

Elisa Fazzi

List of Publications by Year in descending order

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120
papers

8,563
citations

100601

38
h-index

56606

87
g-index

123
all docs

123
docs citations

123
times ranked

9967
citing authors

#	ARTICLE	IF	CITATIONS
1	Mutations in ADAR1 cause Aicardi-GoutiÃres syndrome associated with a type I interferon signature. <i>Nature Genetics</i> , 2012, 44, 1243-1248.	9.4	712
2	Mutations involved in Aicardi-GoutiÃres syndrome implicate SAMHD1 as regulator of the innate immune response. <i>Nature Genetics</i> , 2009, 41, 829-832.	9.4	610
3	Mutations in genes encoding ribonuclease H2 subunits cause Aicardi-GoutiÃres syndrome and mimic congenital viral brain infection. <i>Nature Genetics</i> , 2006, 38, 910-916.	9.4	592
4	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.	9.4	490
5	Assessment of interferon-related biomarkers in Aicardi-GoutiÃres syndrome associated with mutations in TREX1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, and ADAR: a case-control study. <i>Lancet Neurology</i> , The, 2013, 12, 1159-1169.	4.9	473
6	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.	0.7	447
7	Mutations in INPP5E, encoding inositol polyphosphate-5-phosphatase E, link phosphatidylinositol signaling to the ciliopathies. <i>Nature Genetics</i> , 2009, 41, 1032-1036.	9.4	383
8	Clinical and Molecular Phenotype of Aicardi-GoutiÃres Syndrome. <i>American Journal of Human Genetics</i> , 2007, 81, 713-725.	2.6	375
9	Mutations in CEP290, which encodes a centrosomal protein, cause pleiotropic forms of Joubert syndrome. <i>Nature Genetics</i> , 2006, 38, 623-625.	9.4	368
10	Spectrum of NPHP6/CEP290 mutations in Leber congenital amaurosis and delineation of the associated phenotype. <i>Human Mutation</i> , 2007, 28, 416-416.	1.1	224
11	Cognitive visual dysfunctions in preterm children with periventricular leukomalacia. <i>Developmental Medicine and Child Neurology</i> , 2009, 51, 974-981.	1.1	160
12	Spectrum of Visual Disorders in Children With Cerebral Visual Impairment. <i>Journal of Child Neurology</i> , 2007, 22, 294-301.	0.7	156
13	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.	1.4	137
14	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.	2.6	137
15	Neuroophthalmological disorders in cerebral palsy: ophthalmological, oculomotor, and visual aspects. <i>Developmental Medicine and Child Neurology</i> , 2012, 54, 730-736.	1.1	137
16	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.	0.7	134
17	Clinical and Molecular Genetics of Leber's Congenital Amaurosis: A Multicenter Study of Italian Patients. , 2007, 48, 4284.		131
18	Effect of preterm premature rupture of membranes on neurodevelopmental outcome: follow up at two years of age. <i>BJOG: an International Journal of Obstetrics and Gynaecology</i> , 1995, 102, 882-887.	1.1	123

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19	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.	1.1	122
20	Visual perceptual impairment in children with periventricular leukomalacia. <i>Brain and Development</i> , 2004, 26, 506-512.	0.6	115
21	Intra-Erythrocyte Infusion of Dexamethasone Reduces Neurological Symptoms in Ataxia Telangiectasia Patients: Results of a Phase 2 Trial. <i>Orphanet Journal of Rare Diseases</i> , 2014, 9, 5.	1.2	114
22	<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.	1.1	96
23	Mutation analysis of NPHP6/CEP290 in patients with Joubert syndrome and Senior Loken syndrome. <i>Journal of Medical Genetics</i> , 2007, 44, 657-663.	1.5	93
24	Stereotyped behaviours in blind children. <i>Brain and Development</i> , 1999, 21, 522-528.	0.6	79
25	Genetic and phenotypic spectrum associated with IFIH1 gain of function. <i>Human Mutation</i> , 2020, 41, 837-849.	1.1	63
26	Genetic, Phenotypic, and Interferon Biomarker Status in ADAR1-Related Neurological Disease. <i>Neuropediatrics</i> , 2017, 48, 166-184.	0.3	62
27	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.	0.7	59
28	Positive effect of erythrocyte-delivered dexamethasone in ataxia-telangiectasia. <i>Neurology: Neuroimmunology and Neuroinflammation</i> , 2015, 2, e98.	3.1	59
29	Neuroradiologic patterns and novel imaging findings in Aicardi-Goutières syndrome. <i>Neurology</i> , 2016, 86, 28-35.	1.5	59
30	Infant sex, obstetric risk factors, and 2-year neurodevelopmental outcome among preterm infants. <i>Developmental Medicine and Child Neurology</i> , 2009, 51, 518-525.	1.1	58
31	Molecular and Clinical Characterization of Albinism in a Large Cohort of Italian Patients. , 2011, 52, 1281.		58
32	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.	0.9	57
33	The influence of presentation and method of delivery on neonatal mortality and infant neurodevelopmental outcome in nondiscordant low-birthweight (< 2500 g) twin gestations. <i>European Journal of Obstetrics, Gynecology and Reproductive Biology</i> , 1992, 47, 189-194.	0.5	55
34	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.	1.0	51
35	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.	0.7	50
36	Aicardi-Goutières syndrome, a rare neurological disease in children: A new autoimmune disorder?. <i>Autoimmunity Reviews</i> , 2013, 12, 506-509.	2.5	50

