Elisa Fazzi

List of Publications by Year
in descending order

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120	8,563	87888	49909
papers	citations	38 h-index	87 g-index

123	123	123	9267
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Mutations in ADAR1 cause Aicardi-GoutiÃ¨res syndrome associated with a type I interferon signature. Nature Genetics, 2012, 44, 1243-1248.	21.4	712
2	Mutations involved in Aicardi-GoutiÃ¨res syndrome implicate SAMHD1 as regulator of the innate immune response. Nature Genetics, 2009, 41, 829-832.	21.4	610
3	Mutations in genes encoding ribonuclease H2 subunits cause Aicardi-GoutiÃ¨res syndrome and mimic congenital viral brain infection. Nature Genetics, 2006, 38, 910-916.	21.4	592
4	Gain-of-function mutations in IFIH1 cause a spectrum of human disease phenotypes associated with upregulated type I interferon signaling. Nature Genetics, 2014, 46, 503-509.	21.4	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. Lancet Neurology, The, 2013, 12, 1159-1169.	10.2	473
6	Characterization of human disease phenotypes associated with mutations in TREX1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, ADAR, and IFIH1. American Journal of Medical Genetics, Part A, 2015, 167, 296-312.	1.2	447
7	Mutations in INPP5E, encoding inositol polyphosphate-5-phosphatase E, link phosphatidyl inositol signaling to the ciliopathies. Nature Genetics, 2009, 41, 1032-1036.	21.4	383
8	Clinical and Molecular Phenotype of Aicardi-GoutiÃ¨res Syndrome. American Journal of Human Genetics, 2007, 81, 713-725.	6.2	375
9	Mutations in CEP290, which encodes a centrosomal protein, cause pleiotropic forms of Joubert syndrome. Nature Genetics, 2006, 38, 623-625.	21.4	368
10	Spectrum of NPHP6/CEP290 mutations in Leber congenital amaurosis and delineation of the associated phenotype. Human Mutation, 2007, 28, 416-416.	2.5	224
11	Cognitive visual dysfunctions in preterm children with periventricular leukomalacia. Developmental Medicine and Child Neurology, 2009, 51, 974-981.	2.1	160
12	Spectrum of Visual Disorders in Children With Cerebral Visual Impairment. Journal of Child Neurology, 2007, 22, 294-301.	1.4	156
13	Neurochemical Evidence to Implicate Elevated Glutamate in the Mechanisms of High Intraocular Pressure (IOP)-induced Retinal Ganglion Cell Death in Rat. NeuroToxicology, 2005, 26, 935-941.	3.0	137
14	CEP290 Mutations Are Frequently Identified in the Oculo-Renal Form of Joubert Syndromeâ€“Related Disorders. American Journal of Human Genetics, 2007, 81, 104-113.	6.2	137
15	Neuroâ€ophthalmological disorders in cerebral palsy: ophthalmological, oculomotor, and visual aspects. Developmental Medicine and Child Neurology, 2012, 54, 730-736.	2.1	137
16	Two-year infant neurodevelopmental outcome after single or multiple antenatal courses of corticosteroids to prevent complications of prematurity. American Journal of Obstetrics and Gynecology, 2004, 191, 217-224.	1.3	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. BJOG: an International Journal of Obstetrics and Gynaecology, 1995, 102, 882-887.	2.3	123

