

# Christine E Seidman

## List of Publications by Year in Descending Order

**Source:** <https://exaly.com/author-pdf/660296/christine-e-seidman-publications-by-year.pdf>

**Version:** 2024-04-23

This document has been generated based on the publications and citations recorded by exaly.com. For the latest version of this publication list, visit the link given above.

The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

207  
papers

24,364  
citations

73  
h-index

155  
g-index

224  
ext. papers

29,468  
ext. citations

16.4  
avg, IF

6.3  
L-index

#	Paper	IF	Citations
207	Genetic determinants of telomere length from 109,122 ancestrally diverse whole-genome sequences in TOPMed.. <i>Cell Genomics</i> , <b>2022</b> , 2, 100084-100084		1
206	Cell cycle defects underlie childhood-onset cardiomyopathy associated with Noonan syndrome.. <i>IScience</i> , <b>2022</b> , 25, 103596	6.1	0
205	Genome-Wide De Novo Variants in Congenital Heart Disease Are Not Associated With Maternal Diabetes or Obesity.. <i>Circulation Genomic and Precision Medicine</i> , <b>2022</b> , CIRCGEN121003500	5.2	0
204	Transcription factor protein interactomes reveal genetic determinants in heart disease.. <i>Cell</i> , <b>2022</b> ,	56.2	3
203	Neither cardiac mitochondrial DNA variation or copy number contribute to congenital heart disease risk.. <i>American Journal of Human Genetics</i> , <b>2022</b> ,	11	1
202	Genetics of cancer therapy-associated cardiotoxicity.. <i>Journal of Molecular and Cellular Cardiology</i> , <b>2022</b> ,	5.8	2
201	Engineering a living cardiac pump on a chip using high-precision fabrication.. <i>Science Advances</i> , <b>2022</b> , 8, eabm3791	14.3	4
200	Population Prevalence of Premature Truncating Variants in Plakophilin-2 and Association With Arrhythmogenic Right Ventricular Cardiomyopathy: a UK Biobank Analysis.. <i>Circulation Genomic and Precision Medicine</i> , <b>2022</b> , 101161CIRCGEN121003507	5.2	0
199	An ancient founder mutation located between and is responsible for increased microtia risk in Amerindigenous populations.. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , <b>2022</b> , 119, e2203928119	11.5	0
198	Associations Between Female Sex, Sarcomere Variants, and Clinical Outcomes in Hypertrophic Cardiomyopathy. <i>Circulation Genomic and Precision Medicine</i> , <b>2021</b> , 14, e003062	5.2	10
197	Targeted therapies in genetic dilated and hypertrophic cardiomyopathies: From molecular mechanisms to therapeutic targets.. <i>European Journal of Heart Failure</i> , <b>2021</b> ,	12.3	2
196	Plakophilin-2 truncating variants impair cardiac contractility by disrupting sarcomere stability and organization. <i>Science Advances</i> , <b>2021</b> , 7, eabh3995	14.3	1
195	Mechanisms of Congenital Heart Disease Caused by NAA15 Haploinsufficiency. <i>Circulation Research</i> , <b>2021</b> , 128, 1156-1169	15.7	2
194	Genetic and Phenotypic Landscape of Peripartum Cardiomyopathy. <i>Circulation</i> , <b>2021</b> , 143, 1852-1862	16.7	11
193	Isolation of Nuclei from Mammalian Cells and Tissues for Single-Nucleus Molecular Profiling. <i>Current Protocols</i> , <b>2021</b> , 1, e132		4
192	Cardiomyocyte Proliferative Capacity Is Restricted in Mice With Mutation. <i>Frontiers in Cardiovascular Medicine</i> , <b>2021</b> , 8, 639148	5.4	3
191	CalTrack: High-Throughput Automated Calcium Transient Analysis in Cardiomyocytes. <i>Circulation Research</i> , <b>2021</b> , 129, 326-341	15.7	8

