

Elisa Fazzi

List of Publications by Year in descending order

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120
papers

8,563
citations

87888
38
h-index

49909
87
g-index

123
all docs

123
docs citations

123
times ranked

9267
citing authors

#	ARTICLE	IF	CITATIONS
1	Action Observation Treatment in a tele-rehabilitation setting: a pilot study in children with cerebral palsy. Disability and Rehabilitation, 2022, 44, 1107-1112.	1.8	21
2	Age-Related Effects on the Spectrum of Cerebral Visual Impairment in Children With Cerebral Palsy. Frontiers in Human Neuroscience, 2022, 16, 750464.	2.0	12
3	DNA damage contributes to neurotoxic inflammation in Aicardi-GoutiÃres syndrome astrocytes. Journal of Experimental Medicine, 2022, 219, .	8.5	35
4	Basal Ganglia Dysmorphism in Patients With Aicardi Syndrome. Neurology, 2021, 96, e1319-e1333.	1.1	6
5	Late-Onset Aicardi-GoutiÃres Syndrome: A Characterization of Presenting Clinical Features. Pediatric Neurology, 2021, 115, 1-6.	2.1	18
6	Molecular characterization of a complex small supernumerary marker chromosome derived from chromosome 18p: an addition to the literature. Molecular Cytogenetics, 2021, 14, 6.	0.9	2
7	The Influence of Treatment of Inflammatory Arthritis During Pregnancy on the Long-Term Childrenâs Outcome. Frontiers in Pharmacology, 2021, 12, 626258.	3.5	5
8	The epileptology of Aicardi-GoutiÃres syndrome: electro-clinical-radiological findings. Seizure: the Journal of the British Epilepsy Association, 2021, 86, 197-209.	2.0	2
9	Early visual training and environmental adaptation for infants with visual impairment. Developmental Medicine and Child Neurology, 2021, 63, 1180-1193.	2.1	22
10	IFN-Î± levels in ruxolitinib-treated Aicardi-GoutiÃres patient during SARS-CoV-2 infection: A case report. Clinical Immunology, 2021, 227, 108743.	3.2	1
11	Neurodevelopmental Profile in Children Affected by Ocular Albinism. Neuropediatrics, 2021, , .	0.6	5
12	Case Report: The JAK-Inhibitor Ruxolitinib Use in Aicardi-Goutieres Syndrome Due to ADAR1 Mutation. Frontiers in Pediatrics, 2021, 9, 725868.	1.9	9
13	Impaired urinary concentration ability is a sensitive predictor of renal disease progression in Joubert syndrome. Nephrology Dialysis Transplantation, 2020, 35, 1195-1202.	0.7	15
14	Visual Function Classification System for children with cerebral palsy: development and validation. Developmental Medicine and Child Neurology, 2020, 62, 104-110.	2.1	46
15	Developmental Outcomes of Aicardi GoutiÃres Syndrome. Journal of Child Neurology, 2020, 35, 7-16.	1.4	40
16	Novel and emerging treatments for Aicardi-GoutiÃres syndrome. Expert Review of Clinical Immunology, 2020, 16, 189-198.	3.0	27
17	Long-term outcome of children born from mothers with autoimmune diseases. Best Practice and Research in Clinical Obstetrics and Gynaecology, 2020, 64, 107-116.	2.8	6
18	Genetic and phenotypic spectrum associated with IFIH1 gain-of-function. Human Mutation, 2020, 41, 837-849.	2.5	63

