Combination of Whole Genome Sequencing, Linkage, and Functional Studies Implicates a Missense Mutation in Titin as a Cause of Autosomal Dominant Cardiomyopathy With Features of Left Ventricular Noncomp

DOI: 10.1161/circgenetics.116.001431
Circulation: Cardiovascular Genetics, 2016, 9, 426-435.

#	Paper	IF	Citations
60	Letter by Finsterer and Zarrouk-Mahjoub Regarding Article, "Combination of Whole Genome Sequencing, Linkage, and Functional Studies Implicates a Missense Mutation in Titin as a Cause of Autosomal Dominant Cardiomyopathy With Features of Left Ventricular Noncompaction". *Circulation: Cardiovascular Genetics*, **2016**, 9, 579		0
59	Wrestling the Giant: New Approaches for Assessing Titin Variant Pathogenicity. *Circulation: Cardiovascular Genetics*, **2016**, 9, 392-394		3
58	Titin-Truncating Variants Increase the Risk of Cardiovascular Death in Patients With Hypertrophic Cardiomyopathy. *Canadian Journal of Cardiology*, **2017**, 33, 1292-1297	3.8	10
57	Clinical genetics and outcome of left ventricular non-compaction cardiomyopathy. *European Heart Journal*, **2017**, 38, 3449-3460	9.5	102
56	Structural consequences of mutations associated with idiopathic restrictive cardiomyopathy. *Amino Acids*, **2017**, 49, 1815-1829	3.5	3
55	Genomic Characteristics of Gender Dysphoria Patients and Identification of Rare Mutations in RYR3 Gene. *Scientific Reports*, **2017**, 7, 8339	4.9	12
54	Navigating Genetic and Phenotypic Uncertainty in Left Ventricular Noncompaction. *Circulation: Cardiovascular Genetics*, **2017**, 10,		3
53	TITINdb-a computational tool to assess titinŠs role as a disease gene. *Bioinformatics*, **2017**, 33, 3482-3485	7.2	22
52	When signalling goes wrong: pathogenic variants in structural and signalling proteins causing cardiomyopathies. *Journal of Muscle Research and Cell Motility*, **2017**, 38, 303-316	3.5	9
51	High proportion of genetic cases in patients with advanced cardiomyopathy including a novel homozygous Plakophilin 2-gene mutation. *PLoS ONE*, **2017**, 12, e0189489	3.7	20
50	Fabry Disease and/or Hypertrophic Cardiomyopathy. *International Heart Journal*, **2017**, 58, 305-306	1.8	1
49	Recent Advances in Understanding and Managing Cardiomyopathy. *F1000Research*, **2017**, 6, 1659	3.6	1
48	Systems analysis of dilated cardiomyopathy in the next generation sequencing era. *Wiley Interdisciplinary Reviews: Systems Biology and Medicine*, **2018**, 10, e1419	6.6	8
47	Reasons for missing noncompaction in myopathies and vice versa. *Cardiovascular Pathology*, **2018**, 35, 20-22	3.8	
46	Heart Disease and Stroke Statistics-2018 Update: A Report From the American Heart Association. *Circulation*, **2018**, 137, e67-e492	16.7	3848
45	Precision medicine for cardiovascular disease : Learning lessons from cardiomyopathies. *Herz*, **2018**, 43, 123-130	2.6	6
44	Left Ventricular Noncompaction Cardiomyopathy. **2018**, 269-290		4

43	Role of titin in cardiomyopathy: from DNA variants to patient stratification. *Nature Reviews Cardiology*, **2018**, 15, 241-252	14.8	69
42	A novel SPEG mutation causes non-compaction cardiomyopathy and neuropathy in a floppy infant with centronuclear myopathy. *Acta Neuropathologica Communications*, **2018**, 6, 83	7.3	13
41	Non-sarcomeric causes of heart failure. *Biophysical Reviews*, **2018**, 10, 943-947	3.7	1
40	Takotsubo as Initial Manifestation of Non-Myopathic Cardiomyopathy Due to the Titin Variant c.1489G > T. *Medicines (Basel, Switzerland)*, **2018**, 5,	4.1	2
39	Genetics of Dilated Cardiomyopathy: Clinical Implications. *Current Cardiology Reports*, **2018**, 20, 83	4.2	18
38	Omics studies for comprehensive understanding of immunoglobulin A nephropathy: state-of-the-art and future directions. *Nephrology Dialysis Transplantation*, **2018**, 33, 2101-2112	4.3	4
37	Pathogenic Variant Rs1471414348of the TTN Gene in the Patient with Familial Left Venticular Noncompaction Cardiomyopathy. *Rational Pharmacotherapy in Cardiology*, **2019**, 15, 524-529	0.5	
36	Heart Disease and Stroke Statistics-2019 Update: A Report From the American Heart Association. *Circulation*, **2019**, 139, e56-e528	16.7	3937
35	The giant titin: how to evaluate its role in cardiomyopathies. *Journal of Muscle Research and Cell Motility*, **2019**, 40, 159-167	3.5	6
34	Genetics of Dilated Cardiomyopathy: Current Knowledge and Future Perspectives. **2019**, 45-69		1
33	Titin-truncating variants are associated with heart failure events in patients with left ventricular non-compaction cardiomyopathy. *Clinical Cardiology*, **2019**, 42, 530-535	3.3	6
32	Relevance of Titin Missense and Non-Frameshifting Insertions/Deletions Variants in Dilated Cardiomyopathy. *Scientific Reports*, **2019**, 9, 4093	4.9	19
31	Titin in muscular dystrophy and cardiomyopathy: Urinary titin as a novel marker. *Clinica Chimica Acta*, **2019**, 495, 123-128	6.2	9
30	Expanding the genetic and clinical spectrum of the NONO-associated X-linked intellectual disability syndrome. *American Journal of Medical Genetics, Part A*, **2019**, 179, 792-796	2.5	9
29	Targeted panel sequencing in adult patients with left ventricular non-compaction reveals a large genetic heterogeneity. *Clinical Genetics*, **2019**, 95, 356-367	4	28
28	Left Ventricular Noncompaction Syndrome: Genetic Insights and Therapeutic Perspectives. *Current Cardiology Reports*, **2020**, 22, 84	4.2	11
27	Improved Criteria for the Classification of Titin Variants in Inherited Skeletal Myopathies. *Journal of Neuromuscular Diseases*, **2020**, 7, 153-166	5	10
26	Cardiomyopathy due to PRDM16 mutation: First description of a fetal presentation, with possible modifier genes. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2020**, 184, 129-135	3.1	10

