Elaine Spector

List of Publications by Year
in descending order

13 papers	20,843 citations	1307594 7 h-index	1281871 11 g-index
14 all docs	14 docs citations	14 times ranked	33947 citing authors

#	Article	IF	Citations
1	Laboratory testing for fragile X, 2021 revision: a technical standard of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2021, 23, 799-812.	2.4	18
2	The genotypic spectrum of ALDH7A1 mutations resulting in pyridoxine dependent epilepsy: A common epileptic encephalopathy. Journal of Inherited Metabolic Disease, 2019, 42, 353-361.	3.6	54
3	Further delineation of achondroplasiaâ€“hypochondroplasia complex with longâ€term survival. American Journal of Medical Genetics, Part A, 2018, 176, 1225-1231.	1.2	0
4	Response to Biesecker and Harrison. Genetics in Medicine, 2018, 20, 1689-1690.	2.4	7
5	The genetic basis of classic nonketotic hyperglycinemia due to mutations in GLDC and AMT. Genetics in Medicine, 2017, 19, 104-111.	2.4	71
6	d -Glyceric aciduria does not cause nonketotic hyperglycinemia: A historic co-occurrence. Molecular Genetics and Metabolism, 2017, 121, 80-82.	1.1	2
7	Clinical Phenotype of Adult Fragile X Gray Zone Allele Carriers: a Case Series. Cerebellum, 2016, 15, 623-631.	2.5	20
8	The M405V allele of the glutaryl-CoA dehydrogenase gene is an important marker for glutaric aciduria type I (GA-I) low excretors. Molecular Genetics and Metabolism, 2016, 119, 50-56.	1.1	12
9	Detection of gonadal mosaicism in Hartsfield syndrome by next generation sequencing. American Journal of Medical Genetics, Part A, 2016, 170, 3359-3359.	1.2	5
10	Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology. Genetics in Medicine, 2015, 17, 405-424.	2.4	20,455
11	Variant non ketotic hyperglycinemia is caused by mutations in LIAS, BOLA3 and the novel gene GLRX5. Brain, 2014, 137, 366-379.	7.6	187
12	Concurrent non-ketotic hyperglycinemia and propionic acidemia in an eight year old boy. Molecular Genetics and Metabolism Reports, 2014, 1, 237-240.	1.1	5
13	The genotypic spectrum of ALDH7A1 mutations resulting in pyridoxine dependent epilepsy: a common epileptic encephalopathy. Journal of Inherited Metabolic Disease, 0, , .	3.6	6