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19	Commentary on “Catatonia in a Patient with Aicardi-Goutières Syndrome Efficiently Treated with Immunoabsorption”. Schizophrenia Research, 2020, 224, 188-189.	2.0	0
20	Autistic-Like Features in Visually Impaired Children: A Review of Literature and Directions for Future Research. Brain Sciences, 2020, 10, 507.	2.3	21
21	New clinical needs and strategies for care in children with neurodisability during COVID-19. Developmental Medicine and Child Neurology, 2020, 62, 879-880.	2.1	43
22	Morpho-functional survey in children suspected of inherited retinal dystrophies via video recording, electrophysiology and genetic analysis. International Ophthalmology, 2020, 40, 2523-2534.	1.4	2
23	Questionnaires as screening tools for children with cerebral visual impairment. Developmental Medicine and Child Neurology, 2020, 62, 891-891.	2.1	8
24	Development of a neurologic severity scale for Aicardi Goutières Syndrome. Molecular Genetics and Metabolism, 2020, 130, 153-160.	1.1	25
25	Establishment of three iPSC lines from fibroblasts of a patient with Aicardi Goutières syndrome mutated in RNaseH2B. Stem Cell Research, 2019, 41, 101620.	0.7	6
26	Generation of three isogenic induced Pluripotent Stem Cell lines (iPSCs) from fibroblasts of a patient with Aicardi Goutières Syndrome carrying a c.2471G>A dominant mutation in IFIH1 gene. Stem Cell Research, 2019, 41, 101623.	0.7	4
27	Generation of three iPSC lines from fibroblasts of a patient with Aicardi Goutières Syndrome mutated in TREX1. Stem Cell Research, 2019, 41, 101580.	0.7	8
28	Molecular Genetics and Interferon Signature in the Italian Aicardi Goutières Syndrome Cohort: Report of 12 New Cases and Literature Review. Journal of Clinical Medicine, 2019, 8, 750.	2.4	29
29	Cerebral Visual Impairment and Clinical Assessment: The European Perspective. Seminars in Pediatric Neurology, 2019, 31, 15-24.	2.0	44
30	Autism in Children With Cerebral and Peripheral Visual Impairment: Fact or Artifact?. Seminars in Pediatric Neurology, 2019, 31, 57-67.	2.0	18
31	FRI0060...NEUROCOGNITIVE PROFILE IN CHILDREN BORN TO MOTHERS WITH CHRONIC ARTHRITIS: WHICH RELATIONSHIP WITH MATERNAL FEELING OF DISEASE?. , 2019, , .		0
32	SLC19A3 related disorder: Treatment implication and clinical outcome of 2 new patients. European Journal of Paediatric Neurology, 2018, 22, 332-335.	1.6	6
33	Neurovisual Assessment in Children with Ataxia Telangiectasia. Neuropediatrics, 2018, 49, 026-034.	0.6	6
34	Sine causa tetraparesis. Medicine (United States), 2018, 97, e13893.	1.0	9
35	Music reduces pain perception in healthy newborns: A comparison between different music tracks and recorded heartbeat. Early Human Development, 2018, 124, 7-10.	1.8	19
36	Childhood Absence Epilepsy evolving to Eyelid Myoclonia with Absence Epilepsy. Seizure: the Journal of the British Epilepsy Association, 2018, 61, 1-3.	2.0	8

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37	White matter changes associated with cognitive visual dysfunctions in children with cerebral palsy: A diffusion tensor imaging study. <i>Journal of Neuroscience Research</i> , 2018, 96, 1766-1774.	2.9	17
38	Action Observation Treatment Improves Upper Limb Motor Functions in Children with Cerebral Palsy: A Combined Clinical and Brain Imaging Study. <i>Neural Plasticity</i> , 2018, 2018, 1-11.	2.2	51
39	Global motion and form processing and attention deficits in multiple child cohorts with neurodevelopmental disorders: Dorsal vulnerability or dorsal/ventral integration?. <i>Journal of Vision</i> , 2018, 18, 546.	0.3	0
40	Genetic, Phenotypic, and Interferon Biomarker Status in ADAR1-Related Neurological Disease. <i>Neuropediatrics</i> , 2017, 48, 166-184.	0.6	62
41	Neonatal Assessment Visual European Grid (NAVEG): Unveiling neurological risk. , 2017, 49, 21-30.		12
42	Ãvaluation et prise en charge des troubles visuels de lâ™ancien prÃ©maturÃ©. <i>Contraste</i> , 2016, NÂ° 43, 89-102.		2
43	Exploring Autoimmunity in a Cohort of Children with Genetically Confirmed Aicardi-GoutiÃ©res Syndrome. <i>Journal of Clinical Immunology</i> , 2016, 36, 693-699.	3.8	21
44	Patient-reported outcomes measure for children born preterm: validation of the <scp>SOLE VLBWI</scp> Questionnaire, a new quality of life self-assessment tool. <i>Developmental Medicine and Child Neurology</i> , 2016, 58, 957-964.	2.1	6
45	Neuroradiologic patterns and novel imaging findings in Aicardi-GoutiÃ©res syndrome. <i>Neurology</i> , 2016, 86, 28-35.	1.1	59
46	Positive effect of erythrocyte-delivered dexamethasone in ataxia-telangiectasia. <i>Neurology: Neuroimmunology and NeuroInflammation</i> , 2015, 2, e98.	6.0	59
47	Characterization of human disease phenotypes associated with mutations in <i>TREX1</i>, <i>RNASEH2A</i>, <i>RNASEH2B</i>, <i>RNASEH2C</i>, <i>SAMHD1</i>, <i>ADAR</i>, and <i>IFIH1</i>. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 296-312.	1.2	447
48	Gain-of-function mutations in IFIH1 cause a spectrum of human disease phenotypes associated with upregulated type I interferon signaling. <i>Nature Genetics</i> , 2014, 46, 503-509.	21.4	490
49	Intra-Erythrocyte Infusion of Dexamethasone Reduces Neurological Symptoms in Ataxia Teleangiectasia Patients: Results of a Phase 2 Trial. <i>Orphanet Journal of Rare Diseases</i> , 2014, 9, 5.	2.7	114
50	Bilateral striatal necrosis in two subjects with Aicardi-GoutiÃ©res syndrome due to mutations in <i>ADAR1</i> (<i>AGS6</i>). <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 815-819.	1.2	30
51	Long-Term Outcome of Children of Rheumatic Disease Patients. , 2014, , 289-303.		1
52	Neuroni specchio in etÃ evolutiva: prospettive cliniche e di ricerca. , 2014, , 191-204.		0
53	Body experiences, emotional competence, and psychosocial functioning in juvenile idiopathic arthritis. <i>Rheumatology International</i> , 2013, 33, 2045-2052.	3.0	39
54	Aicardi-GoutiÃ©res syndrome, a rare neurological disease in children: A new autoimmune disorder?. <i>Autoimmunity Reviews</i> , 2013, 12, 506-509.	5.8	50