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19	Improving upper limb motor functions through action observation treatment: a pilot study in children with cerebral palsy. Developmental Medicine and Child Neurology, 2012, 54, 822-828.	2.1	122
20	Visualâ€“perceptual impairment in children with periventricular leukomalacia. Brain and Development, 2004, 26, 506-512.	1.1	115
21	Intra-Erythrocyte Infusion of Dexamethasone Reduces Neurological Symptoms in Ataxia Teleangiectasia Patients: Results of a Phase 2 Trial. Orphanet Journal of Rare Diseases, 2014, 9, 5.	2.7	114
22	MKS3/TMEM67mutations are a major cause of COACH Syndrome, a Joubert Syndrome related disorder with liver involvement. Human Mutation, 2009, 30, E432-E442.	2.5	96
23	Mutation analysis of NPHP6/CEP290 in patients with Joubert syndrome and Senior Loken syndrome. Journal of Medical Genetics, 2007, 44, 657-663.	3.2	93
24	Stereotyped behaviours in blind children. Brain and Development, 1999, 21, 522-528.	1.1	79
25	Genetic and phenotypic spectrum associated with IFIH1 gainâ€ofâ€function. Human Mutation, 2020, 41, 837-849.	2.5	63
26	Genetic, Phenotypic, and Interferon Biomarker Status in ADAR1-Related Neurological Disease. Neuropediatrics, 2017, 48, 166-184.	0.6	62
27	Cognitive Profiles and Visuoperceptual Abilities in Preterm and Term Spastic Diplegic Children With Periventricular Leukomalacia. Journal of Child Neurology, 2007, 22, 282-288.	1.4	59
28	Positive effect of erythrocyte-delivered dexamethasone in ataxia-telangiectasia. Neurology: Neuroimmunology and NeuroInflammation, 2015, 2, e98.	6.0	59
29	Neuroradiologic patterns and novel imaging findings in Aicardi-GoutiÃ¨res syndrome. Neurology, 2016, 86, 28-35.	1.1	59
30	Infant sex, obstetric risk factors, and 2â€year neurodevelopmental outcome among preterm infants. Developmental Medicine and Child Neurology, 2009, 51, 518-525.	2.1	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. Progress in Brain Research, 2008, 173, 575-582.	1.4	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. European Journal of Obstetrics, Gynecology and Reproductive Biology, 1992, 47, 189-194.	1.1	55
34	Action Observation Treatment Improves Upper Limb Motor Functions in Children with Cerebral Palsy: A Combined Clinical and Brain Imaging Study. Neural Plasticity, 2018, 2018, 1-11.	2.2	51
35	Aicardi-GoutiÃ¨res syndrome: a description of 21 new cases and a comparison with the literature. European Journal of Paediatric Neurology, 2002, 6, A9-A22.	1.6	50
36	Aicardiâ€“Goutieres syndrome, a rare neurological disease in children: A new autoimmune disorder?. Autoimmunity Reviews, 2013, 12, 506-509.	5.8	50

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37	Leber's congenital amaurosis: an update. European Journal of Paediatric Neurology, 2003, 7, 13-22.	1.6	49
38	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
39	Visual Function Classification System for children with cerebral palsy: development and validation. Developmental Medicine and Child Neurology, 2020, 62, 104-110.	2.1	46
40	Severity of abruptio placentae and neurodevelopmental outcome in low birth weight infants. Early Human Development, 1993, 35, 45-54.	1.8	44
41	Cerebral Visual Impairment and Clinical Assessment: The European Perspective. Seminars in Pediatric Neurology, 2019, 31, 15-24.	2.0	44
42	New clinical needs and strategies for care in children with neurodisability during COVIDâ€19. Developmental Medicine and Child Neurology, 2020, 62, 879-880.	2.1	43
43	The Development of Visual Object Recognition in School-Age Children. Developmental Neuropsychology, 2007, 31, 79-102.	1.4	40
44	Developmental Outcomes of Aicardi GoutiÃ¨res Syndrome. Journal of Child Neurology, 2020, 35, 7-16.	1.4	40
45	Body experiences, emotional competence, and psychosocial functioning in juvenile idiopathic arthritis. Rheumatology International, 2013, 33, 2045-2052.	3.0	39
46	Meconium-Stained Amniotic Fluid and Risk for Cerebral Palsy in Preterm Infants. Obstetrics and Gynecology, 1997, 90, 519-523.	2.4	38
47	Preeclampsia, preterm delivery and infant cerebral palsy. European Journal of Obstetrics, Gynecology and Reproductive Biology, 1998, 77, 151-155.	1.1	36
48	Predictors of Independent Walking in Children With Spastic Diplegia. Journal of Child Neurology, 2000, 15, 228-234.	1.4	36
49	DNA damage contributes to neurotoxic inflammation in Aicardi-GoutiÃ¨res syndrome astrocytes. Journal of Experimental Medicine, 2022, 219, .	8.5	35
50	Unimanual and Bimanual Intensive Training in Children With Hemiplegic Cerebral Palsy and Persistence in Time of Hand Function Improvement. Journal of Child Neurology, 2013, 28, 161-175.	1.4	34
51	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
52	Factors Predicting the Efficacy of Botulinum Toxin-A Treatment of the Lower Limb in Children With Cerebral Palsy. Journal of Child Neurology, 2005, 20, 661-666.	1.4	32
53	Prognostic Value of Umbilical Artery Doppler Studies in Unselected Preterm Deliveries. Obstetrics and Gynecology, 2005, 105, 613-620.	2.4	31
54	Visual Function in Infants with West Syndrome: Correlation with EEG Patterns. Epilepsia, 2004, 45, 781-786.	5.1	30