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37	Leber's congenital amaurosis: an update. <i>European Journal of Paediatric Neurology</i> , 2003, 7, 13-22.	0.7	49
38	Antenatal and delivery risk factors simultaneously associated with neonatal death and cerebral palsy in preterm infants. <i>Early Human Development</i> , 1997, 48, 81-91.	0.8	46
39	Visual Function Classification System for children with cerebral palsy: development and validation. <i>Developmental Medicine and Child Neurology</i> , 2020, 62, 104-110.	1.1	46
40	Severity of abruptio placentae and neurodevelopmental outcome in low birth weight infants. <i>Early Human Development</i> , 1993, 35, 45-54.	0.8	44
41	Cerebral Visual Impairment and Clinical Assessment: The European Perspective. <i>Seminars in Pediatric Neurology</i> , 2019, 31, 15-24.	1.0	44
42	New clinical needs and strategies for care in children with neurodisability during COVID-19. <i>Developmental Medicine and Child Neurology</i> , 2020, 62, 879-880.	1.1	43
43	The Development of Visual Object Recognition in School-Age Children. <i>Developmental Neuropsychology</i> , 2007, 31, 79-102.	1.0	40
44	Developmental Outcomes of Aicardi Goutières Syndrome. <i>Journal of Child Neurology</i> , 2020, 35, 7-16.	0.7	40
45	Body experiences, emotional competence, and psychosocial functioning in juvenile idiopathic arthritis. <i>Rheumatology International</i> , 2013, 33, 2045-2052.	1.5	39
46	Meconium-Stained Amniotic Fluid and Risk for Cerebral Palsy in Preterm Infants. <i>Obstetrics and Gynecology</i> , 1997, 90, 519-523.	1.2	38
47	Preeclampsia, preterm delivery and infant cerebral palsy. <i>European Journal of Obstetrics, Gynecology and Reproductive Biology</i> , 1998, 77, 151-155.	0.5	36
48	Predictors of Independent Walking in Children With Spastic Diplegia. <i>Journal of Child Neurology</i> , 2000, 15, 228-234.	0.7	36
49	DNA damage contributes to neurotoxic inflammation in Aicardi-Goutières syndrome astrocytes. <i>Journal of Experimental Medicine</i> , 2022, 219, .	4.2	35
50	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.	0.7	34
51	Infant neurodevelopmental outcome in pregnancies complicated by gestational hypertension and intra-uterine growth retardation. <i>Journal of Perinatal Medicine</i> , 1993, 21, 195-203.	0.6	33
52	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.	0.7	32
53	Prognostic Value of Umbilical Artery Doppler Studies in Unselected Preterm Deliveries. <i>Obstetrics and Gynecology</i> , 2005, 105, 613-620.	1.2	31
54	Visual Function in Infants with West Syndrome: Correlation with EEG Patterns. <i>Epilepsia</i> , 2004, 45, 781-786.	2.6	30

