Sara Seneca

List of Publications by Year
in descending order

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42 papers	1,239 citations	394421 19 h-index	377865 34 g-index
44 all docs	44 docs citations	44 times ranked	2266 citing authors

#	Article	IF	Citations
1	Translation of MT-ATP6 pathogenic variants reveals distinct regulatory consequences from the co-translational quality control of mitochondrial protein synthesis. Human Molecular Genetics, 2022, 31, 1230-1241.	2.9	5
2	Clinical Heterogeneity in MT-ATP6 Pathogenic Variants: Same Genotypeâ€"Different Onset. Cells, 2022, 11, 489.	4.1	6
3	Mild Leber hereditary optic neuropathy (LHON) in a Western European family due to the rare Asian m.14502T>C variant in the MT-ND6 gene. Ophthalmic Genetics, 2021, 42, 440-445.	1.2	2
4	Frequency of Participation in External Quality Assessment Programs Focused on Rare Diseases: Belgian Guidelines for Human Genetics Centers. JMIR Medical Informatics, 2021, 9, e27980.	2.6	0
5	Mild myopathic phenotype in a patient with homozygous c.416C > T mutation in gene. Acta Myologica, 2020, 39, 94-97.	1.5	1
6	TANGO2 deficiency as a cause of neurodevelopmental delay with indirect effects on mitochondrial energy metabolism. Journal of Inherited Metabolic Disease, 2019, 42, 898-908.	3.6	32
7	Clinical implementation of gene panel testing for lysosomal storage diseases. Molecular Genetics & Genomic Medicine, 2019, 7, e00527.	1.2	18
8	Rare genetic variants potentially involved in ovarian hyperstimulation syndrome. Journal of Assisted Reproduction and Genetics, 2019, 36, 491-497.	2.5	12
9	Near-Infrared Spectroscopy Screening to Allow Detection of Pathogenic Mitochondrial DNA Variants in Individuals with Unexplained Abnormal Fatigue: A Preliminary Study. Applied Spectroscopy, 2018, 72, 715-724.	2.2	1
10	Leigh syndrome followed by parkinsonism in an adult with homozygous c.626C>T mutation in MTFMT. Neurology: Genetics, 2018, 4, e298.	1.9	6
11	Efficient CRISPR/Cas9-mediated editing of trinucleotide repeat expansion in myotonic dystrophy patient-derived iPS and myogenic cells. Nucleic Acids Research, 2018, 46, 8275-8298.	14.5	78
12	Clinical, biochemical and genetic spectrum of 70 patients with ACAD9 deficiency: is riboflavin supplementation effective?. Orphanet Journal of Rare Diseases, 2018, 13, 120.	2.7	61
13	Biallelic mutations in RTTN are associated with microcephaly, short stature and a wide range of brain malformations. European Journal of Medical Genetics, 2018, 61, 733-737.	1.3	11
14	Random Mutagenesis, Clonal Events, and Embryonic or Somatic Origin Determine the mtDNA Variant Type and Load in Human Pluripotent StemÂCells. Stem Cell Reports, 2018, 11, 102-114.	4.8	23
15	New insights into the phenotype of FARS2 deficiency. Molecular Genetics and Metabolism, 2017, 122, 172-181.	1.1	38
16	Accurate and comprehensive analysis of single nucleotide variants and large deletions of the human mitochondrial genome in DNA and single cells. European Journal of Human Genetics, 2017, 25, 1229-1236.	2.8	16
17	Sertoli Cell-Only Syndrome: Behind the Genetic Scenes. BioMed Research International, 2016, 2016, 1-7.	1.9	22
18	Biallelic Mutations in TMEM126B Cause Severe Complex I Deficiency with a Variable Clinical Phenotype. American Journal of Human Genetics, 2016, 99, 217-227.	6.2	57