190	Association of Damaging Variants in Genes With Increased Cancer Risk Among Patients With Congenital Heart Disease. <i>JAMA Cardiology</i> , <b>2021</b> , 6, 457-462	16.2	12
189	Modeling Human TBX5 Haploinsufficiency Predicts Regulatory Networks for Congenital Heart Disease. <i>Developmental Cell</i> , <b>2021</b> , 56, 292-309.e9	10.2	17
188	Probing the subcellular nanostructure of engineered human cardiomyocytes in 3D tissue. <i>Microsystems and Nanoengineering</i> , <b>2021</b> , 7, 10	7.7	1
187	Discordant clinical features of identical hypertrophic cardiomyopathy twins. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , <b>2021</b> , 118,	11.5	5
186	Genomic frontiers in congenital heart disease. <i>Nature Reviews Cardiology</i> , <b>2021</b> ,	14.8	15
185	Contribution of Noncanonical Splice Variants to Truncating Variant Cardiomyopathy. <i>Circulation Genomic and Precision Medicine</i> , <b>2021</b> , 14, e003389	5.2	0
184	Valsartan in early-stage hypertrophic cardiomyopathy: a randomized phase 2 trial. <i>Nature Medicine</i> , <b>2021</b> , 27, 1818-1824	50.5	7
183	Filamin C Cardiomyopathy Variants Cause Protein and Lysosome Accumulation. <i>Circulation Research</i> , <b>2021</b> , 129, 751-766	15.7	2
182	Systems Analysis Implicates WAVE2 Complex in the Pathogenesis of Developmental Left-Sided Obstructive Heart Defects. <i>JACC Basic To Translational Science</i> , <b>2020</b> , 5, 376-386	8.7	2
181	Complement genes contribute sex-biased vulnerability in diverse disorders. <i>Nature</i> , <b>2020</b> , 582, 577-581	50.4	71
180	Congenital Heart Defects Due to Missense Variants. <i>Circulation Genomic and Precision Medicine</i> , <b>2020</b> , 13, e002843	5.2	3
179	Expanding the clinical and genetic spectrum of ALPK3 variants: Phenotypes identified in pediatric cardiomyopathy patients and adults with heterozygous variants. <i>American Heart Journal</i> , <b>2020</b> , 225, 1084-1097	4.9	7
178	Hypertrophic cardiomyopathy in myosin-binding protein C () Icelandic founder mutation carriers. <i>Open Heart</i> , <b>2020</b> , 7, e001220	3	2
177	Genomic analyses implicate noncoding de novo variants in congenital heart disease. <i>Nature Genetics</i> , <b>2020</b> , 52, 769-777	36.3	33
176	De Novo Damaging Variants, Clinical Phenotypes, and Post-Operative Outcomes in Congenital Heart Disease. <i>Circulation Genomic and Precision Medicine</i> , <b>2020</b> , 13, e002836	5.2	15
175	Myosin Sequestration Regulates Sarcomere Function, Cardiomyocyte Energetics, and Metabolism, Informing the Pathogenesis of Hypertrophic Cardiomyopathy. <i>Circulation</i> , <b>2020</b> , 141, 828-842	16.7	66
174	EM-mosaic detects mosaic point mutations that contribute to congenital heart disease. <i>Genome Medicine</i> , <b>2020</b> , 12, 42	14.4	8
173	BET bromodomain proteins regulate transcriptional reprogramming in genetic dilated cardiomyopathy. <i>JCI Insight</i> , <b>2020</b> , 5,	9.9	12

172	Rare genetic variation at transcription factor binding sites modulates local DNA methylation profiles. <i>PLoS Genetics</i> , <b>2020</b> , 16, e1009189	6	7
171	mutations in hiPSCs inform mechanisms for maldevelopment of the heart, pancreas, and diaphragm. <i>ELife</i> , <b>2020</b> , 9,	8.9	9
170	Marked Up-Regulation of ACE2 in Hearts of Patients With Obstructive Hypertrophic Cardiomyopathy: Implications for SARS-CoV-2-Mediated COVID-19. <i>Mayo Clinic Proceedings</i> , <b>2020</b> , 95, 1354-1368	6.4	31
169	Genetic Cardiomyopathies <b>2020</b> , 77-114		1
168	Scientists on the Spot: Putting a halt to hypertrophic cardiomyopathy. <i>Cardiovascular Research</i> , <b>2020</b> , 116, e42-e43	9.9	
167	Association of Race With Disease Expression and Clinical Outcomes Among Patients With Hypertrophic Cardiomyopathy. <i>JAMA Cardiology</i> , <b>2020</b> , 5, 83-91	16.2	23
166	Cells of the adult human heart. <i>Nature</i> , <b>2020</b> , 588, 466-472	50.4	274
165	Founder Mutation in N Terminus of Cardiac Troponin I Causes Malignant Hypertrophic Cardiomyopathy. <i>Circulation Genomic and Precision Medicine</i> , <b>2020</b> , 13, 444-452	5.2	3
164	Genetic Studies of Hypertrophic Cardiomyopathy in Singaporeans Identify Variants in and That Are Common in Chinese Patients. <i>Circulation Genomic and Precision Medicine</i> , <b>2020</b> , 13, 424-434	5.2	4
163	Abnormal Left-Hemispheric Sulcal Patterns Correlate with Neurodevelopmental Outcomes in Subjects with Single Ventricular Congenital Heart Disease. <i>Cerebral Cortex</i> , <b>2020</b> , 30, 476-487	5.1	11
162	Yin Yang 1 Suppresses Dilated Cardiomyopathy and Cardiac Fibrosis Through Regulation of and. <i>Circulation Research</i> , <b>2019</b> , 125, 834-846	15.7	20
161	SarcTrack. <i>Circulation Research</i> , <b>2019</b> , 124, 1172-1183	15.7	56
160	Hypertrophic cardiomyopathy mutations in dysregulate myosin. <i>Science Translational Medicine</i> , <b>2019</b> , 11,	17.5	73
159	Novel Therapies for Prevention and Early Treatment of Cardiomyopathies. <i>Circulation Research</i> , <b>2019</b> , 124, 1536-1550	15.7	22
158	Histone H2B monoubiquitination regulates heart development via epigenetic control of cilia motility. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , <b>2019</b> , 116, 14049-14054	11.5	17
157	The Translational Landscape of the Human Heart. <i>Cell</i> , <b>2019</b> , 178, 242-260.e29	56.2	210
156	Genetic Variants Associated With Cancer Therapy-Induced Cardiomyopathy. <i>Circulation</i> , <b>2019</b> , 140, 31-41	16.7	110
155	Response by Ho et al to Letter Regarding Article, "Genotype and Lifetime Burden of Disease in Hypertrophic Cardiomyopathy: Insights From the Sarcomeric Human Cardiomyopathy Registry (SHaRe)". <i>Circulation</i> , <b>2019</b> , 139, 1559-1560	16.7	2