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55	Bilateral striatal necrosis in two subjects with Aicardi-Goutières syndrome due to mutations in <i>ADAR1</i> (<i>AGS6</i>). <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 815-819.	0.7	30
56	Changes in the Optic Disc Excavation of Children Affected by Cerebral Visual Impairment: A Tomographic Analysis. , 2006, 47, 484.		29
57	Molecular Genetics and Interferon Signature in the Italian Aicardi Goutières Syndrome Cohort: Report of 12 New Cases and Literature Review. <i>Journal of Clinical Medicine</i> , 2019, 8, 750.	1.0	29
58	Novel and emerging treatments for Aicardi-Goutières syndrome. <i>Expert Review of Clinical Immunology</i> , 2020, 16, 189-198.	1.3	27
59	Neurodevelopmental outcome in very low birth weight infants at 24 months and 5 to 7 years of age: Changing diagnosis. <i>Pediatric Neurology</i> , 1997, 17, 240-248.	1.0	26
60	Interferon-Related Transcriptome Alterations in the Cerebrospinal Fluid Cells of Aicardi-Goutières Patients ^{<sup> </sup>} . <i>Brain Pathology</i> , 2009, 19, 650-660.	2.1	26
61	Development of a neurologic severity scale for Aicardi Goutières Syndrome. <i>Molecular Genetics and Metabolism</i> , 2020, 130, 153-160.	0.5	25
62	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.	0.7	22
63	Early visual training and environmental adaptation for infants with visual impairment. <i>Developmental Medicine and Child Neurology</i> , 2021, 63, 1180-1193.	1.1	22
64	New Case of 4H Syndrome and a Review of the Literature. <i>Pediatric Neurology</i> , 2010, 42, 359-364.	1.0	21
65	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.	0.8	21
66	Exploring Autoimmunity in a Cohort of Children with Genetically Confirmed Aicardi-Goutières Syndrome. <i>Journal of Clinical Immunology</i> , 2016, 36, 693-699.	2.0	21
67	Autistic-Like Features in Visually Impaired Children: A Review of Literature and Directions for Future Research. <i>Brain Sciences</i> , 2020, 10, 507.	1.1	21
68	Action Observation Treatment in a tele-rehabilitation setting: a pilot study in children with cerebral palsy. <i>Disability and Rehabilitation</i> , 2022, 44, 1107-1112.	0.9	21
69	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.	1.1	20
70	Reach on sound: A key to object permanence in visually impaired children. <i>Early Human Development</i> , 2011, 87, 289-296.	0.8	19
71	Music reduces pain perception in healthy newborns: A comparison between different music tracks and recorded heartbeat. <i>Early Human Development</i> , 2018, 124, 7-10.	0.8	19
72	Sleep disturbances in visually impaired toddlers. <i>Brain and Development</i> , 2008, 30, 572-578.	0.6	18

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73	Autism in Children With Cerebral and Peripheral Visual Impairment: Fact or Artifact?. <i>Seminars in Pediatric Neurology</i> , 2019, 31, 57-67.	1.0	18
74	Late-Onset Aicardi-Goutières Syndrome: A Characterization of Presenting Clinical Features. <i>Pediatric Neurology</i> , 2021, 115, 1-6.	1.0	18
75	Neurodevelopmental evolution of West syndrome: A 2-year prospective study. <i>European Journal of Paediatric Neurology</i> , 2008, 12, 387-397.	0.7	17
76	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.	1.3	17
77	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.	1.1	16
78	Fetal Growth and Infant Neurodevelopmental Outcome After Preterm Premature Rupture of Membranes. <i>Obstetrics and Gynecology</i> , 2004, 103, 1286-1293.	1.2	15
79	Impaired urinary concentration ability is a sensitive predictor of renal disease progression in Joubert syndrome. <i>Nephrology Dialysis Transplantation</i> , 2020, 35, 1195-1202.	0.4	15
80	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
81	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.	0.7	14
82	Visual Impairment: A Common Sequela of Preterm Birth. <i>NeoReviews</i> , 2012, 13, e542-e550.	0.4	12
83	Neonatal Assessment Visual European Grid (NAVEG): Unveiling neurological risk. , 2017, 49, 21-30.		12
84	Age-Related Effects on the Spectrum of Cerebral Visual Impairment in Children With Cerebral Palsy. <i>Frontiers in Human Neuroscience</i> , 2022, 16, 750464.	1.0	12
85	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.	1.1	11
86	Aicardi-Goutières syndrome: differential diagnosis and aetiopathogenesis. <i>Functional Neurology</i> , 2003, 18, 71-5.	1.3	11
87	Early predictors of neurodevelopmental outcome at 12–36 months in very low-birthweight infants. <i>Brain and Development</i> , 1990, 12, 482-487.	0.6	10
88	Ring chromosome 9: An atypical case. <i>Brain and Development</i> , 1996, 18, 216-219.	0.6	10
89	Migraine, Mitral Valve Prolapse and Platelet Function in the Pediatric Age Group. <i>Headache</i> , 1986, 26, 142-145.	1.8	9
90	Sine causa tetraparesis. <i>Medicine (United States)</i> , 2018, 97, e13893.	0.4	9

