

Sebahattin Cirak

List of Publications by Year in descending order

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Version: 2024-02-01

24
papers

3,418
citations

331670

21
h-index

610901

24
g-index

25
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25
docs citations

25
times ranked

4086
citing authors

#	ARTICLE	IF	CITATIONS
1	Exon skipping and dystrophin restoration in patients with Duchenne muscular dystrophy after systemic phosphorodiamidate morpholino oligomer treatment: an open-label, phase 2, dose-escalation study. <i>Lancet</i> , The, 2011, 378, 595-605.	13.7	803
2	Local restoration of dystrophin expression with the morpholino oligomer AVI-4658 in Duchenne muscular dystrophy: a single-blind, placebo-controlled, dose-escalation, proof-of-concept study. <i>Lancet Neurology</i> , The, 2009, 8, 918-928.	10.2	617
3	ISPD loss-of-function mutations disrupt dystroglycan O-mannosylation and cause Walker-Warburg syndrome. <i>Nature Genetics</i> , 2012, 44, 575-580.	21.4	212
4	A Homozygous Mutation in Human PRICKLE1 Causes an Autosomal-Recessive Progressive Myoclonus Epilepsy-Ataxia Syndrome. <i>American Journal of Human Genetics</i> , 2008, 83, 572-581.	6.2	199
5	Mutations in GDP-Mannose Pyrophosphorylase B Cause Congenital and Limb-Girdle Muscular Dystrophies Associated with Hypoglycosylation of Î±-Dystroglycan. <i>American Journal of Human Genetics</i> , 2013, 93, 29-41.	6.2	197
6	Mutations in B3GALNT2 Cause Congenital Muscular Dystrophy and Hypoglycosylation of Î±-Dystroglycan. <i>American Journal of Human Genetics</i> , 2013, 92, 354-365.	6.2	172
7	Dystromirs as Serum Biomarkers for Monitoring the Disease Severity in Duchenne Muscular Dystrophy. <i>PLoS ONE</i> , 2013, 8, e80263.	2.5	137
8	Dystrophin quantification and clinical correlations in Becker muscular dystrophy: implications for clinical trials. <i>Brain</i> , 2011, 134, 3547-3559.	7.6	125
9	<i>DMD</i> genotypes and loss of ambulation in the CINRG Duchenne Natural History Study. <i>Neurology</i> , 2016, 87, 401-409.	1.1	119
10	Discovery of serum protein biomarkers in the mdx mouse model and cross-species comparison to Duchenne muscular dystrophy patients. <i>Human Molecular Genetics</i> , 2014, 23, 6458-6469.	2.9	106
11	Novel mutations expand the clinical spectrum of <i>DYNC1H1</i> -associated spinal muscular atrophy. <i>Neurology</i> , 2015, 84, 668-679.	1.1	106
12	Restoration of the Dystrophin-associated Glycoprotein Complex After Exon Skipping Therapy in Duchenne Muscular Dystrophy. <i>Molecular Therapy</i> , 2012, 20, 462-467.	8.2	99
13	Validation of genetic modifiers for Duchenne muscular dystrophy: a multicentre study assessing <i>SPP1</i> and <i>LTBP4</i> variants. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2015, 86, 1060-1065.	1.9	86
14	ISPD gene mutations are a common cause of congenital and limb-girdle muscular dystrophies. <i>Brain</i> , 2013, 136, 269-281.	7.6	80
15	What Can We Learn From Clinical Trials of Exon Skipping for DMD?. <i>Molecular Therapy - Nucleic Acids</i> , 2014, 3, e152.	5.1	80
16	Zebrafish Fukutin family proteins link the unfolded protein response with dystroglycanopathies. <i>Human Molecular Genetics</i> , 2011, 20, 1763-1775.	2.9	72
17	Myoblasts and macrophages are required for therapeutic morpholino antisense oligonucleotide delivery to dystrophic muscle. <i>Nature Communications</i> , 2017, 8, 941.	12.8	44
18	Transgenic Overexpression of LARGE Induces Î±-Dystroglycan Hyperglycosylation in Skeletal and Cardiac Muscle. <i>PLoS ONE</i> , 2010, 5, e14434.	2.5	42

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19	Exon Skipping Quantification by Quantitative Reverse-Transcription Polymerase Chain Reaction in Duchenne Muscular Dystrophy Patients Treated with the Antisense Oligomer Eteplirsen. <i>Human Gene Therapy Methods</i> , 2012, 23, 336-345.	2.1	38
20	Exome Sequencing Identifies DYNC1H1 Variant Associated With Vertebral Abnormality and Spinal Muscular Atrophy With Lower Extremity Predominance. <i>Pediatric Neurology</i> , 2015, 52, 239-244.	2.1	27
21	Dropped head congenital muscular dystrophy caused by de novo mutations in LMNA. <i>Brain and Development</i> , 2017, 39, 361-364.	1.1	12
22	Shorter Phosphorodiamidate Morpholino Splice-Switching Oligonucleotides May Increase Exon-Skipping Efficacy in DMD. <i>Molecular Therapy - Nucleic Acids</i> , 2018, 13, 534-542.	5.1	7
23	Exon-skipping therapy for Duchenne muscular dystrophy – Authors' reply. <i>Lancet, The</i> , 2012, 379, e10-e11.	13.7	2
24	On the differential diagnosis of neuropathy in neurogenetic disorders. <i>Medizinische Genetik</i> , 2020, 32, 243-261.	0.2	0