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	UBA2 variants underlie a recognizable syndrome with variable aplasia cutis congenita and ectrodactyly. Genetics in Medicine, 2021, 23, 1624-1635.	2.4	7
2	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
3	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
4	Somatic Mosaicism in a Male Patient WithÂX-linked Alport Syndrome. Kidney International Reports, 2019, 4, 1031-1035.	0.8	6
5	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
6	Cardiac transplantation in children with Noonan syndrome. Pediatric Transplantation, 2019, 23, e13535.	1.0	12
7	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
8	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
9	Functional Dysregulation of CDC42 Causes Diverse Developmental Phenotypes. American Journal of Human Genetics, 2018, 102, 309-320.	6.2	138
10	Cardiovascular disease in Noonan syndrome. Current Opinion in Pediatrics, 2018, 30, 601-608.	2.0	42
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	Restrictive cardiomyopathy: an unusual phenotype of a lamin A variant. ESC Heart Failure, 2018, 5, 724-726.	3.1	9
13	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
14	Retinal dystrophy in two boys with Costello syndrome due to the HRAS p.Gly13Cys mutation. , 2017, 173, 1342-1347.		7
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	A specific mutation inTBL1XR1causes Pierpont syndrome. Journal of Medical Genetics, 2016, 53, 330-337.	3.2	51
17	PIK3CA-associated developmental disorders exhibit distinct classes of mutations with variable expression and tissue distribution. JCI Insight, 2016, 1, .	5.0	134
18	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

#	Article	IF	Citations
19	Mutations in transcriptional regulator ATRX establish the functional significance of a PHD-like domain. Nature Genetics, 1997, 17, 146-148.	21.4	196
20	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
21	Clinical phenotype associated with terminal 2q37 deletion. Clinical Genetics, 1995, 48, 134-139.	2.0	47