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91	Case Report: The JAK-Inhibitor Ruxolitinib Use in Aicardi-Goutieres Syndrome Due to ADAR1 Mutation. <i>Frontiers in Pediatrics</i> , 2021, 9, 725868.	0.9	9
92	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
93	Childhood Absence Epilepsy evolving to Eyelid Myoclonia with Absence Epilepsy. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2018, 61, 1-3.	0.9	8
94	Generation of three iPSC lines from fibroblasts of a patient with Aicardi Gouti�res Syndrome mutated in TREX1. <i>Stem Cell Research</i> , 2019, 41, 101580.	0.3	8
95	Questionnaires as screening tools for children with cerebral visual impairment. <i>Developmental Medicine and Child Neurology</i> , 2020, 62, 891-891.	1.1	8
96	Patient�reported outcomes measure for children born preterm: validation of the <sc>SOLE VLBWI</sc> Questionnaire, a new quality of life self�assessment tool. <i>Developmental Medicine and Child Neurology</i> , 2016, 58, 957-964.	1.1	6
97	SLC19A3 related disorder: Treatment implication and clinical outcome of 2 new patients. <i>European Journal of Paediatric Neurology</i> , 2018, 22, 332-335.	0.7	6
98	Neurovisual Assessment in Children with Ataxia Telangiectasia. <i>Neuropediatrics</i> , 2018, 49, 026-034.	0.3	6
99	Establishment of three iPSC lines from fibroblasts of a patient with Aicardi Gouti�res syndrome mutated in RNaseH2B. <i>Stem Cell Research</i> , 2019, 41, 101620.	0.3	6
100	Long-term outcome of children born from mothers with autoimmune diseases. <i>Best Practice and Research in Clinical Obstetrics and Gynaecology</i> , 2020, 64, 107-116.	1.4	6
101	Basal Ganglia Dysmorphism in Patients With Aicardi Syndrome. <i>Neurology</i> , 2021, 96, e1319-e1333.	1.5	6
102	The Influence of Treatment of Inflammatory Arthritis During Pregnancy on the Long-Term Children�s Outcome. <i>Frontiers in Pharmacology</i> , 2021, 12, 626258.	1.6	5
103	Neurodevelopmental Profile in Children Affected by Ocular Albinism. <i>Neuropediatrics</i> , 2021, , .	0.3	5
104	Developmental sequence of postural control in prone position in children with spastic diplegia. <i>Brain and Development</i> , 2000, 22, 436-444.	0.6	4
105	Family History of Autoimmune Disease in Patients with Aicardi-Gouti�res Syndrome. <i>Clinical and Developmental Immunology</i> , 2012, 2012, 1-6.	3.3	4
106	Different Mutations in Three Prime Repair Exonuclease 1 and Ribonuclease H2 Genes Affect Clinical Features in Aicardi-Gouti�res Syndrome. <i>Journal of Child Neurology</i> , 2012, 27, 51-60.	0.7	4
107	Generation of three isogenic induced Pluripotent Stem Cell lines (iPSCs) from fibroblasts of a patient with Aicardi Gouti�res Syndrome carrying a c.2471G>A dominant mutation in IFIH1 gene. <i>Stem Cell Research</i> , 2019, 41, 101623.	0.3	4
108	�valuation et prise en charge des troubles visuels de lâancien pr�matur�. <i>Contraste</i> , 2016, N� 43, 89-102		2

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109	Morpho-functional survey in children suspected of inherited retinal dystrophies via video recording, electrophysiology and genetic analysis. <i>International Ophthalmology</i> , 2020, 40, 2523-2534.	0.6	2
110	Molecular characterization of a complex small supernumerary marker chromosome derived from chromosome 18p: an addition to the literature. <i>Molecular Cytogenetics</i> , 2021, 14, 6.	0.4	2
111	The epileptology of Aicardi-GoutiÃ¨res syndrome: electro-clinical-radiological findings. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2021, 86, 197-209.	0.9	2
112	IFN-Î± levels in ruxolitinib-treated Aicardi-GoutiÃ¨res patient during SARS-CoV-2 infection: A case report. <i>Clinical Immunology</i> , 2021, 227, 108743.	1.4	1
113	Long-Term Outcome of Children of Rheumatic Disease Patients. , 2014, , 289-303.		1
114	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.	1.1	0
115	Ã©RandÃ© et al. replyÃ©. <i>Developmental Medicine and Child Neurology</i> , 2007, 48, 942-943.	1.1	0
116	FRI0060Ã©...NEUROCOGNITIVE PROFILE IN CHILDREN BORN TO MOTHERS WITH CHRONIC ARTHRITIS: WHICH RELATIONSHIP WITH MATERNAL FEELING OF DISEASE?. , 2019, , .		0
117	Commentary on Ã©Catatonia in a Patient with Aicardi-GoutiÃ¨res Syndrome Efficiently Treated with ImmunoabsorptionÃ©. <i>Schizophrenia Research</i> , 2020, 224, 188-189.	1.1	0
118	RandÃ© et al. reply. <i>Developmental Medicine and Child Neurology</i> , 2006, 48, 942.	1.1	0
119	Neuroni specchio in etÃ© evolutiva: prospettive cliniche e di ricerca. , 2014, , 191-204.		0
120	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.	0.1	0