core rod myopathy. European Journal of Neurology, 2008, 15, e31-2.	3.3	21
61	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
62	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
63	Encephalitis patient-derived monoclonal GABAA receptor antibodies cause epileptic seizures. Journal of Experimental Medicine, 2021, 218, .	8.5	19
64	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
65	Glutamate antagonists are neurotoxins for the developing brain. Neurotoxicity Research, 2007, 11, 203-218.	2.7	17
66	CDK5RAP2 Is Required to Maintain the Germ Cell Pool during Embryonic Development. Stem Cell Reports, 2017, 8, 198-204.	4.8	17
67	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
68	Loss of CDK5RAP2 affects neural but not non-neural mESC differentiation into cardiomyocytes. Cell Cycle, 2015, 14, 2044-2057.	2.6	15
69	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
70	SIGLEC1 (CD169) as a potential diagnostical screening marker for monogenic interferonopathies. Pediatric Allergy and Immunology, 2021, 32, 621-625.	2.6	15
71	Selenoprotein N Muscular Dystrophy. Journal of Child Neurology, 2006, 21, 316-320.	1.4	14
72	Reference genes in the developing murine brain and in differentiating embryonic stem cells. Neurological Research, 2012, 34, 664-668.	1.3	14

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73	Abnormal brain structure and behavior in MyD88-deficient mice. Brain, Behavior, and Immunity, 2021, 91, 181-193.	4.1	14
74	Real-World Experience Treating Pediatric Epilepsy Patients With Cenobamate. Frontiers in Neurology, 0, 13, .	2.4	14
75	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
76	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
77	Phenotype variability of infantile-onset multisystem neurologic, endocrine, and pancreatic disease IMNEPD. Orphanet Journal of Rare Diseases, 2016, 11, 52.	2.7	13
78	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
79	Acute Disseminated Encephalomyelitis After Human Parechovirus Infection. Pediatric Infectious Disease Journal, 2016, 35, 35-38.	2.0	12
80	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
81	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
82	PTRH2 gene mutation causes progressive congenital skeletal muscle pathology. Human Molecular Genetics, 2017, 26, 1458-1464.	2.9	10
83	Brain malformations and cognitive performance in spina bifida. Developmental Medicine and Child Neurology, 2021, 63, 295-302.	2.1	9
84	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
85	Previously reported new type of autosomal recessive primary microcephaly is caused by compound heterozygousASPMgene mutations. Cell Cycle, 2014, 13, 1650-1651.	2.6	8
86	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
87	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
88	Altered inhibition and excitation in neocortical circuits in congenital microcephaly. Neurobiology of Disease, 2019, 129, 130-143.	4.4	7
89	Clinical Outcome of Children With Corpus Callosum Agenesis. Pediatric Neurology, 2020, 112, 47-52.	2.1	7
90	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

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91	OUP accepted manuscript. Human Molecular Genetics, 2021, 30, 2068-2081.	2.9	7
92	Genetic causes of MCPH in consanguineous Pakistani families. Clinical Genetics, 2016, 89, 744-745.	2.0	6
93	Lacosamide Lowers Valproate and Levetiracetam Levels. Neuropediatrics, 2017, 48, 188-189.	0.6	6
94	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
95	Intravenous Nimodipine Treatment for Severe Episode of ATP1A2 Hemiplegic Migraine. Pediatric Neurology, 2020, 112, 71-72.	2.1	6
96	Systematic Classification of Spina Bifida. Journal of Neuropathology and Experimental Neurology, 2021, 80, 294-305.	1.7	6
97	Case Report: Hemispherotomy in the First Days of Life to Treat Drug-Resistant Lesional Epilepsy. Frontiers in Neurology, 2021, 12, 818972.	2.4	6
98	Large homozygous RAB3GAP1 gene microdeletion causes Warburg Micro Syndrome 1. Orphanet Journal of Rare Diseases, 2014, 9, 113.	2.7	5
99	Homozygous mutation in MCM7 causes autosomal recessive primary microcephaly and intellectual disability. Journal of Medical Genetics, 2022, 59, 453-461.	3.2	5
100	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
101	Lessons learned from drug trials in neurofibromatosis: A systematic review. European Journal of Medical Genetics, 2021, 64, 104281.	1.3	5
102	Autosomal recessive primary microcephalies (MCPH). European Journal of Paediatric Neurology, 2009, 13, 458.	1.6	4
103	Combined deletion 18q22.2 and duplication/triplication 18q22.1 causes microcephaly, mental retardation and leukencephalopathy. Gene, 2013, 523, 92-98.	2.2	4
104	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
105	Congenital microcephalyâ€linked CDK5RAP2 affects eye development. Annals of Human Genetics, 2020, 84, 87-91.	0.8	4
106	Age-specific occurrence of pathological fractures in patients with spina bifida. European Journal of Pediatrics, 2020, 179, 773-779.	2.7	4
107	Presence of anti-neuronal antibodies in children with neurological disorders beyond encephalitis. European Journal of Paediatric Neurology, 2020, 28, 159-166.	1.6	4
108	Lumbar Puncture Opening Pressure in Patients with Spinal Muscular Atrophy. Neuropediatrics, 2021, 52, 219-223.	0.6	4