154	Activin type II receptor signaling in cardiac aging and heart failure. <i>Science Translational Medicine</i> , <b>2019</b> , 11,	17.5	43
153	Advances in the Genetic Basis and Pathogenesis of Sarcomere Cardiomyopathies. <i>Annual Review of Genomics and Human Genetics</i> , <b>2019</b> , 20, 129-153	9.7	59
152	A gene-centric strategy for identifying disease-causing rare variants in dilated cardiomyopathy. <i>Genetics in Medicine</i> , <b>2019</b> , 21, 133-143	8.1	20
151	Genome sequencing as a first-line genetic test in familial dilated cardiomyopathy. <i>Genetics in Medicine</i> , <b>2019</b> , 21, 650-662	8.1	36
150	Paternal-age-related de novo mutations and risk for five disorders. <i>Nature Communications</i> , <b>2019</b> , 10, 3043	17.4	33
149	De novo and recessive forms of congenital heart disease have distinct genetic and phenotypic landscapes. <i>Nature Communications</i> , <b>2019</b> , 10, 4722	17.4	25
148	The uptake of family screening in hypertrophic cardiomyopathy and an online video intervention to facilitate family communication. <i>Molecular Genetics &amp; Genomic Medicine</i> , <b>2019</b> , 7, e940	2.3	5
147	Precision Medicine in the Management of Dilated Cardiomyopathy: JACC State-of-the-Art Review. <i>Journal of the American College of Cardiology</i> , <b>2019</b> , 74, 2921-2938	15.1	33
146	Genetic Testing and Counseling for Hypertrophic Cardiomyopathy. <i>Cardiology Clinics</i> , <b>2019</b> , 37, 35-43	2.5	8
145	Sex differences in gene expression in response to ischemia in the human left ventricular myocardium. <i>Human Molecular Genetics</i> , <b>2019</b> , 28, 1682-1693	5.6	18
144	Response to Brodehl et al. <i>Genetics in Medicine</i> , <b>2019</b> , 21, 1248-1249	8.1	
143	Human Induced Pluripotent Stem Cell Production and Expansion from Blood using a Non-Integrating Viral Reprogramming Vector. <i>Current Protocols in Molecular Biology</i> , <b>2018</b> , 122, e58	2.9	5
142	Robust identification of mosaic variants in congenital heart disease. <i>Human Genetics</i> , <b>2018</b> , 137, 183-193.6.3		26
141	CRISPR/Cas9-Mediated Fluorescent Tagging of Endogenous Proteins in Human Pluripotent Stem Cells. <i>Current Protocols in Human Genetics</i> , <b>2018</b> , 96, 21.11.1-21.11.20	3.2	30
140	Differentiation and Contractile Analysis of GFP-Sarcomere Reporter hiPSC-Cardiomyocytes. <i>Current Protocols in Human Genetics</i> , <b>2018</b> , 96, 21.12.1-21.12.12	3.2	18
139	Force Generation via $\beta$ Cardiac Myosin, Titin, and $\beta$ Actinin Drives Cardiac Sarcomere Assembly from Cell-Matrix Adhesions. <i>Developmental Cell</i> , <b>2018</b> , 44, 87-96.e5	10.2	75
138	Spatiotemporal Multi-Omics Mapping Generates a Molecular Atlas of the Aortic Valve and Reveals Networks Driving Disease. <i>Circulation</i> , <b>2018</b> , 138, 377-393	16.7	102
137	Genetic Pathogenesis of Hypertrophic and Dilated Cardiomyopathy. <i>Heart Failure Clinics</i> , <b>2018</b> , 14, 139-146	13.6	49

