Julie Steffann

List of Publications by Year
in descending order

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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	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
2	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
3	<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
4	Improving post-natal detection of mitochondrial DNA mutations. Expert Review of Molecular Diagnostics, 2020, 20, 1003-1008.	3.1	2
5	After the Storm â€" A Responsible Path for Genome Editing. New England Journal of Medicine, 2019, 380, 897-899.	27.0	50
6	Could Failure in Preimplantation Genetic Diagnosis Justify Editing the Human Embryo Genome?. Cell Stem Cell, 2018, 22, 481-482.	11.1	33
7	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
8	Pitfalls in molecular diagnosis of Friedreich ataxia. European Journal of Medical Genetics, 2018, 61, 455-458.	1.3	5
9	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
10	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
11	Lack of interaction between NEMO and SHARPIN impairs linear ubiquitination and NF-Îº B activation and leads to incontinentia pigmenti. Journal of Allergy and Clinical Immunology, 2017, 140, 1671-1682.e2.	2.9	13
12	A novel recurrent LIS1 splice site mutation in classic lissencephaly. American Journal of Medical Genetics, Part A, 2017, 173, 561-564.	1.2	6
13	Recurrent KIF2A mutations are responsible for classic lissencephaly. Neurogenetics, 2017, 18, 73-79.	1.4	41
14	Genome Editing and Dialogic Responsibility: â€œWhat's in a Name?â€• American Journal of Bioethics, 2015, 15, 54-57.	0.9	1
15	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
16	Parental mosaicism is a pitfall in preimplantation genetic diagnosis of dominant disorders. European Journal of Human Genetics, 2014, 22, 711-712.	2.8	5
17	Data from Artificial Models of Mitochondrial DNA Disorders Are Not Always Applicable to Humans. Cell Reports, 2014, 7, 933-934.	6.4	23
18	CFTR p.Arg117His associated with CBAVD and other CFTR-related disorders. Journal of Medical Genetics, 2013, 50, 220-227.	3.2	31

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19	Clinical Utility Gene Card for: incontinentia pigmenti. European Journal of Human Genetics, 2013, 21, 792-792.	2.8	20
20	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
21	Pre-implantation genetic diagnosis in pulmonary arterial hypertension due toBMPR2mutation: Figure 1â€“. European Respiratory Journal, 2012, 39, 1534-1535.	6.7	35
22	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
23	Segregation of mtDNA throughout human embryofetal development: m.3243A>G as a model system. Human Mutation, 2011, 32, 116-125.	2.5	103
24	Single-sperm analysis for recurrence risk assessment of spinal muscular atrophy. European Journal of Human Genetics, 2010, 18, 505-508.	2.8	7
25	Structural insights on pathogenic effects of novel mutations causing pyruvate carboxylase deficiency. Human Mutation, 2009, 30, 734-740.	2.5	34
26	Preimplantation genetic diagnosis: State of the art. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2009, 145, 9-13.	1.1	94
27	NDUFS4 mutations cause Leigh syndrome with predominant brainstem involvement. Molecular Genetics and Metabolism, 2009, 97, 185-189.	1.1	54
28	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
29	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
30	Improved single-cell protocol for preimplantation genetic diagnosis of spinal muscular atrophy. Fertility and Sterility, 2005, 84, 734-739.	1.0	17
31	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
32	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
33	A novel PCR approach for prenatal detection of the common NEMO rearrangement in incontinentia pigmenti. Prenatal Diagnosis, 2004, 24, 384-388.	2.3	45
34	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
35	Fetal DNA in maternal serum: does it persist after pregnancy?. Human Genetics, 2003, 113, 76-79.	3.8	43
36	Segregation at three loci explains familial and population risk in Hirschsprung disease. Nature Genetics, 2002, 31, 89-93.	21.4	269

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37	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
38	MECP2 is highly mutated in X-linked mental retardation. Human Molecular Genetics, 2001, 10, 941-946.	2.9	238