

AmÃ©lie C Pinard

List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/8082504/publications.pdf>

Version: 2024-02-01

18
papers

890
citations

840585

11
h-index

887953

17
g-index

20
all docs

20
docs citations

20
times ranked

2042
citing authors

#	ARTICLE	IF	CITATIONS
1	Genetics of Thoracic and Abdominal Aortic Diseases. <i>Circulation Research</i> , 2019, 124, 588-606.	2.0	253
2	Comparative genomics of emerging pathogens in the <i>Candida glabrata</i> clade. <i>BMC Genomics</i> , 2013, 14, 623.	1.2	174
3	UMDâ€Predictor: A Highâ€Throughput Sequencing Compliant System for Pathogenicity Prediction of any Human cDNA Substitution. <i>Human Mutation</i> , 2016, 37, 439-446.	1.1	104
4	A genome-wide approach reveals novel imprinted genes expressed in the human placenta. <i>Epigenetics</i> , 2012, 7, 1079-1090.	1.3	81
5	LTBP3 Pathogenic Variants Predispose Individuals to Thoracic Aortic Aneurysms and Dissections. <i>American Journal of Human Genetics</i> , 2018, 102, 706-712.	2.6	51
6	Piezo1 is required for outflow tract and aortic valve development.. <i>Journal of Molecular and Cellular Cardiology</i> , 2020, 143, 51-62.	0.9	44
7	The revised ghent nosology; reclassifying isolated ectopia lentis. <i>Clinical Genetics</i> , 2015, 87, 284-287.	1.0	41
8	The pleiotropy associated with de novo variants in CHD4, CNOT3, and SETD5 extends to moyamoya angiopathy. <i>Genetics in Medicine</i> , 2020, 22, 427-431.	1.1	34
9	<i>DIAPH1</i> Variants in Nonâ€East Asian Patients With Sporadic Moyamoya Disease. <i>JAMA Neurology</i> , 2021, 78, 993.	4.5	33
10	Association of De Novo <i>RNF213</i> Variants With Childhood Onset Moyamoya Disease and Diffuse Occlusive Vasculopathy. <i>Neurology</i> , 2021, 96, e1783-e1791.	1.5	21
11	Rare deleterious variants of <i>NOTCH1</i>, <i>GATA4</i>, <i>SMAD6</i>, and <i>ROBO4</i> are enriched in BAV with early onset complications but not in BAV with heritable thoracic aortic disease. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1406.	0.6	17
12	Update on the genetic risk for thoracic aortic aneurysms and acute aortic dissections: implications for clinical care. <i>Journal of Cardiovascular Surgery</i> , 2021, 62, 203-210.	0.3	11
13	A genome-wide search for new imprinted genes in the human placenta identifies DSCAM as the first imprinted gene on chromosome 21. <i>European Journal of Human Genetics</i> , 2019, 27, 49-60.	1.4	8
14	Actionable Genes, Core Databases, and Locus-Specific Databases. <i>Human Mutation</i> , 2016, 37, 1299-1307.	1.1	6
15	WES/WGS Reporting of Mutations from Cardiovascular â€Actionableâ€Genes in Clinical Practice: A Key Role for UMD Knowledgebases in the Era of Big Databases. <i>Human Mutation</i> , 2016, 37, 1308-1317.	1.1	5
16	An uncommon cause of tricuspid regurgitation: three-dimensional echocardiographic incremental value, surgical and genetic insights. <i>European Journal of Cardio-thoracic Surgery</i> , 2016, 50, 180-182.	0.6	1
17	Preventing Acute Aortic Dissections: The Power of Familial Screening and Risk Assessment. <i>Journal of the American Heart Association</i> , 2022, 11, e025441.	1.6	1
18	Analysis of HOXB1 gene in a cohort of patients with sporadic ventricular septal defect. <i>Molecular Biology Reports</i> , 2018, 45, 1507-1513.	1.0	0