136	Robust identification of deletions in exome and genome sequence data based on clustering of Mendelian errors. <i>Human Mutation</i> , <b>2018</b> , 39, 870-881	4.7	1
135	Genetics of pediatric cardiomyopathies. <i>Progress in Pediatric Cardiology</i> , <b>2018</b> , 49, 18-19	0.4	2
134	An integrated clinical program and crowdsourcing strategy for genomic sequencing and Mendelian disease gene discovery. <i>Npj Genomic Medicine</i> , <b>2018</b> , 3, 21	6.2	15
133	Association of Variants in BAG3 With Cardiomyopathy Outcomes in African American Individuals. <i>JAMA Cardiology</i> , <b>2018</b> , 3, 929-938	16.2	30
132	ViroFind: A novel target-enrichment deep-sequencing platform reveals a complex JC virus population in the brain of PML patients. <i>PLoS ONE</i> , <b>2018</b> , 13, e0186945	3.7	16
131	In vivo and In vitro methods to identify DNA sequence variants that alter RNA Splicing. <i>Current Protocols in Human Genetics</i> , <b>2018</b> , 97, e60	3.2	2
130	Cardiac-enriched BAF chromatin-remodeling complex subunit Baf60c regulates gene expression programs essential for heart development and function. <i>Biology Open</i> , <b>2018</b> , 7,	2.2	19
129	Genotype and Lifetime Burden of Disease in Hypertrophic Cardiomyopathy: Insights from the Sarcomeric Human Cardiomyopathy Registry (SHaRe). <i>Circulation</i> , <b>2018</b> , 138, 1387-1398	16.7	210
128	Hierarchical and stage-specific regulation of murine cardiomyocyte maturation by serum response factor. <i>Nature Communications</i> , <b>2018</b> , 9, 3837	17.4	36
127	Multilayer Myocardial Mechanics in Genotype-Positive Left Ventricular Hypertrophy-Negative Patients With Hypertrophic Cardiomyopathy. <i>American Journal of Cardiology</i> , <b>2018</b> , 122, 1754-1760	3	11
126	Molecular Genetics of Lidocaine-Containing Cardioplegia in the Human Heart During Cardiac Surgery. <i>Annals of Thoracic Surgery</i> , <b>2018</b> , 106, 1379-1387	2.7	6
125	The Transcriptional Signature of Growth in Human Fetal Aortic Valve Development. <i>Annals of Thoracic Surgery</i> , <b>2018</b> , 106, 1834-1840	2.7	5
124	Telomere shortening is a hallmark of genetic cardiomyopathies. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , <b>2018</b> , 115, 9276-9281	11.5	30
123	Dynamic Cellular Integration Drives Functional Assembly of the Heart's Pacemaker Complex. <i>Cell Reports</i> , <b>2018</b> , 23, 2283-2291	10.6	11
122	The Long Noncoding RNA Landscape of the Ischemic Human Left Ventricle. <i>Circulation: Cardiovascular Genetics</i> , <b>2017</b> , 10,		20
121	The Burden of Early Phenotypes and the Influence of Wall Thickness in Hypertrophic Cardiomyopathy Mutation Carriers: Findings From the HCMNet Study. <i>JAMA Cardiology</i> , <b>2017</b> , 2, 419-428	16.2	35
120	Macrophages Facilitate Electrical Conduction in the Heart. <i>Cell</i> , <b>2017</b> , 169, 510-522.e20	56.2	438
119	Cardiac Myosin Binding Protein-C Autoantibodies are Potential Early Indicators of Cardiac Dysfunction and Patient Outcome in Acute Coronary Syndrome. <i>JACC Basic To Translational Science</i> , <b>2017</b> , 2, 122-131	8.7	1

118	Bedside Back to Bench: Building Bridges between Basic and Clinical Genomic Research. <i>Cell</i> , <b>2017</b> , 169, 6-12	56.2	81
117	Early remodeling of repolarizing K currents in the MHC mouse model of familial hypertrophic cardiomyopathy. <i>Journal of Molecular and Cellular Cardiology</i> , <b>2017</b> , 103, 93-101	5.8	3
116	Cardiovascular homeostasis dependence on MICU2, a regulatory subunit of the mitochondrial calcium uniporter. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , <b>2017</b> , 114, E9096-E9104	11.5	35
115	Contribution of rare inherited and de novo variants in 2,871 congenital heart disease probands. <i>Nature Genetics</i> , <b>2017</b> , 49, 1593-1601	36.3	348
114	A Comparison of Whole Genome Sequencing to Multigene Panel Testing in Hypertrophic Cardiomyopathy Patients. <i>Circulation: Cardiovascular Genetics</i> , <b>2017</b> , 10,		45
113	Joint analysis of left ventricular expression and circulating plasma levels of Omentin after myocardial ischemia. <i>Cardiovascular Diabetology</i> , <b>2017</b> , 16, 87	8.7	12
112	Fabry Disease in Families With Hypertrophic Cardiomyopathy: Clinical Manifestations in the Classic and Later-Onset Phenotypes. <i>Circulation: Cardiovascular Genetics</i> , <b>2017</b> , 10,		13
111	IL-11 is a crucial determinant of cardiovascular fibrosis. <i>Nature</i> , <b>2017</b> , 552, 110-115	50.4	267
110	Identification of pathogenic gene mutations in and that alter RNA splicing. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , <b>2017</b> , 114, 7689-7694	11.5	42
109	Titin-truncating variants affect heart function in disease cohorts and the general population. <i>Nature Genetics</i> , <b>2017</b> , 49, 46-53	36.3	179
108	Effects of myosin variants on interacting-heads motif explain distinct hypertrophic and dilated cardiomyopathy phenotypes. <i>ELife</i> , <b>2017</b> , 6,	8.9	87
107	A Novel Role for in a Lebanese Family with Congenital Cardiac Defects. <i>Frontiers in Genetics</i> , <b>2017</b> , 8, 217	4.5	4
106	Detection of Cell Proliferation Markers by Immunofluorescence Staining and Microscopy Imaging in Paraffin-Embedded Tissue Sections. <i>Current Protocols in Molecular Biology</i> , <b>2016</b> , 115, 14.25.1-14.25.14	2.9	11
105	Aggregate penetrance of genomic variants for actionable disorders in European and African Americans. <i>Science Translational Medicine</i> , <b>2016</b> , 8, 364ra151	17.5	41
104	Single-Cell Resolution of Temporal Gene Expression during Heart Development. <i>Developmental Cell</i> , <b>2016</b> , 39, 480-490	10.2	231
103	Loss of RNA expression and allele-specific expression associated with congenital heart disease. <i>Nature Communications</i> , <b>2016</b> , 7, 12824	17.4	33
102	Pitx2 modulates a Tbx5-dependent gene regulatory network to maintain atrial rhythm. <i>Science Translational Medicine</i> , <b>2016</b> , 8, 354ra115	17.5	79
101	Preparation of rAAV9 to Overexpress or Knockdown Genes in Mouse Hearts. <i>Journal of Visualized Experiments</i> , <b>2016</b> ,	1.6	4