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19	GTF2E2 Mutations Destabilize the General Transcription Factor Complex TFIIE in Individuals with DNA Repair-Proficient Trichothiodystrophy. American Journal of Human Genetics, 2016, 98, 627-642.	6.2	49
20	Evidence of a wide spectrum of cardiac involvement due to ACAD9 mutations: Report on nine patients. Molecular Genetics and Metabolism, 2016, 118, 185-189.	1.1	21
21	Polyneuropathy in a young Belgian patient: A novel heterozygous mutation in the WNK1/HSN2 gene. Neurology: Genetics, 2016, 2, e42.	1.9	3
22	Megaconial muscular dystrophy caused by mitochondrial membrane homeostasis defect, new insights from skeletal and heart muscle analyses. Mitochondrion, 2016, 27, 32-38.	3.4	9
23	Clinical, biochemical, and genetic spectrum of seven patients with NFU1 deficiency. Frontiers in Genetics, 2015, 06, 123.	2.3	81
24	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. Biochemical Journal, 2015, 466, 601-611.	3.7	16
25	Mutation of the ironâ€sulfur cluster assembly gene IBA57 causes fatal infantile leukodystrophy. Journal of Inherited Metabolic Disease, 2015, 38, 1147-1153.	3.6	43
26	Analysis of the whole mitochondrial genome: translation of the Ion Torrent Personal Genome Machine system to the diagnostic bench?. European Journal of Human Genetics, 2015, 23, 41-48.	2.8	33
27	Differential proteomic profiling unveils new molecular mechanisms associated with mitochondrial complex III deficiency. Journal of Proteomics, 2015, 113, 38-56.	2.4	21
28	Genetic causes of male infertility. Annales D'Endocrinologie, 2014, 75, 109-111.	1.4	23
29	Mitochondrial encephalomyopathy with cytochrome c oxidase deficiency caused by a novel mutation in the MTCO1 gene. Mitochondrion, 2014, 17, 101-105.	3.4	10
30	Clinical variability in neurohepatic syndrome due to combined mitochondrial DNA depletion and Gaucher disease. Molecular Genetics and Metabolism Reports, 2014, 1, 223-231.	1.1	2
31	A Bumpy Ride on the Diagnostic Bench of Massive Parallel Sequencing, the Case of the Mitochondrial Genome. PLoS ONE, 2014, 9, e112950.	2.5	13
32	Fluorescence imaging of mitochondria in cultured skin fibroblasts: a useful method for the detection of oxidative phosphorylation defects. Pediatric Research, 2012, 72, 232-240.	2.3	16
33	Reliable and Sensitive Detection of Fragile X (Expanded) Alleles in Clinical Prenatal DNA Samples with a Fast Turnaround Time. Journal of Molecular Diagnostics, 2012, 14, 560-568.	2.8	10
34	Complex III staining in blue native polyacrylamide gels. Journal of Inherited Metabolic Disease, 2011, 34, 741-747.	3.6	21
35	Defining the Pathogenesis of the Human Atp12p W94R Mutation Using a Saccharomyces cerevisiae Yeast Model. Journal of Biological Chemistry, 2010, 285, 4099-4109.	3.4	17
36	Experience and outcome of 3 years of a European EQA scheme for genetic testing of the spinocerebellar ataxias. European Journal of Human Genetics, 2008, 16, 913-920.	2.8	23

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37	Early onset Huntington disease: a neuronal degeneration syndrome. European Journal of Pediatrics, 2004, 163, 717-721.	2.7	48
38	Blue Native Polyacrylamide Gel Electrophoresis: A Powerful Tool in Diagnosis of Oxidative Phosphorylation Defects. Pediatric Research, 2001, 50, 658-665.	2.3	117
39	Mutations in the X-linked pyruvate dehydrogenase (E1) ? subunit gene (PDHA1) in patients with a pyruvate dehydrogenase complex deficiency. Human Mutation, 2000, 15, 209-219.	2.5	191
40	Mutations in the X-linked pyruvate dehydrogenase (E1) Î± subunit gene (PDHA1) in patients with a pyruvate dehydrogenase complex deficiency. Human Mutation, 2000, 15, 209.	2.5	9
41	Pearson marrow pancreas syndrome: a molecular study and clinical management. Clinical Genetics, 1997, 51, 338-342.	2.0	37
42	Mutation analysis of the pyruvate dehydrogenase E1Î± gene in eight patients with a pyruvate dehydrogenase complex deficiency. , 1996, 7, 46-51.		36