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55	Bilateral striatal necrosis in two subjects with Aicardiâ€“GoutiÃ¨res syndrome due to mutations in ADAR1 (AGS6). American Journal of Medical Genetics, Part A, 2014, 164, 815-819.	1.2	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 GoutiÃ¨res Syndrome Cohort: Report of 12 New Cases and Literature Review. Journal of Clinical Medicine, 2019, 8, 750.	2.4	29
58	Novel and emerging treatments for Aicardi-GoutiÃ¨res syndrome. Expert Review of Clinical Immunology, 2020, 16, 189-198.	3.0	27
59	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
60	Interferonâ€Related Transcriptome Alterations in the Cerebrospinal Fluid Cells of Aicardiâ€GoutiÃ¨res Patients^{â€}. Brain Pathology, 2009, 19, 650-660.	4.1	26
61	Development of a neurologic severity scale for Aicardi GoutiÃ¨res Syndrome. Molecular Genetics and Metabolism, 2020, 130, 153-160.	1.1	25
62	Towards improved clinical characterization of Leber congenital amaurosis: Neurological and systemic findings. American Journal of Medical Genetics, Part A, 2005, 132A, 13-19.	1.2	22
63	Early visual training and environmental adaptation for infants with visual impairment. Developmental Medicine and Child Neurology, 2021, 63, 1180-1193.	2.1	22
64	New Case of 4H Syndrome and a Review of the Literature. Pediatric Neurology, 2010, 42, 359-364.	2.1	21
65	Outcome of extremely low birth weight infants: What's new in the third millennium? Neuropsychological profiles at four years. Early Human Development, 2012, 88, 241-250.	1.8	21
66	Exploring Autoimmunity in a Cohort of Children with Genetically Confirmed Aicardiâ€“GoutiÃ¨res Syndrome. Journal of Clinical Immunology, 2016, 36, 693-699.	3.8	21
67	Autistic-Like Features in Visually Impaired Children: A Review of Literature and Directions for Future Research. Brain Sciences, 2020, 10, 507.	2.3	21
68	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
69	Cognitive competence at the onset of West syndrome: correlation with EEG patterns and visual function. Developmental Medicine and Child Neurology, 2005, 47, 760.	2.1	20
70	Reach on sound: A key to object permanence in visually impaired children. Early Human Development, 2011, 87, 289-296.	1.8	19
71	Music reduces pain perception in healthy newborns: A comparison between different music tracks and recoded heartbeat. Early Human Development, 2018, 124, 7-10.	1.8	19
72	Sleep disturbances in visually impaired toddlers. Brain and Development, 2008, 30, 572-578.	1.1	18

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73	Autism in Children With Cerebral and Peripheral Visual Impairment: Fact or Artifact?. Seminars in Pediatric Neurology, 2019, 31, 57-67.	2.0	18
74	Late-Onset Aicardi-GoutiÃ¨res Syndrome: A Characterization of Presenting Clinical Features. Pediatric Neurology, 2021, 115, 1-6.	2.1	18
75	Neurodevelopmental evolution of West syndrome: A 2-year prospective study. European Journal of Paediatric Neurology, 2008, 12, 387-397.	1.6	17
76	White matter changes associated with cognitive visual dysfunctions in children with cerebral palsy: A diffusion tensor imaging study. Journal of Neuroscience Research, 2018, 96, 1766-1774.	2.9	17
77	Synonymous Mutations inRNASEH2ACreate Cryptic Splice Sites Impairing RNase H2 Enzyme Function in Aicardi-GoutiÃ¨res Syndrome. Human Mutation, 2013, 34, 1066-1070.	2.5	16
78	Fetal Growth and Infant Neurodevelopmental Outcome After Preterm Premature Rupture of Membranes. Obstetrics and Gynecology, 2004, 103, 1286-1293.	2.4	15
79	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
80	Visual acuity in the first two years of life in healthy term newborns: an experience with the teller acuity cards. Functional Neurology, 2002, 17, 87-92.	1.3	15
81	Multisite Trial on Efficacy of Constraint-Induced Movement Therapy in Children with Hemiplegia. American Journal of Physical Medicine and Rehabilitation, 2009, 88, 216-230.	1.4	14
82	Visual Impairment: A Common Sequela of Preterm Birth. NeoReviews, 2012, 13, e542-e550.	0.8	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. Frontiers in Human Neuroscience, 2022, 16, 750464.	2.0	12
85	Obstetric risk factors and persistent increases in brain parenchymal echogenicity in preterm infants. BJOG: an International Journal of Obstetrics and Gynaecology, 2004, 111, 913-918.	2.3	11
86	Aicardi-GoutiÃ¨res syndrome: differential diagnosis and aetiopathogenesis. Functional Neurology, 2003, 18, 71-5.	1.3	11
87	Early predictors of neurodevelopmental outcome at 12â€“36 months in very low-birthweight infants. Brain and Development, 1990, 12, 482-487.	1.1	10
88	Ring chromosome 9: An atypical case. Brain and Development, 1996, 18, 216-219.	1.1	10
89	Migraine, Mitral Valve Prolapse and Platelet Function in the Pediatric Age Group. Headache, 1986, 26, 142-145.	3.9	9
90	Sine causa tetraparesis. Medicine (United States), 2018, 97, e13893.	1.0	9

