Elaine Spector

List of Publications by Year
in descending order

13 papers	20,843 citations	1464605 7 h-index	1427216 11 g-index
14 all docs	14 docs citations	14 times ranked	35776 citing authors

#	Article	IF	Citations
1	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.	1.1	20,455
2	Variant non ketotic hyperglycinemia is caused by mutations in LIAS, BOLA3 and the novel gene GLRX5. Brain, 2014, 137, 366-379.	3.7	187
3	The genetic basis of classic nonketotic hyperglycinemia due to mutations in GLDC and AMT. Genetics in Medicine, 2017, 19, 104-111.	1.1	71
4	The genotypic spectrum of ALDH7A1 mutations resulting in pyridoxine dependent epilepsy: A common epileptic encephalopathy. Journal of Inherited Metabolic Disease, 2019, 42, 353-361.	1.7	54
5	Clinical Phenotype of Adult Fragile X Gray Zone Allele Carriers: a Case Series. Cerebellum, 2016, 15, 623-631.	1.4	20
6	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.	1.1	18
7	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.	0.5	12
8	Response to Biesecker and Harrison. Genetics in Medicine, 2018, 20, 1689-1690.	1.1	7
9	The genotypic spectrum of ALDH7A1 mutations resulting in pyridoxine dependent epilepsy: a common epileptic encephalopathy. Journal of Inherited Metabolic Disease, 0, , .	1.7	6
10	Concurrent non-ketotic hyperglycinemia and propionic acidemia in an eight year old boy. Molecular Genetics and Metabolism Reports, 2014, 1, 237-240.	0.4	5
11	Detection of gonadal mosaicism in Hartsfield syndrome by next generation sequencing. American Journal of Medical Genetics, Part A, 2016, 170, 3359-3359.	0.7	5
12	d -Glyceric aciduria does not cause nonketotic hyperglycinemia: A historic co-occurrence. Molecular Genetics and Metabolism, 2017, 121, 80-82.	0.5	2
13	Further delineation of achondroplasiaâ€“hypochondroplasia complex with longâ€term survival. American Journal of Medical Genetics, Part A, 2018, 176, 1225-1231.	0.7	0