100	Mutation analysis of the phospholamban gene in 315 South Africans with dilated, hypertrophic, peripartum and arrhythmogenic right ventricular cardiomyopathies. <i>Scientific Reports</i> , <b>2016</b> , 6, 22235	4.9	23
99	The Role of the L-Type Ca Channel in Altered Metabolic Activity in a Murine Model of Hypertrophic Cardiomyopathy. <i>JACC Basic To Translational Science</i> , <b>2016</b> , 1, 61-72	8.7	9
98	A small-molecule inhibitor of sarcomere contractility suppresses hypertrophic cardiomyopathy in mice. <i>Science</i> , <b>2016</b> , 351, 617-21	33.3	282
97	The Muscle-Bound Heart. <i>Cardiac Electrophysiology Clinics</i> , <b>2016</b> , 8, 223-31	1.4	3
96	Shared Genetic Predisposition in Peripartum and Dilated Cardiomyopathies. <i>New England Journal of Medicine</i> , <b>2016</b> , 374, 233-41	59.2	290
95	Molecular profiling of dilated cardiomyopathy that progresses to heart failure. <i>JCI Insight</i> , <b>2016</b> , 1,	9.9	55
94	THSD1 (Thrombospondin Type 1 Domain Containing Protein 1) Mutation in the Pathogenesis of Intracranial Aneurysm and Subarachnoid Hemorrhage. <i>Stroke</i> , <b>2016</b> , 47, 3005-3013	6.7	26
93	Integrative Analysis of PRKAG2 Cardiomyopathy iPS and Microtissue Models Identifies AMPK as a Regulator of Metabolism, Survival, and Fibrosis. <i>Cell Reports</i> , <b>2016</b> , 17, 3292-3304	10.6	51
92	Clinical and Mechanistic Insights Into the Genetics of Cardiomyopathy. <i>Journal of the American College of Cardiology</i> , <b>2016</b> , 68, 2871-2886	15.1	164
91	A Tension-Based Model Distinguishes Hypertrophic versus Dilated Cardiomyopathy. <i>Cell</i> , <b>2016</b> , 165, 1147-1159	15.2	122
90	Cardiac myosin binding protein C regulates postnatal myocyte cytokinesis. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , <b>2015</b> , 112, 9046-51	11.5	28
89	Integrated allelic, transcriptional, and phenomic dissection of the cardiac effects of titin truncations in health and disease. <i>Science Translational Medicine</i> , <b>2015</b> , 7, 270ra6	17.5	267
88	HEART DISEASE. Titin mutations in iPS cells define sarcomere insufficiency as a cause of dilated cardiomyopathy. <i>Science</i> , <b>2015</b> , 349, 982-6	33.3	379
87	Haploinsufficiency of MYBPC3 exacerbates the development of hypertrophic cardiomyopathy in heterozygous mice. <i>Journal of Molecular and Cellular Cardiology</i> , <b>2015</b> , 79, 234-43	5.8	41
86	163 Integrated allelic, transcriptional, and phenotypic dissection of the cardiac effects of titin variation in health and disease. <i>Heart</i> , <b>2015</b> , 101, A93.1-A93	5.1	
85	Phenotype and prognostic correlations of the converter region mutations affecting the $\beta$ myosin heavy chain. <i>Heart</i> , <b>2015</b> , 101, 1047-53	5.1	34
84	NKX2-5 mutations in an inbred consanguineous population: genetic and phenotypic diversity. <i>Scientific Reports</i> , <b>2015</b> , 5, 8848	4.9	32
83	De novo mutations in congenital heart disease with neurodevelopmental and other congenital anomalies. <i>Science</i> , <b>2015</b> , 350, 1262-6	33.3	406



