

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