

Sara Seneca

List of Publications by Year in descending order

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42
papers

1,239
citations

393982

19
h-index

377514

34
g-index

44
all docs

44
docs citations

44
times ranked

2266
citing authors

#	ARTICLE	IF	CITATIONS
1	Mutations in the X-linked pyruvate dehydrogenase (E1) γ subunit gene (PDHA1) in patients with a pyruvate dehydrogenase complex deficiency. <i>Human Mutation</i> , 2000, 15, 209-219.	1.1	191
2	Blue Native Polyacrylamide Gel Electrophoresis: A Powerful Tool in Diagnosis of Oxidative Phosphorylation Defects. <i>Pediatric Research</i> , 2001, 50, 658-665.	1.1	117
3	Clinical, biochemical, and genetic spectrum of seven patients with NFU1 deficiency. <i>Frontiers in Genetics</i> , 2015, 06, 123.	1.1	81
4	Efficient CRISPR/Cas9-mediated editing of trinucleotide repeat expansion in myotonic dystrophy patient-derived iPS and myogenic cells. <i>Nucleic Acids Research</i> , 2018, 46, 8275-8298.	6.5	78
5	Clinical, biochemical and genetic spectrum of 70 patients with ACAD9 deficiency: is riboflavin supplementation effective?. <i>Orphanet Journal of Rare Diseases</i> , 2018, 13, 120.	1.2	61
6	Biallelic Mutations in TMEM126B Cause Severe Complex I Deficiency with a Variable Clinical Phenotype. <i>American Journal of Human Genetics</i> , 2016, 99, 217-227.	2.6	57
7	GTF2E2 Mutations Destabilize the General Transcription Factor Complex TFIIE in Individuals with DNA Repair-Proficient Trichothiodystrophy. <i>American Journal of Human Genetics</i> , 2016, 98, 627-642.	2.6	49
8	Early onset Huntington disease: a neuronal degeneration syndrome. <i>European Journal of Pediatrics</i> , 2004, 163, 717-721.	1.3	48
9	Mutation of the iron-sulfur cluster assembly gene <i>IBA57</i> causes fatal infantile leukodystrophy. <i>Journal of Inherited Metabolic Disease</i> , 2015, 38, 1147-1153.	1.7	43
10	New insights into the phenotype of FARS2 deficiency. <i>Molecular Genetics and Metabolism</i> , 2017, 122, 172-181.	0.5	38
11	Pearson marrow pancreas syndrome: a molecular study and clinical management. <i>Clinical Genetics</i> , 1997, 51, 338-342.	1.0	37
12	Mutation analysis of the pyruvate dehydrogenase E1 α gene in eight patients with a pyruvate dehydrogenase complex deficiency. , 1996, 7, 46-51.		36
13	Analysis of the whole mitochondrial genome: translation of the Ion Torrent Personal Genome Machine system to the diagnostic bench?. <i>European Journal of Human Genetics</i> , 2015, 23, 41-48.	1.4	33
14	<i>TANGO2</i> deficiency as a cause of neurodevelopmental delay with indirect effects on mitochondrial energy metabolism. <i>Journal of Inherited Metabolic Disease</i> , 2019, 42, 898-908.	1.7	32
15	Experience and outcome of 3 years of a European EQA scheme for genetic testing of the spinocerebellar ataxias. <i>European Journal of Human Genetics</i> , 2008, 16, 913-920.	1.4	23
16	Genetic causes of male infertility. <i>Annales D'Endocrinologie</i> , 2014, 75, 109-111.	0.6	23
17	Random Mutagenesis, Clonal Events, and Embryonic or Somatic Origin Determine the mtDNA Variant Type and Load in Human Pluripotent Stem Cells. <i>Stem Cell Reports</i> , 2018, 11, 102-114.	2.3	23
18	Sertoli Cell-Only Syndrome: Behind the Genetic Scenes. <i>BioMed Research International</i> , 2016, 2016, 1-7.	0.9	22

