Mary Ella Pierpont

List of Publications by Year
in descending order

21	982	623734 14	713466 21
papers	citations	h-index	g-index

22	22	22	2339
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Mutations in transcriptional regulator ATRX establish the functional significance of a PHD-like domain. Nature Genetics, 1997, 17, 146-148.	21.4	196
2	Functional Dysregulation of CDC42 Causes Diverse Developmental Phenotypes. American Journal of Human Genetics, 2018, 102, 309-320.	6.2	138
3	PIK3CA-associated developmental disorders exhibit distinct classes of mutations with variable expression and tissue distribution. JCI Insight, 2016, 1, .	5.0	134
4	Clinical spectrum of individuals with pathogenic N F1 missense variants affecting p.Met1149, p.Arg1276, and p.Lys1423: genotypeâ€“phenotype study in neurofibromatosis type 1. Human Mutation, 2020, 41, 299-315.	2.5	80
5	Expanding the clinical phenotype of individuals with a 3-bp in-frame deletion of the NF1 gene (c.2970_2972del): an update of genotypeâ€“phenotype correlation. Genetics in Medicine, 2019, 21, 867-876.	2.4	62
6	A specific mutation inTBL1XR1causes Pierpont syndrome. Journal of Medical Genetics, 2016, 53, 330-337.	3.2	51
7	CTCF variants in 39 individuals with a variable neurodevelopmental disorder broaden the mutational and clinical spectrum. Genetics in Medicine, 2019, 21, 2723-2733.	2.4	48
8	Clinical phenotype associated with terminal 2q37 deletion. Clinical Genetics, 1995, 48, 134-139.	2.0	47
9	Cardiovascular disease in Noonan syndrome. Current Opinion in Pediatrics, 2018, 30, 601-608.	2.0	42
10	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
11	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
12	Expanding the clinical and genetic spectrum of ALPK3 variants: Phenotypes identified in pediatric cardiomyopathy patients and adults with heterozygous variants. American Heart Journal, 2020, 225, 108-119.	2.7	25
13	A point mutation in the cytb gene of cardiac mtDNA associated with Complex III deficiency in ischemic cardiomyopathy. IUBMB Life, 1996, 40, 487-495.	3.4	17
14	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
15	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
16	Cardiac transplantation in children with Noonan syndrome. Pediatric Transplantation, 2019, 23, e13535.	1.0	12
17	Restrictive cardiomyopathy: an unusual phenotype of a lamin A variant. ESC Heart Failure, 2018, 5, 724-726.	3.1	9
18	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

#	Article	IF	Citations
19	Retinal dystrophy in two boys with Costello syndrome due to the HRAS p.Gly13Cys mutation. , 2017, 173, 1342-1347.		7
20	UBA2 variants underlie a recognizable syndrome with variable aplasia cutis congenita and ectrodactyly. Genetics in Medicine, 2021, 23, 1624-1635.	2.4	7
21	Somatic Mosaicism in a Male Patient WithÂX-linked Alport Syndrome. Kidney International Reports, 2019, 4, 1031-1035.	0.8	6