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55	Unimanual and Bimanual Intensive Training in Children With Hemiplegic Cerebral Palsy and Persistence in Time of Hand Function Improvement. <i>Journal of Child Neurology</i> , 2013, 28, 161-175.	1.4	34
56	Assessment of interferon-related biomarkers in Aicardi-Goutières syndrome associated with mutations in TREX1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, and ADAR: a case-control study. <i>Lancet Neurology</i> , 2013, 12, 1159-1169.	10.2	473
57	Synonymous Mutations in <i>RNASEH2A</i> Create Cryptic Splice Sites Impairing RNase H2 Enzyme Function in Aicardi-Goutières Syndrome. <i>Human Mutation</i> , 2013, 34, 1066-1070.	2.5	16
58	Family History of Autoimmune Disease in Patients with Aicardi-Goutières Syndrome. <i>Clinical and Developmental Immunology</i> , 2012, 2012, 1-6.	3.3	4
59	Visual Impairment: A Common Sequela of Preterm Birth. <i>NeoReviews</i> , 2012, 13, e542-e550.	0.8	12
60	Neuro-ophthalmological disorders in cerebral palsy: ophthalmological, oculomotor, and visual aspects. <i>Developmental Medicine and Child Neurology</i> , 2012, 54, 730-736.	2.1	137
61	Improving upper limb motor functions through action observation treatment: a pilot study in children with cerebral palsy. <i>Developmental Medicine and Child Neurology</i> , 2012, 54, 822-828.	2.1	122
62	Mutations in ADAR1 cause Aicardi-Goutières syndrome associated with a type I interferon signature. <i>Nature Genetics</i> , 2012, 44, 1243-1248.	21.4	712
63	Different Mutations in Three Prime Repair Exonuclease 1 and Ribonuclease H2 Genes Affect Clinical Features in Aicardi-Goutières Syndrome. <i>Journal of Child Neurology</i> , 2012, 27, 51-60.	1.4	4
64	Outcome of extremely low birth weight infants: What's new in the third millennium? Neuropsychological profiles at four years. <i>Early Human Development</i> , 2012, 88, 241-250.	1.8	21
65	Reach on sound: A key to object permanence in visually impaired children. <i>Early Human Development</i> , 2011, 87, 289-296.	1.8	19
66	Molecular and Clinical Characterization of Albinism in a Large Cohort of Italian Patients. , 2011, 52, 1281.		58
67	New Case of 4H Syndrome and a Review of the Literature. <i>Pediatric Neurology</i> , 2010, 42, 359-364.	2.1	21
68	<i>MKS3/TMEM67</i> mutations are a major cause of COACH Syndrome, a Joubert Syndrome related disorder with liver involvement. <i>Human Mutation</i> , 2009, 30, E432-E442.	2.5	96
69	Mutations involved in Aicardi-Goutières syndrome implicate SAMHD1 as regulator of the innate immune response. <i>Nature Genetics</i> , 2009, 41, 829-832.	21.4	610
70	Mutations in INPP5E, encoding inositol polyphosphate-5-phosphatase E, link phosphatidylinositol signaling to the ciliopathies. <i>Nature Genetics</i> , 2009, 41, 1032-1036.	21.4	383
71	Interferon-Related Transcriptome Alterations in the Cerebrospinal Fluid Cells of Aicardi-Goutières Patients ^{<sup>} . <i>Brain Pathology</i> , 2009, 19, 650-660.	4.1	26
72	Cognitive visual dysfunctions in preterm children with periventricular leukomalacia. <i>Developmental Medicine and Child Neurology</i> , 2009, 51, 974-981.	2.1	160