82	Diltiazem treatment for pre-clinical hypertrophic cardiomyopathy sarcomere mutation carriers: a pilot randomized trial to modify disease expression. <i>JACC: Heart Failure</i> , <b>2015</b> , 3, 180-8	7.9	105
81	Results of clinical genetic testing of 2,912 probands with hypertrophic cardiomyopathy: expanded panels offer limited additional sensitivity. <i>Genetics in Medicine</i> , <b>2015</b> , 17, 880-8	8.1	236
80	Genetic Pathways in Congenital Heart Development. <i>FASEB Journal</i> , <b>2015</b> , 29, 215.3	0.9	
79	Increased frequency of de novo copy number variants in congenital heart disease by integrative analysis of single nucleotide polymorphism array and exome sequence data. <i>Circulation Research</i> , <b>2014</b> , 115, 884-896	15.7	158
78	126 Advanced Assessment of Cardiac Morphology and Prediction of Gene Carriage by CMR in Hypertrophic Cardiomyopathy - The HCMNET/UCL Collaboration. <i>Heart</i> , <b>2014</b> , 100, A72-A73	5.1	1
77	Biobank participants' preferences for disclosure of genetic research results: perspectives from the OurGenes, OurHealth, OurCommunity project. <i>Mayo Clinic Proceedings</i> , <b>2014</b> , 89, 738-46	6.4	42
76	Alpha blockade potentiates CPVT therapy in calsequestrin-mutant mice. <i>Heart Rhythm</i> , <b>2014</b> , 11, 1471-96.7		16
75	Nationwide study on hypertrophic cardiomyopathy in Iceland: evidence of a MYBPC3 founder mutation. <i>Circulation</i> , <b>2014</b> , 130, 1158-67	16.7	44
74	Tbx5 is required for avian and Mammalian epicardial formation and coronary vasculogenesis. <i>Circulation Research</i> , <b>2014</b> , 115, 834-44	15.7	23
73	UBQLN2 mutation causing heterogeneous X-linked dominant neurodegeneration. <i>Annals of Neurology</i> , <b>2014</b> , 75, 793-798	9.4	38
72	Targeted and genome-wide sequencing reveal single nucleotide variations impacting specificity of Cas9 in human stem cells. <i>Nature Communications</i> , <b>2014</b> , 5, 5507	17.4	106
71	Myosin heavy chain variant Val606Met causes very mild hypertrophic cardiomyopathy in mice, but exacerbates HCM phenotypes in mice carrying other HCM mutations. <i>Circulation Research</i> , <b>2014</b> , 115, 227-37	15.7	29
70	5RNA-Seq identifies Fhl1 as a genetic modifier in cardiomyopathy. <i>Journal of Clinical Investigation</i> , <b>2014</b> , 124, 1364-70	15.9	44
69	Genetics of congenital heart disease: the glass half empty. <i>Circulation Research</i> , <b>2013</b> , 112, 707-20	15.7	369
68	Effects of losartan on left ventricular hypertrophy and fibrosis in patients with nonobstructive hypertrophic cardiomyopathy. <i>JACC: Heart Failure</i> , <b>2013</b> , 1, 480-7	7.9	75
67	Elevated rates of force development and MgATP binding in F764L and S532P myosin mutations causing dilated cardiomyopathy. <i>Journal of Molecular and Cellular Cardiology</i> , <b>2013</b> , 57, 23-31	5.8	15
66	De novo mutations in histone-modifying genes in congenital heart disease. <i>Nature</i> , <b>2013</b> , 498, 220-3	50.4	591
65	HOXA2 haploinsufficiency in dominant bilateral microtia and hearing loss. <i>Human Mutation</i> , <b>2013</b> , 34, 1347-51	4.7	30

64	The Congenital Heart Disease Genetic Network Study: rationale, design, and early results. <i>Circulation Research</i> , <b>2013</b> , 112, 698-706	15.7	104
63	Subtle abnormalities in contractile function are an early manifestation of sarcomere mutations in dilated cardiomyopathy. <i>Circulation: Cardiovascular Genetics</i> , <b>2012</b> , 5, 503-10		56
62	Truncations of titin causing dilated cardiomyopathy. <i>New England Journal of Medicine</i> , <b>2012</b> , 366, 619-28	59.2	874
61	Hypertrophic cardiomyopathy: translating cellular cross talk into therapeutics. <i>Journal of Cell Biology</i> , <b>2012</b> , 199, 417-21	7.3	34
60	2011 ACCF/AHA guideline for the diagnosis and treatment of hypertrophic cardiomyopathy: executive summary: a report of the American College of Cardiology Foundation/American Heart Association Task Force on Practice Guidelines. <i>Journal of Thoracic and Cardiovascular Surgery</i> , <b>2011</b> , 142, 1303-38	1.5	59
59	2011 ACCF/AHA guideline for the diagnosis and treatment of hypertrophic cardiomyopathy: executive summary: a report of the American College of Cardiology Foundation/American Heart Association Task Force on Practice Guidelines. <i>Circulation</i> , <b>2011</b> , 124, 2761-96	16.7	587
58	Identifying sarcomere gene mutations in hypertrophic cardiomyopathy: a personal history. <i>Circulation Research</i> , <b>2011</b> , 108, 743-50	15.7	167
57	Activation of multiple signaling pathways causes developmental defects in mice with a Noonan syndrome-associated <i>Sos1</i> mutation. <i>Journal of Clinical Investigation</i> , <b>2010</b> , 120, 4353-65	15.9	79
56	Short communication: the cardiac myosin binding protein C Arg502Trp mutation: a common cause of hypertrophic cardiomyopathy. <i>Circulation Research</i> , <b>2010</b> , 106, 1549-52	15.7	56
55	Myocardial fibrosis as an early manifestation of hypertrophic cardiomyopathy. <i>New England Journal of Medicine</i> , <b>2010</b> , 363, 552-63	59.2	452
54	Genetics of hypertrophic cardiomyopathy. <i>Current Opinion in Cardiology</i> , <b>2010</b> , 25, 205-9	2.1	90
53	Cardiac fibrosis in mice with hypertrophic cardiomyopathy is mediated by non-myocyte proliferation and requires Tgf- $\beta$ . <i>Journal of Clinical Investigation</i> , <b>2010</b> , 120, 3520-9	15.9	301
52	A classic twin study of external ear malformations, including microtia. <i>New England Journal of Medicine</i> , <b>2009</b> , 361, 1216-8	59.2	23
51	De novo copy number variants identify new genes and loci in isolated sporadic tetralogy of Fallot. <i>Nature Genetics</i> , <b>2009</b> , 41, 931-5	36.3	325
50	Lamin A/C haploinsufficiency causes dilated cardiomyopathy and apoptosis-triggered cardiac conduction system disease. <i>Journal of Molecular and Cellular Cardiology</i> , <b>2008</b> , 44, 293-303	5.8	122
49	Sarcomere Protein Gene Mutations and Inherited Heart Disease: A $\beta$ -Cardiac Myosin Heavy Chain Mutation Causing Endocardial Fibroelastosis and Heart Failure. <i>Novartis Foundation Symposium</i> , <b>2008</b> , 176-195		10
48	Polony multiplex analysis of gene expression (PMAGE) in mouse hypertrophic cardiomyopathy. <i>Science</i> , <b>2007</b> , 316, 1481-4	33.3	155
47	Cardiac myosin missense mutations cause dilated cardiomyopathy in mouse models and depress molecular motor function. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , <b>2006</b> , 103, 14525-30	11.5	76