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109	Arhgef2 regulates neural differentiation in the cerebral cortex through mRNA m6A-methylation of Npdc1 and Cend1. IScience, 2021, 24, 102645.	4.1	4
110	Infratentorial MRI Findings in Rasmussen Encephalitis Suggest Primary Cerebellar Involvement. Neurology: Neuroimmunology and NeuroInflammation, 2021, 8, .	6.0	4
111	Novel Alternative Splice Variants of Mouse Cdk5rap2. PLoS ONE, 2015, 10, e0136684.	2.5	4
112	Cerebral Abnormalities in Spina Bifida: A Neuropathological Study. Pediatric and Developmental Pathology, 2022, 25, 107-123.	1.0	3
113	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
114	Is cannabidiol worth a trial in Rasmussen encephalitis?. European Journal of Paediatric Neurology, 2022, 37, 53-55.	1.6	3
115	Cross-sectional Neuromuscular Phenotyping Study of Patients With Arhinia With SMCHD1 Variants. Neurology, 2022, 98, .	1.1	3
116	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
117	Is microcephaly a so-far unrecognized feature of XYY syndrome?. Meta Gene, 2014, 2, 160-163.	0.6	2
118	Selenium Status in Paediatric Patients with Neurodevelopmental Diseases. Nutrients, 2022, 14, 2375.	4.1	2
119	Interferon receptor dysfunction in a child with malignant atrophic papulosis and CNS involvement. Lancet Neurology, The, 2022, 21, 682-686.	10.2	2
120	Common molecular causes for congenital heart defects and microcephaly. American Journal of Obstetrics and Gynecology, 2010, 202, e7.	1.3	1
121	Phenotype of five patients with dopa-responsive dystonia and mutations in GCH1. Journal of Pediatric Neurology, 2015, 03, 083-087.	0.2	1
122	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
123	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
124	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
125	Zebrafish modeling mimics developmental phenotype of patients with <scp>RAPGEF1</scp> mutation. Clinical Genetics, 2021, 100, 144-155.	2.0	1
126	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

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127	Expanding the Phenotype of NUP85 Mutations beyond Nephrotic Syndrome to Primary Autosomal Recessive Microcephaly and Seckel Syndrome Spectrum Disorders. , 2021, 52, .		1
128	Case Report: Behavioral Disorder Following Hemispherotomy: A Valproate Effect?. Frontiers in Neurology, 2021, 12, 764376.	2.4	1
129	Standard values for MRI brain biometry throughout the first year of life. Pediatrics and Neonatology, 2022, 63, 255-261.	0.9	1
130	Evaluation of Metabolic Effects of Nusinersen in Patients with Spinal Muscular Atrophy. Journal of Pediatric Neurology, 0, , .	0.2	0
131	Neuroprotective Strategies. , 2012, , 1173-1179.		0
132	P 306. Role of Cdk5rap2 in Neocortical Development. , 2018, 49, .		0
133	FV 757. Clinical, Radiological and Genetic Spectrum of a Large Pediatric Cohort with Epilepsy. Neuropediatrics, 2018, 49, .	0.6	0
134	Advantages of Botulinum Toxin A Treatment in Combination with Controlled Dynamic Stretching Orthotics for the Treatment of Contractures. , 2021, 52, .		0
135	First Report of Glioblastoma and Associated PNKP Mutation. Neuropediatrics, 2021, 52, .	0.6	0
136	Beware of Neurotoxins in Common Plants: Water Hemlock Poisoning Presenting as Convulsive Status Epilepticus. Pediatric Neurology, 2021, 127, 39-40.	2.1	0
137	Modified Zipper Method: A Promising Treatment Option in Severe Pediatric Cases of Immune-Mediated Neurological Disorders. Neuropediatrics, 2021, 52, .	0.6	0
138	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
139	Relationship between cerebral palsy severity and cognition, aids and education. Minerva Pediatrics, 2022, , .	0.4	0