Elizabeth I Pierpont

List of Publications by Year
in descending order

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	Social behavior in RASopathies and idiopathic autism. Journal of Neurodevelopmental Disorders, 2022, 14, 5.	3.1	1
2	The seventh international <scp>RASopathies</scp> symposium: Pathways to a cureâ€"expanding knowledge, enhancing research, and therapeutic discovery. American Journal of Medical Genetics, Part A, 2022, 188, 1915-1927.	1.2	10
3	Primary Adrenal Insufficiency in a Boy with Type I Diabetes: The Importance of Considering X-linked Adrenoleukodystrophy. Journal of the Endocrine Society, 2022, 6, bvac039.	0.2	3
4	Neurologic and neurodevelopmental complications in cardiofaciocutaneous syndrome are associated with genotype: A multinational cohort study. Genetics in Medicine, 2022, 24, 1556-1566.	2.4	15
5	Mary Ella Mascia Pierpont: Geneticist, scientist, mentor, friend (1945â€"2020). American Journal of Medical Genetics, Part A, 2021, 185, 319-323.	1.2	0
6	Impact of pediatric hypophosphatasia on behavioral health and quality of life. Orphanet Journal of Rare Diseases, 2021, 16, 80.	2.7	8
7	Cobalamin J disease detected on newborn screening: Novel variant and normal neurodevelopmental course. American Journal of Medical Genetics, Part A, 2021, 185, 1870-1874.	1.2	0
8	Differential outcomes for frontal versus posterior demyelination in childhood cerebral adrenoleukodystrophy. Journal of Inherited Metabolic Disease, 2021, 44, 1434-1440.	3.6	3
9	Neurocognitive benchmarks following transplant for emerging cerebral adrenoleukodystrophy. Neurology, 2020, 95, e591-e600.	1.1	11
10	Emotional functioning among children with neurofibromatosis type 1 or Noonan syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 2433-2446.	1.2	9
11	Association between APOE4 and biomarkers in cerebral adrenoleukodystrophy. Scientific Reports, 2019, 9, 7858.	3.3	10
12	A report on stateâ€wide implementation of newborn screening for Xâ€linked Adrenoleukodystrophy. American Journal of Medical Genetics, Part A, 2019, 179, 1205-1213.	1.2	56
13	Intrathecal enzyme replacement for Hurler syndrome: biomarker association with neurocognitive outcomes. Genetics in Medicine, 2019, 21, 2552-2560.	2.4	25
14	Postâ€transplant adaptive function in childhood cerebral adrenoleukodystrophy. Annals of Clinical and Translational Neurology, 2018, 5, 252-261.	3.7	7
15	Associations Between Medical History, Cognition, and Behavior in Youth With Down Syndrome: A Report From the Down Syndrome Cognition Project. American Journal on Intellectual and Developmental Disabilities, 2018, 123, 514-528.	1.6	25
16	Social skills in children with RASopathies: a comparison of Noonan syndrome and neurofibromatosis type 1. Journal of Neurodevelopmental Disorders, 2018, 10, 21.	3.1	25
17	Mosaicism of the UDP-Galactose transporter SLC35A2 in a female causing a congenital disorder of glycosylation: a case report. BMC Medical Genetics, 2018, 19, 100.	2.1	14
18	Neurocognitive Trajectory of Boys Who Received a Hematopoietic Stem Cell Transplant at an Early Stage of Childhood Cerebral Adrenoleukodystrophy. JAMA Neurology, 2017, 74, 710.	9.0	55

#	Article	IF	Citations
19	The Arizona Cognitive Test Battery for Down Syndrome: Test-Retest Reliability and Practice Effects. American Journal on Intellectual and Developmental Disabilities, 2017, 122, 215-234.	1.6	35
20	Variability in clinical and neuropsychological features of individuals with MAP2K1 mutations. American Journal of Medical Genetics, Part A, 2017, 173, 452-459.	1.2	12
21	Behavioral functioning in cardiofaciocutaneous syndrome: Risk factors and impact on parenting experience. American Journal of Medical Genetics, Part A, 2016, 170, 1974-1988.	1.2	17
22	Neuropsychological Functioning in Individuals with Noonan Syndrome: a Systematic Literature Review with Educational and Treatment Recommendations. Journal of Pediatric Neuropsychology, 2016, 2, 14-33.	0.6	25
23	Neurocognitive Trajectory of Patients with Childhood Cerebral Adrenoleukodystrophy Who Received Allogeneic Hematopoietic Cell Transplantation at an Early Stage of Cerebral Disease. Blood, 2016, 128, 4682-4682.	1.4	0
24	Attention skills and executive functioning in children with Noonan syndrome and their unaffected siblings. Developmental Medicine and Child Neurology, 2015, 57, 385-392.	2.1	42
25	Cardio-Facio-Cutaneous Syndrome: Clinical Features, Diagnosis, and Management Guidelines. Pediatrics, 2014, 134, e1149-e1162.	2.1	148
26	A Neurodevelopmental Perspective on the Acquisition of Nonverbal Cognitive Skills in Adolescents With Fragile X Syndrome. Developmental Neuropsychology, 2013, 38, 445-460.	1.4	31
27	Learning and memory in children with Noonan syndrome. American Journal of Medical Genetics, Part A, 2013, 161, 2250-2257.	1.2	29
28	Contributions of phonological and verbal working memory to language development in adolescents with fragile X syndrome. Journal of Neurodevelopmental Disorders, 2011, 3, 335-347.	3.1	36
29	Effects of germline mutations in the Ras/MAPK signaling pathway on adaptive behavior: Cardiofaciocutaneous syndrome and Noonan syndrome. American Journal of Medical Genetics, Part A, 2010, 152A, 591-600.	1.2	40
30	The Language Phenotype of Children and Adolescents With Noonan Syndrome. Journal of Speech, Language, and Hearing Research, 2010, 53, 917-932.	1.6	39
31	Genotype differences in cognitive functioning in Noonan syndrome. Genes, Brain and Behavior, 2009, 8, 275-282.	2.2	95
32	Specific Language Impairment is not Specific to Language: the Procedural Deficit Hypothesis. Cortex, 2005, 41, 399-433.	2.4	662