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91	Case Report: The JAK-Inhibitor Ruxolitinib Use in Aicardi-Goutieres Syndrome Due to ADAR1 Mutation. Frontiers in Pediatrics, 2021, 9, 725868.	1.9	9
92	A questionnaire on sleep behaviour in the first years of life: preliminary results from a normative sample. Functional Neurology, 2006, 21, 151-8.	1.3	9
93	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
94	Generation of three iPSC lines from fibroblasts of a patient with Aicardi GoutiÃ¨res Syndrome mutated in TREX1. Stem Cell Research, 2019, 41, 101580.	0.7	8
95	Questionnaires as screening tools for children with cerebral visual impairment. Developmental Medicine and Child Neurology, 2020, 62, 891-891.	2.1	8
96	Patientâ€reported outcomes measure for children born preterm: validation of the <scp>SOLE VLBWI</scp> Questionnaire, a new quality of life selfâ€assessment tool. Developmental Medicine and Child Neurology, 2016, 58, 957-964.	2.1	6
97	SLC19A3 related disorder: Treatment implication and clinical outcome of 2 new patients. European Journal of Paediatric Neurology, 2018, 22, 332-335.	1.6	6
98	Neurovisual Assessment in Children with Ataxia Telangiectasia. Neuropediatrics, 2018, 49, 026-034.	0.6	6
99	Establishment of three iPSC lines from fibroblasts of a patient with Aicardi GoutiÃ¨res syndrome mutated in RNaseH2B. Stem Cell Research, 2019, 41, 101620.	0.7	6
100	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
101	Basal Ganglia Dysmorphism in Patients With Aicardi Syndrome. Neurology, 2021, 96, e1319-e1333.	1.1	6
102	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
103	Neurodevelopmental Profile in Children Affected by Ocular Albinism. Neuropediatrics, 2021, , .	0.6	5
104	Developmental sequence of postural control in prone position in children with spastic diplegia. Brain and Development, 2000, 22, 436-444.	1.1	4
105	Family History of Autoimmune Disease in Patients with Aicardi-GoutiÃ¨res Syndrome. Clinical and Developmental Immunology, 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-GoutiÃ¨res Syndrome. Journal of Child Neurology, 2012, 27, 51-60.	1.4	4
107	Generation of three isogenic induced Pluripotent Stem Cell lines (iPSCs) from fibroblasts of a patient with Aicardi GoutiÃ¨res Syndrome carrying a c.2471G>A dominant mutation in IFIH1 gene. Stem Cell Research, 2019, 41, 101623.	0.7	4
108	Ã‰valuation et prise en charge des troubles visuels de lâ€™ancien prÃ©maturÃ©. Contraste, 2016, NÂ° 43, 89-102.	0.1	2

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109	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
110	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
111	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
112	IFN-Î± levels in ruxolitinib-treatead Aicardi-GoutiÃ¨res patient during SARS-CoV-2 infection: A case report. Clinical Immunology, 2021, 227, 108743.	3.2	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. Developmental Medicine and Child Neurology, 2005, 47, 760-765.	2.1	0
115	â€˜RandÃ² et al. replyâ€™. Developmental Medicine and Child Neurology, 2007, 48, 942-943.	2.1	0
116	FRI0060â€…NEUROCOGNITIVE PROFILE IN CHILDREN BORN TO MOTHERS WITH CHRONIC ARTHRITIS: WHICH RELATIONSHIP WITH MATERNAL FEELING OF DISEASE?. , 2019, , .		0
117	Commentary on â€œCatatonia in a Patient with Aicardi-GoutiÃ¨res Syndrome Efficiently Treated with Immunoadsorptionâ€• Schizophrenia Research, 2020, 224, 188-189.	2.0	0
118	RandÃ² et al. reply. Developmental Medicine and Child Neurology, 2006, 48, 942.	2.1	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?. Journal of Vision, 2018, 18, 546.	0.3	0