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19	Complex III staining in blue native polyacrylamide gels. <i>Journal of Inherited Metabolic Disease</i> , 2011, 34, 741-747.	1.7	21
20	Differential proteomic profiling unveils new molecular mechanisms associated with mitochondrial complex III deficiency. <i>Journal of Proteomics</i> , 2015, 113, 38-56.	1.2	21
21	Evidence of a wide spectrum of cardiac involvement due to ACAD9 mutations: Report on nine patients. <i>Molecular Genetics and Metabolism</i> , 2016, 118, 185-189.	0.5	21
22	Clinical implementation of gene panel testing for lysosomal storage diseases. <i>Molecular Genetics & Genomic Medicine</i> , 2019, 7, e00527.	0.6	18
23	Defining the Pathogenesis of the Human Atp12p W94R Mutation Using a <i>Saccharomyces cerevisiae</i> Yeast Model. <i>Journal of Biological Chemistry</i> , 2010, 285, 4099-4109.	1.6	17
24	Fluorescence imaging of mitochondria in cultured skin fibroblasts: a useful method for the detection of oxidative phosphorylation defects. <i>Pediatric Research</i> , 2012, 72, 232-240.	1.1	16
25	Alteration of structure and function of ATP synthase and cytochrome c oxidase by lack of Fo-a and Cox3 subunits caused by mitochondrial DNA 9205delTA mutation. <i>Biochemical Journal</i> , 2015, 466, 601-611.	1.7	16
26	Accurate and comprehensive analysis of single nucleotide variants and large deletions of the human mitochondrial genome in DNA and single cells. <i>European Journal of Human Genetics</i> , 2017, 25, 1229-1236.	1.4	16
27	A Bumpy Ride on the Diagnostic Bench of Massive Parallel Sequencing, the Case of the Mitochondrial Genome. <i>PLoS ONE</i> , 2014, 9, e112950.	1.1	13
28	Rare genetic variants potentially involved in ovarian hyperstimulation syndrome. <i>Journal of Assisted Reproduction and Genetics</i> , 2019, 36, 491-497.	1.2	12
29	Biallelic mutations in RTTN are associated with microcephaly, short stature and a wide range of brain malformations. <i>European Journal of Medical Genetics</i> , 2018, 61, 733-737.	0.7	11
30	Reliable and Sensitive Detection of Fragile X (Expanded) Alleles in Clinical Prenatal DNA Samples with a Fast Turnaround Time. <i>Journal of Molecular Diagnostics</i> , 2012, 14, 560-568.	1.2	10
31	Mitochondrial encephalomyopathy with cytochrome c oxidase deficiency caused by a novel mutation in the MTCO1 gene. <i>Mitochondrion</i> , 2014, 17, 101-105.	1.6	10
32	Megaconial muscular dystrophy caused by mitochondrial membrane homeostasis defect, new insights from skeletal and heart muscle analyses. <i>Mitochondrion</i> , 2016, 27, 32-38.	1.6	9
33	Mutations in the X-linked pyruvate dehydrogenase (E1) β subunit gene (PDHA1) in patients with a pyruvate dehydrogenase complex deficiency. <i>Human Mutation</i> , 2000, 15, 209.	1.1	9
34	Leigh syndrome followed by parkinsonism in an adult with homozygous c.626C>T mutation in <i>MTFMT</i> . <i>Neurology: Genetics</i> , 2018, 4, e298.	0.9	6
35	Clinical Heterogeneity in MT-ATP6 Pathogenic Variants: Same Genotypeâ€”Different Onset. <i>Cells</i> , 2022, 11, 489.	1.8	6
36	Translation of <i>MT-ATP6</i> pathogenic variants reveals distinct regulatory consequences from the co-translational quality control of mitochondrial protein synthesis. <i>Human Molecular Genetics</i> , 2022, 31, 1230-1241.	1.4	5

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37	Polyneuropathy in a young Belgian patient: A novel heterozygous mutation in the <i>WNK1/HSN2</i> gene. <i>Neurology: Genetics</i> , 2016, 2, e42.	0.9	3
38	Clinical variability in neurohepatic syndrome due to combined mitochondrial DNA depletion and Gaucher disease. <i>Molecular Genetics and Metabolism Reports</i> , 2014, 1, 223-231.	0.4	2
39	Mild Leber hereditary optic neuropathy (LHON) in a Western European family due to the rare Asian m.14502T>C variant in the <i>MT-ND6</i> gene. <i>Ophthalmic Genetics</i> , 2021, 42, 440-445.	0.5	2
40	Near-Infrared Spectroscopy Screening to Allow Detection of Pathogenic Mitochondrial DNA Variants in Individuals with Unexplained Abnormal Fatigue: A Preliminary Study. <i>Applied Spectroscopy</i> , 2018, 72, 715-724.	1.2	1
41	Mild myopathic phenotype in a patient with homozygous c.416C > T mutation in gene. <i>Acta Myologica</i> , 2020, 39, 94-97.	1.5	1
42	Frequency of Participation in External Quality Assessment Programs Focused on Rare Diseases: Belgian Guidelines for Human Genetics Centers. <i>JMIR Medical Informatics</i> , 2021, 9, e27980.	1.3	0