46	Gene mutations in apical hypertrophic cardiomyopathy. <i>Circulation</i> , <b>2005</b> , 112, 2805-11	16.7	165
45	Differential cross-bridge kinetics of FHC myosin mutations R403Q and R453C in heterozygous mouse myocardium. <i>American Journal of Physiology - Heart and Circulatory Physiology</i> , <b>2004</b> , 287, H91-9	5.2	61
44	American College of Cardiology/European Society of Cardiology clinical expert consensus document on hypertrophic cardiomyopathy. A report of the American College of Cardiology Foundation Task Force on Clinical Expert Consensus Documents and the European Society of Cardiology Committee for Practice Guidelines. <i>Journal of the American College of Cardiology</i> , <b>2003</b> ,	15.1	1187
43	Dilated cardiomyopathy and heart failure caused by a mutation in phospholamban. <i>Science</i> , <b>2003</b> , 299, 1410-3	33.3	492
42	Assessment of diastolic function with Doppler tissue imaging to predict genotype in preclinical hypertrophic cardiomyopathy. <i>Circulation</i> , <b>2002</b> , 105, 2992-7	16.7	298
41	Sarcomere protein gene mutations in hypertrophic cardiomyopathy of the elderly. <i>Circulation</i> , <b>2002</b> , 105, 446-51	16.7	269
40	Genetic causes of inherited cardiac hypertrophy: Robert L. Frye Lecture. <i>Mayo Clinic Proceedings</i> , <b>2002</b> , 77, 1315-9	6.4	12
39	The L-type calcium channel inhibitor diltiazem prevents cardiomyopathy in a mouse model. <i>Journal of Clinical Investigation</i> , <b>2002</b> , 109, 1013-20	15.9	106
38	The L-type calcium channel inhibitor diltiazem prevents cardiomyopathy in a mouse model. <i>Journal of Clinical Investigation</i> , <b>2002</b> , 109, 1013-1020	15.9	199
37	Introduction of plasmid DNA into cells. <i>Current Protocols in Neuroscience</i> , <b>2001</b> , Appendix 1, Appendix 1L	2.7	
36	The genetic basis for cardiomyopathy: from mutation identification to mechanistic paradigms. <i>Cell</i> , <b>2001</b> , 104, 557-67	56.2	846
35	A murine model of Holt-Oram syndrome defines roles of the T-box transcription factor Tbx5 in cardiogenesis and disease. <i>Cell</i> , <b>2001</b> , 106, 709-21	56.2	833
34	Introduction of plasmid DNA into cells. <i>Current Protocols in Protein Science</i> , <b>2001</b> , Appendix 4, 4D	3.1	6
33	Homozygous mutation in cardiac troponin T: implications for hypertrophic cardiomyopathy. <i>Circulation</i> , <b>2000</b> , 102, 1950-5	16.7	123
32	Mutations in sarcomere protein genes as a cause of dilated cardiomyopathy. <i>New England Journal of Medicine</i> , <b>2000</b> , 343, 1688-96	59.2	550
31	Genetics of Cardiovascular Disease. <i>Circulation</i> , <b>2000</b> , 102,	16.7	3
30	An alpha-cardiac myosin heavy chain gene mutation impairs contraction and relaxation function of cardiac myocytes. <i>American Journal of Physiology - Heart and Circulatory Physiology</i> , <b>1999</b> , 276, H1780-7	5.2	36
29	Electrophysiologic characteristics of accessory atrioventricular connections in an inherited form of Wolff-Parkinson-White syndrome. <i>Journal of Cardiovascular Electrophysiology</i> , <b>1999</b> , 10, 629-35	2.7	41

