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List of Publications by Year in descending order

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1478505 1720034 7 293 6 7 citations h-index g-index papers 7 7 7 556 docs citations times ranked citing authors all docs

#	Article	IF	CITATIONS
1	Opportunities and challenges for antisense oligonucleotide therapies. Journal of Inherited Metabolic Disease, 2021, 44, 72-87.	3.6	86
2	A central role for TFIID in the pluripotent transcription circuitry. Nature, 2013, 495, 516-519.	27.8	73
3	Extension of the Pompe mutation database by linking diseaseâ€associated variants to clinical severity. Human Mutation, 2019, 40, 1954-1967.	2.5	47
4	A genetic modifier of symptom onset in Pompe disease. EBioMedicine, 2019, 43, 553-561.	6.1	32
5	Identification and Characterization of Aberrant <i>GAA</i> Pre-mRNA Splicing in Pompe Disease Using a Generic Approach. Human Mutation, 2015, 36, 57-68.	2.5	28
6	Alternative Splicing in Genetic Diseases: Improved Diagnosis and Novel Treatment Options. International Review of Cell and Molecular Biology, 2018, 335, 85-141.	3.2	23
7	Broad variation in phenotypes for common <i>GAA</i> genotypes in Pompe disease. Human Mutation, 2021, 42, 1461-1472.	2.5	4