

Elizabeth I Pierpont

List of Publications by Year in descending order

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Version: 2024-02-01

32
papers

1,488
citations

516710

16
h-index

477307

29
g-index

32
all docs

32
docs citations

32
times ranked

1764
citing authors

#	ARTICLE	IF	CITATIONS
1	Specific Language Impairment is not Specific to Language: the Procedural Deficit Hypothesis. <i>Cortex</i> , 2005, 41, 399-433.	2.4	662
2	Cardio-Facio-Cutaneous Syndrome: Clinical Features, Diagnosis, and Management Guidelines. <i>Pediatrics</i> , 2014, 134, e1149-e1162.	2.1	148
3	Genotype differences in cognitive functioning in Noonan syndrome. <i>Genes, Brain and Behavior</i> , 2009, 8, 275-282.	2.2	95
4	A report on state-wide implementation of newborn screening for X-linked Adrenoleukodystrophy. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 1205-1213.	1.2	56
5	Neurocognitive Trajectory of Boys Who Received a Hematopoietic Stem Cell Transplant at an Early Stage of Childhood Cerebral Adrenoleukodystrophy. <i>JAMA Neurology</i> , 2017, 74, 710.	9.0	55
6	Attention skills and executive functioning in children with Noonan syndrome and their unaffected siblings. <i>Developmental Medicine and Child Neurology</i> , 2015, 57, 385-392.	2.1	42
7	Effects of germline mutations in the Ras/MAPK signaling pathway on adaptive behavior: Cardiofaciocutaneous syndrome and Noonan syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 591-600.	1.2	40
8	The Language Phenotype of Children and Adolescents With Noonan Syndrome. <i>Journal of Speech, Language, and Hearing Research</i> , 2010, 53, 917-932.	1.6	39
9	Contributions of phonological and verbal working memory to language development in adolescents with fragile X syndrome. <i>Journal of Neurodevelopmental Disorders</i> , 2011, 3, 335-347.	3.1	36
10	The Arizona Cognitive Test Battery for Down Syndrome: Test-Retest Reliability and Practice Effects. <i>American Journal on Intellectual and Developmental Disabilities</i> , 2017, 122, 215-234.	1.6	35
11	A Neurodevelopmental Perspective on the Acquisition of Nonverbal Cognitive Skills in Adolescents With Fragile X Syndrome. <i>Developmental Neuropsychology</i> , 2013, 38, 445-460.	1.4	31
12	Learning and memory in children with Noonan syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 2250-2257.	1.2	29
13	Neuropsychological Functioning in Individuals with Noonan Syndrome: a Systematic Literature Review with Educational and Treatment Recommendations. <i>Journal of Pediatric Neuropsychology</i> , 2016, 2, 14-33.	0.6	25
14	Associations Between Medical History, Cognition, and Behavior in Youth With Down Syndrome: A Report From the Down Syndrome Cognition Project. <i>American Journal on Intellectual and Developmental Disabilities</i> , 2018, 123, 514-528.	1.6	25
15	Social skills in children with RASopathies: a comparison of Noonan syndrome and neurofibromatosis type 1. <i>Journal of Neurodevelopmental Disorders</i> , 2018, 10, 21.	3.1	25
16	Intrathecal enzyme replacement for Hurler syndrome: biomarker association with neurocognitive outcomes. <i>Genetics in Medicine</i> , 2019, 21, 2552-2560.	2.4	25
17	Behavioral functioning in cardiofaciocutaneous syndrome: Risk factors and impact on parenting experience. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 1974-1988.	1.2	17
18	Neurologic and neurodevelopmental complications in cardiofaciocutaneous syndrome are associated with genotype: A multinational cohort study. <i>Genetics in Medicine</i> , 2022, 24, 1556-1566.	2.4	15

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19	Mosaicism of the UDP-Galactose transporter SLC35A2 in a female causing a congenital disorder of glycosylation: a case report. <i>BMC Medical Genetics</i> , 2018, 19, 100.	2.1	14
20	Variability in clinical and neuropsychological features of individuals with <i>MAP2K1</i> mutations. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 452-459.	1.2	12
21	Neurocognitive benchmarks following transplant for emerging cerebral adrenoleukodystrophy. <i>Neurology</i> , 2020, 95, e591-e600.	1.1	11
22	Association between APOE4 and biomarkers in cerebral adrenoleukodystrophy. <i>Scientific Reports</i> , 2019, 9, 7858.	3.3	10
23	The seventh international RASopathies symposium: Pathways to a cure“ expanding knowledge, enhancing research, and therapeutic discovery. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 1915-1927.	1.2	10
24	Emotional functioning among children with neurofibromatosis type 1 or Noonan syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 2433-2446.	1.2	9
25	Impact of pediatric hypophosphatasia on behavioral health and quality of life. <i>Orphanet Journal of Rare Diseases</i> , 2021, 16, 80.	2.7	8
26	Post-transplant adaptive function in childhood cerebral adrenoleukodystrophy. <i>Annals of Clinical and Translational Neurology</i> , 2018, 5, 252-261.	3.7	7
27	Differential outcomes for frontal versus posterior demyelination in childhood cerebral adrenoleukodystrophy. <i>Journal of Inherited Metabolic Disease</i> , 2021, 44, 1434-1440.	3.6	3
28	Primary Adrenal Insufficiency in a Boy with Type I Diabetes: The Importance of Considering X-linked Adrenoleukodystrophy. <i>Journal of the Endocrine Society</i> , 2022, 6, bvac039.	0.2	3
29	Social behavior in RASopathies and idiopathic autism. <i>Journal of Neurodevelopmental Disorders</i> , 2022, 14, 5.	3.1	1
30	Mary Ella Mascia Pierpont: Geneticist, scientist, mentor, friend (1945–2020). <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 319-323.	1.2	0
31	Cobalamin J disease detected on newborn screening: Novel variant and normal neurodevelopmental course. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1870-1874.	1.2	0
32	Neurocognitive Trajectory of Patients with Childhood Cerebral Adrenoleukodystrophy Who Received Allogeneic Hematopoietic Cell Transplantation at an Early Stage of Cerebral Disease. <i>Blood</i> , 2016, 128, 4682-4682.	1.4	0