28	QTL influencing autoimmune diabetes and encephalomyelitis map to a 0.15-cM region containing Il2. <i>Nature Genetics</i> , <b>1999</b> , 21, 158-60	36.3	116
27	Regulation of chamber-specific gene expression in the developing heart by Irx4. <i>Science</i> , <b>1999</b> , 283, 1161-4	36.3	199
26	Mutations in the cardiac transcription factor NKX2.5 affect diverse cardiac developmental pathways. <i>Journal of Clinical Investigation</i> , <b>1999</b> , 104, 1567-73	15.9	468
25	Altered cardiac excitation-contraction coupling in mutant mice with familial hypertrophic cardiomyopathy. <i>Journal of Clinical Investigation</i> , <b>1999</b> , 103, 661-6	15.9	46
24	Dilated cardiomyopathy in homozygous myosin-binding protein-C mutant mice. <i>Journal of Clinical Investigation</i> , <b>1999</b> , 104, 1235-44	15.9	179
23	Familial hypertrophic cardiomyopathy mice display gender differences in electrophysiological abnormalities. <i>Journal of Interventional Cardiac Electrophysiology</i> , <b>1998</b> , 2, 7-14	2.4	37
22	Mutations in a novel cochlear gene cause DFNA9, a human nonsyndromic deafness with vestibular dysfunction. <i>Nature Genetics</i> , <b>1998</b> , 20, 299-303	36.3	286
21	Mutations in human TBX5 [corrected] cause limb and cardiac malformation in Holt-Oram syndrome. <i>Nature Genetics</i> , <b>1997</b> , 15, 30-5	36.3	887
20	Mutations in human TBX3 alter limb, apocrine and genital development in ulnar-mammary syndrome. <i>Nature Genetics</i> , <b>1997</b> , 16, 311-5	36.3	456
19	Amyloid-beta proteins activate Ca(2+)-permeable channels through calcium-sensing receptors. <i>Journal of Neuroscience Research</i> , <b>1997</b> , 47, 547-54	4.4	93
18	Targeted ablation of the murine alpha-tropomyosin gene. <i>Circulation Research</i> , <b>1997</b> , 81, 1005-10	15.7	81
17	beta-Arrestin1 knockout mice appear normal but demonstrate altered cardiac responses to beta-adrenergic stimulation. <i>Circulation Research</i> , <b>1997</b> , 81, 1021-6	15.7	155
16	A human MSX1 homeodomain missense mutation causes selective tooth agenesis. <i>Nature Genetics</i> , <b>1996</b> , 13, 417-21	36.3	551
15	Missense mutation in the pore region of HERG causes familial long QT syndrome. <i>Circulation</i> , <b>1996</b> , 93, 1791-5	16.7	55
14	A mouse model of human familial hypocalciuric hypercalcemia and neonatal severe hyperparathyroidism. <i>Nature Genetics</i> , <b>1995</b> , 11, 389-94	36.3	519
13	Mutations in the cardiac myosin binding protein-C gene on chromosome 11 cause familial hypertrophic cardiomyopathy. <i>Nature Genetics</i> , <b>1995</b> , 11, 434-7	36.3	482
12	A de novo mutation in alpha-tropomyosin that causes hypertrophic cardiomyopathy. <i>Circulation</i> , <b>1995</b> , 91, 2302-5	16.7	44
11	A gene for hereditary haemorrhagic telangiectasia maps to chromosome 9q3. <i>Nature Genetics</i> , <b>1994</b> , 6, 205-9	36.3	174

10	A complex bilateral polysyndactyly disease locus maps to chromosome 7q36. <i>Nature Genetics</i> , <b>1994</b> , 6, 282-6	36.3	79
9	Autosomal dominant hypocalcaemia caused by a Ca(2+)-sensing receptor gene mutation. <i>Nature Genetics</i> , <b>1994</b> , 8, 303-7	36.3	513
8	Homozygosity mapping of the gene for alkaptonuria to chromosome 3q2. <i>Nature Genetics</i> , <b>1993</b> , 5, 201-4	36.3	110
7	The gene responsible for familial hypocalciuric hypercalcemia maps to chromosome 3q in four unrelated families. <i>Nature Genetics</i> , <b>1992</b> , 1, 295-300	36.3	135
6	A molecular basis for familial hypertrophic cardiomyopathy: a beta cardiac myosin heavy chain gene missense mutation. <i>Cell</i> , <b>1990</b> , 62, 999-1006	56.2	1042
5	Mapping a gene for familial hypertrophic cardiomyopathy to chromosome 14q1. <i>New England Journal of Medicine</i> , <b>1989</b> , 321, 1372-8	59.2	415
4	Whole Genome De Novo Variant Identification with FreeBayes and Neural Network Approaches		3
3	Cells and gene expression programs in the adult human heart		9
2	MYBPC3 Mutations cause Hypertrophic Cardiomyopathy by Dysregulating Myosin: Implications for Therapy		1
1	Early post-zygotic mutations contribute to congenital heart disease		2