

Stacey Hume

List of Publications by Year in descending order

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Version: 2024-02-01

12
papers

171
citations

1163117

8
h-index

1281871

11
g-index

13
all docs

13
docs citations

13
times ranked

478
citing authors

#	ARTICLE	IF	CITATIONS
1	Methodology for clinical genotyping of CYP2D6 and CYP2C19. <i>Translational Psychiatry</i> , 2021, 11, 596.	4.8	15
2	MITO-FIND: A study in 390 patients to determine a diagnostic strategy for mitochondrial disease. <i>Molecular Genetics and Metabolism</i> , 2020, 131, 66-82.	1.1	17
3	The novel p.Ser263Phe mutation in the human high-affinity choline transporter 1 (CHT1/ <i>SLC5A7</i>) causes a lethal form of fetal akinesia syndrome. <i>Human Mutation</i> , 2019, 40, 1676-1683.	2.5	14
4	Hybrid gel electrophoresis using skin fibroblasts to aid in diagnosing mitochondrial disease. <i>Neurology: Genetics</i> , 2019, 5, e336.	1.9	4
5	CCMG practice guideline: laboratory guidelines for next-generation sequencing. <i>Journal of Medical Genetics</i> , 2019, 56, 792-800.	3.2	29
6	Data sharing as a national quality improvement program: reporting on BRCA1 and BRCA2 variant-interpretation comparisons through the Canadian Open Genetics Repository (COGR). <i>Genetics in Medicine</i> , 2018, 20, 294-302.	2.4	27
7	Plasma-derived cell-free mitochondrial DNA: A novel non-invasive methodology to identify mitochondrial DNA haplogroups in humans. <i>Molecular Genetics and Metabolism</i> , 2018, 125, 332-337.	1.1	20
8	Mitochondrial Replacement Therapy: The Road to the Clinic in Canada. <i>Journal of Obstetrics and Gynaecology Canada</i> , 2017, 39, 916-918.	0.7	10
9	Assessing the cost of implementing the 2011 Society of Obstetricians and Gynecologists of Canada and Canadian College of Medical Genetics practice guidelines on the detection of fetal aneuploidies. <i>Prenatal Diagnosis</i> , 2017, 37, 916-923.	2.3	2
10	Exome Sequencing Identifies a Novel Variant in ACTC1 Associated With Familial Atrial Septal Defect. <i>Canadian Journal of Cardiology</i> , 2014, 30, 181-187.	1.7	30
11	Discrepant HIV results resolved by human DNA testing. <i>Journal of Clinical Virology</i> , 2014, 61, 311-312.	3.1	1
12	An automated hand-held CMOS-based instrument for hand-held mutation detection via electrophoresis. , 2014, , .		2