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73	Infant sex, obstetric risk factors, and 2-year neurodevelopmental outcome among preterm infants. <i>Developmental Medicine and Child Neurology</i> , 2009, 51, 518-525.	2.1	58
74	Multisite Trial on Efficacy of Constraint-Induced Movement Therapy in Children with Hemiplegia. <i>American Journal of Physical Medicine and Rehabilitation</i> , 2009, 88, 216-230.	1.4	14
75	Sleep disturbances in visually impaired toddlers. <i>Brain and Development</i> , 2008, 30, 572-578.	1.1	18
76	Neurodevelopmental evolution of West syndrome: A 2-year prospective study. <i>European Journal of Paediatric Neurology</i> , 2008, 12, 387-397.	1.6	17
77	Rational basis for the development of coenzyme Q10 as a neurotherapeutic agent for retinal protection. <i>Progress in Brain Research</i> , 2008, 173, 575-582.	1.4	57
78	Spectrum of Visual Disorders in Children With Cerebral Visual Impairment. <i>Journal of Child Neurology</i> , 2007, 22, 294-301.	1.4	156
79	The Development of Visual Object Recognition in School-Age Children. <i>Developmental Neuropsychology</i> , 2007, 31, 79-102.	1.4	40
80	Cognitive Profiles and Visuo-perceptual Abilities in Preterm and Term Spastic Diplegic Children With Periventricular Leukomalacia. <i>Journal of Child Neurology</i> , 2007, 22, 282-288.	1.4	59
81	Mutation analysis of NPHP6/CEP290 in patients with Joubert syndrome and Senior Loken syndrome. <i>Journal of Medical Genetics</i> , 2007, 44, 657-663.	3.2	93
82	Clinical and Molecular Genetics of Leber's Congenital Amaurosis: A Multicenter Study of Italian Patients. , 2007, 48, 4284.		131
83	CEP290 Mutations Are Frequently Identified in the Oculo-Renal Form of Joubert Syndrome-Related Disorders. <i>American Journal of Human Genetics</i> , 2007, 81, 104-113.	6.2	137
84	Clinical and Molecular Phenotype of Aicardi-Goutières Syndrome. <i>American Journal of Human Genetics</i> , 2007, 81, 713-725.	6.2	375
85	â€ˆRandÃ² et al. replyâ€™. <i>Developmental Medicine and Child Neurology</i> , 2007, 48, 942-943.	2.1	0
86	Spectrum of NPHP6/CEP290 mutations in Leber congenital amaurosis and delineation of the associated phenotype. <i>Human Mutation</i> , 2007, 28, 416-416.	2.5	224
87	Changes in the Optic Disc Excavation of Children Affected by Cerebral Visual Impairment: A Tomographic Analysis. , 2006, 47, 484.		29
88	Mutations in CEP290, which encodes a centrosomal protein, cause pleiotropic forms of Joubert syndrome. <i>Nature Genetics</i> , 2006, 38, 623-625.	21.4	368
89	Mutations in genes encoding ribonuclease H2 subunits cause Aicardi-Goutières syndrome and mimic congenital viral brain infection. <i>Nature Genetics</i> , 2006, 38, 910-916.	21.4	592
90	RandÃ² et al. reply. <i>Developmental Medicine and Child Neurology</i> , 2006, 48, 942.	2.1	0

