Julie Steffann

List of Publications by Year
in descending order

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Version: 2024-02-01

38 papers	1,699 citations	279798 23 h-index	315739 38 g-index
39 all docs	39 docs citations	39 times ranked	2180 citing authors

#	Article	IF	Citations
1	Segregation at three loci explains familial and population risk in Hirschsprung disease. Nature Genetics, 2002, 31, 89-93.	21.4	269
2	MECP2 is highly mutated in X-linked mental retardation. Human Molecular Genetics, 2001, 10, 941-946.	2.9	238
3	Large-Scale Deletions and SMADIP1 Truncating Mutations in Syndromic Hirschsprung Disease with Involvement of Midline Structures. American Journal of Human Genetics, 2001, 69, 1370-1377.	6.2	105
4	Segregation of mtDNA throughout human embryofetal development: m.3243A>G as a model system. Human Mutation, 2011, 32, 116-125.	2.5	103
5	Preimplantation genetic diagnosis: State of the art. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2009, 145, 9-13.	1.1	94
6	Insight intoIKBKG/NEMOLocus: Report of New Mutations and Complex Genomic Rearrangements Leading to Incontinentia Pigmenti Disease. Human Mutation, 2014, 35, 165-177.	2.5	74
7	Mutation dependance of the mitochondrial DNA copy number in the first stages of human embryogenesis. Human Molecular Genetics, 2013, 22, 1867-1872.	2.9	72
8	Genotyping microsatellite DNA markers at putative disease loci in inbred/multiplex families with respiratory chain complexÂI deficiency allows rapid identification of a novel nonsense mutation (IVS1nt â^'1) in the NDUFS4 gene in Leigh syndrome. Human Genetics, 2003, 112, 563-566.	3.8	54
9	NDUFS4 mutations cause Leigh syndrome with predominant brainstem involvement. Molecular Genetics and Metabolism, 2009, 97, 185-189.	1.1	54
10	Single cell co-amplification of polymorphic markers for the indirect preimplantation genetic diagnosis of hemophiliaï¿½A, X-linked adrenoleukodystrophy, X-linked hydrocephalus and incontinentia pigmenti loci on Xq28. Human Genetics, 2004, 114, 298-305.	3.8	50
11	After the Storm â€" A Responsible Path for Genome Editing. New England Journal of Medicine, 2019, 380, 897-899.	27.0	50
12	A novel PCR approach for prenatal detection of the common NEMO rearrangement in incontinentia pigmenti. Prenatal Diagnosis, 2004, 24, 384-388.	2.3	45
13	Fetal DNA in maternal serum: does it persist after pregnancy?. Human Genetics, 2003, 113, 76-79.	3.8	43
14	Recurrent KIF2A mutations are responsible for classic lissencephaly. Neurogenetics, 2017, 18, 73-79.	1.4	41
15	A novel mutation of the NDUFS7 gene leads to activation of a cryptic exon and impaired assembly of mitochondrial complex I in a patient with Leigh syndrome. Molecular Genetics and Metabolism, 2007, 92, 104-108.	1.1	40
16	Pre-implantation genetic diagnosis in pulmonary arterial hypertension due toBMPR2mutation: Figure 1â€". European Respiratory Journal, 2012, 39, 1534-1535.	6.7	35
17	Five yearsâ€™ experience of preimplantation genetic diagnosis in the Parisian Center: outcome of the first 441 started cycles. Fertility and Sterility, 2007, 87, 60-73.	1.0	34
18	Structural insights on pathogenic effects of novel mutations causing pyruvate carboxylase deficiency. Human Mutation, 2009, 30, 734-740.	2.5	34

#	Article	IF	Citations
19	Poor Correlations in the Levels of Pathogenic Mitochondrial DNA Mutations in Polar Bodies versus Oocytes and Blastomeres in Humans. American Journal of Human Genetics, 2011, 88, 494-498.	6.2	34
20	Single cell quantification of the 8993T>G NARP mitochondrial DNA mutation by fluorescent PCR. Molecular Genetics and Metabolism, 2005, 84, 289-292.	1.1	33
21	Could Failure in Preimplantation Genetic Diagnosis Justify Editing the Human Embryo Genome?. Cell Stem Cell, 2018, 22, 481-482.	11.1	33
22	CFTR p.Arg117His associated with CBAVD and other CFTR-related disorders. Journal of Medical Genetics, 2013, 50, 220-227.	3.2	31
23	Data from Artificial Models of Mitochondrial DNA Disorders Are Not Always Applicable to Humans. Cell Reports, 2014, 7, 933-934.	6.4	23
24	Clinical Utility Gene Card for: incontinentia pigmenti. European Journal of Human Genetics, 2013, 21, 792-792.	2.8	20
25	Improved single-cell protocol for preimplantation genetic diagnosis of spinal muscular atrophy. Fertility and Sterility, 2005, 84, 734-739.	1.0	17
26	Lack of interaction between NEMO and SHARPIN impairs linear ubiquitination and NF-Î²B activation and leads to incontinentia pigmenti. Journal of Allergy and Clinical Immunology, 2017, 140, 1671-1682.e2.	2.9	13
27	Segregation of mitochondrial DNA mutations in the human placenta: implication for prenatal diagnosis of mtDNA disorders. Journal of Medical Genetics, 2018, 55, 131-136.	3.2	11
28	<scp>OTC</scp> deficiency in females: Phenotypeâ€genotype correlation based on a 130â€family cohort. Journal of Inherited Metabolic Disease, 2021, 44, 1235-1247.	3.6	9
29	Single-sperm analysis for recurrence risk assessment of spinal muscular atrophy. European Journal of Human Genetics, 2010, 18, 505-508.	2.8	7
30	A novel recurrent LIS1 splice site mutation in classic lissencephaly. American Journal of Medical Genetics, Part A, 2017, 173, 561-564.	1.2	6
31	Parental mosaicism is a pitfall in preimplantation genetic diagnosis of dominant disorders. European Journal of Human Genetics, 2014, 22, 711-712.	2.8	5
32	No correlation between mtDNA amount and methylation levels at the CpG island of POLG exon 2 in wild-type and mutant human differentiated cells. Journal of Medical Genetics, 2017, 54, 324-329.	3.2	5
33	Pitfalls in molecular diagnosis of Friedreich ataxia. European Journal of Medical Genetics, 2018, 61, 455-458.	1.3	5
34	A retrospective study on the efficacy of prenatal diagnosis for pregnancies at risk of mitochondrial DNA disorders. Genetics in Medicine, 2021, 23, 720-731.	2.4	5
35	Second biopsy for embryos with inconclusive results after preimplantation genetic testing: Impact on pregnancy outcomes. Journal of Gynecology Obstetrics and Human Reproduction, 2022, 51, 102436.	1.3	3
36	Improving post-natal detection of mitochondrial DNA mutations. Expert Review of Molecular Diagnostics, 2020, 20, 1003-1008.	3.1	2

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37	Genome Editing and Dialogic Responsibility: â€œWhat's in a Name?â€• American Journal of Bioethics, 2015, 15, 54-57.	0.9	1
38	FaisabilitÃ© et incertitude du diagnostic prÃ©implantatoire appliquÃ© aux mutations de l'ADN mitochondrial. Revue Francophone Des Laboratoires, 2018, 2018, 58-64.	0.0	0