25	Heart Disease and Stroke Statistics-2020 Update: A Report From the American Heart Association. *Circulation*, **2020**, 141, e139-e596	16.7	2824
24	Is Gene-Size an Issue for the Diagnosis of Skeletal Muscle Disorders?. *Journal of Neuromuscular Diseases*, **2020**, 7, 203-216	5	4
23	Heart Disease and Stroke Statistics-2021 Update: A Report From the American Heart Association. *Circulation*, **2021**, 143, e254-e743	16.7	1087
22	Functional analysis of a gene-edited mouse model to gain insights into the disease mechanisms of a titin missense variant. *Basic Research in Cardiology*, **2021**, 116, 14	11.8	6
21	Genetics of Cardiomyopathy.		
20	Clinical Insights Into Heritable Cardiomyopathies. *Frontiers in Genetics*, **2021**, 12, 663450	4.5	3
19	Order from disorder in the sarcomere: FATZ forms a fuzzy but tight complex and phase-separated condensates with α-actinin. *Science Advances*, **2021**, 7,	14.3	1
18	Microbial production of megadalton titin yields fibers with advantageous mechanical properties. *Nature Communications*, **2021**, 12, 5182	17.4	6
17	Making sense of missense variants in TTN-related congenital myopathies. *Acta Neuropathologica*, **2021**, 141, 431-453	14.3	6
16	The Genetic Landscape of Cardiomyopathies. *Cardiac and Vascular Biology*, **2019**, 45-91	0.2	3
15	Pedigree-Based Gene Mapping Supports Previous Loci and Reveals Novel Suggestive Loci in Specific Language Impairment. *Journal of Speech, Language, and Hearing Research*, **2020**, 63, 4046-4061	2.8	7
14	The application of big data to cardiovascular disease: paths to precision medicine. *Journal of Clinical Investigation*, **2020**, 130, 29-38	15.9	34
13	Noncompaction Cardiomyopathy in Childhood. **2019**, 95-126		
12	Neuromuscular Disorders and Noncompaction Cardiomyopathy. **2019**, 41-60		
11	Understanding the Molecular Basis of Cardiomyopathy. *American Journal of Physiology - Heart and Circulatory Physiology*, **2021**,	5.2	1
10	Overlap phenotypes of the left ventricular noncompaction and hypertrophic cardiomyopathy with complex arrhythmias and heart failure induced by the novel truncated DSC2 mutation. *Orphanet Journal of Rare Diseases*, **2021**, 16, 496	4.2	2
9	Heart Disease and Stroke Statistics-2022 Update: A Report From the American Heart Association.. *Circulation*, **2022**, CIR0000000000001052	16.7	196
8	Discerning the Ambiguous Role of Missense Variants in Inherited Arrhythmogenic Syndromes.. *Journal of Personalized Medicine*, **2022**, 12,	3.6	

7	Implication of a novel truncating mutation in titin as a cause of autosomal dominant left ventricular noncompaction.. *Journal of Geriatric Cardiology*, **2022**, 19, 301-314	1.7	
6	Protein Quality Control at the Sarcomere: Titin Protection and Turnover and Implications for Disease Development. *Frontiers in Physiology*, 13,	4.6	1
5	The use of pharmacological chaperones in rare diseases caused by reduced protein stability. 2200222		0
4	Genetics and Genomics of Congenital and Acquired Cardiovascular Disease. **2021**, 1-41		0
3	Heart Disease and Stroke Statistics—2023 Update: A Report From the American Heart Association.		9
2	Beyond gene-disease validity: capturing structured data on inheritance, allelic-requirement, disease-relevant variant classes, and disease mechanism for inherited cardiac conditions.		0
1	Structural and signaling proteins in the Z-disk and their role in cardiomyopathies. 14,		0