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91	A questionnaire on sleep behaviour in the first years of life: preliminary results from a normative sample. <i>Functional Neurology</i> , 2006, 21, 151-8.	1.3	9
92	Prognostic Value of Umbilical Artery Doppler Studies in Unselected Preterm Deliveries. <i>Obstetrics and Gynecology</i> , 2005, 105, 613-620.	2.4	31
93	Towards improved clinical characterization of Leber congenital amaurosis: Neurological and systemic findings. <i>American Journal of Medical Genetics, Part A</i> , 2005, 132A, 13-19.	1.2	22
94	Factors Predicting the Efficacy of Botulinum Toxin-A Treatment of the Lower Limb in Children With Cerebral Palsy. <i>Journal of Child Neurology</i> , 2005, 20, 661-666.	1.4	32
95	Neurochemical Evidence to Implicate Elevated Glutamate in the Mechanisms of High Intraocular Pressure (IOP)-induced Retinal Ganglion Cell Death in Rat. <i>NeuroToxicology</i> , 2005, 26, 935-941.	3.0	137
96	Cognitive competence at the onset of West syndrome: correlation with EEG patterns and visual function. <i>Developmental Medicine and Child Neurology</i> , 2005, 47, 760-765.	2.1	0
97	Cognitive competence at the onset of West syndrome: correlation with EEG patterns and visual function. <i>Developmental Medicine and Child Neurology</i> , 2005, 47, 760.	2.1	20
98	Obstetric risk factors and persistent increases in brain parenchymal echogenicity in preterm infants. <i>BJOG: an International Journal of Obstetrics and Gynaecology</i> , 2004, 111, 913-918.	2.3	11
99	Visual Function in Infants with West Syndrome: Correlation with EEG Patterns. <i>Epilepsia</i> , 2004, 45, 781-786.	5.1	30
100	Visual perceptual impairment in children with periventricular leukomalacia. <i>Brain and Development</i> , 2004, 26, 506-512.	1.1	115
101	Two-year infant neurodevelopmental outcome after single or multiple antenatal courses of corticosteroids to prevent complications of prematurity. <i>American Journal of Obstetrics and Gynecology</i> , 2004, 191, 217-224.	1.3	134
102	Fetal Growth and Infant Neurodevelopmental Outcome After Preterm Premature Rupture of Membranes. <i>Obstetrics and Gynecology</i> , 2004, 103, 1286-1293.	2.4	15
103	Leber's congenital amaurosis: an update. <i>European Journal of Paediatric Neurology</i> , 2003, 7, 13-22.	1.6	49
104	Aicardi-Goutières syndrome: differential diagnosis and aetiopathogenesis. <i>Functional Neurology</i> , 2003, 18, 71-5.	1.3	11
105	Aicardi-Goutières syndrome: a description of 21 new cases and a comparison with the literature. <i>European Journal of Paediatric Neurology</i> , 2002, 6, A9-A22.	1.6	50
106	Visual acuity in the first two years of life in healthy term newborns: an experience with the teller acuity cards. <i>Functional Neurology</i> , 2002, 17, 87-92.	1.3	15
107	Predictors of Independent Walking in Children With Spastic Diplegia. <i>Journal of Child Neurology</i> , 2000, 15, 228-234.	1.4	36
108	Developmental sequence of postural control in prone position in children with spastic diplegia. <i>Brain and Development</i> , 2000, 22, 436-444.	1.1	4

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109	Stereotyped behaviours in blind children. Brain and Development, 1999, 21, 522-528.	1.1	79
110	Preeclampsia, preterm delivery and infant cerebral palsy. European Journal of Obstetrics, Gynecology and Reproductive Biology, 1998, 77, 151-155.	1.1	36
111	Neurodevelopmental outcome in very low birth weight infants at 24 months and 5 to 7 years of age: Changing diagnosis. Pediatric Neurology, 1997, 17, 240-248.	2.1	26
112	Meconium-Stained Amniotic Fluid and Risk for Cerebral Palsy in Preterm Infants. Obstetrics and Gynecology, 1997, 90, 519-523.	2.4	38
113	Antenatal and delivery risk factors simultaneously associated with neonatal death and cerebral palsy in preterm infants. Early Human Development, 1997, 48, 81-91.	1.8	46
114	Ring chromosome 9: An atypical case. Brain and Development, 1996, 18, 216-219.	1.1	10
115	Effect of preterm premature rupture of membranes on neurodevelopmental outcome: follow up at two years of age. BJOG: an International Journal of Obstetrics and Gynaecology, 1995, 102, 882-887.	2.3	123
116	Severity of abruptio placentae and neurodevelopmental outcome in low birth weight infants. Early Human Development, 1993, 35, 45-54.	1.8	44
117	Infant neurodevelopmental outcome in pregnancies complicated by gestational hypertension and intra-uterine growth retardation. Journal of Perinatal Medicine, 1993, 21, 195-203.	1.4	33
118	The influence of presentation and method of delivery on neonatal mortality and infant neurodevelopmental outcome in nondiscordant low-birthweight (< 2500 g) twin gestations. European Journal of Obstetrics, Gynecology and Reproductive Biology, 1992, 47, 189-194.	1.1	55
119	Early predictors of neurodevelopmental outcome at 12-36 months in very low-birthweight infants. Brain and Development, 1990, 12, 482-487.	1.1	10
120	Migraine, Mitral Valve Prolapse and Platelet Function in the Pediatric Age Group. Headache, 1986, 26, 142-145.	3.9	9