Sebahattin Cirak

List of Publications by Year
in descending order

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24 papers	3,418 citations	331670 21 h-index	610901 24 g-index
25 all docs	25 docs citations	25 times ranked	4086 citing authors

#	Article	IF	Citations
1	On the differential diagnosis of neuropathy in neurogenetic disorders. Medizinische Genetik, 2020, 32, 243-261.	0.2	0
2	Shorter Phosphorodiamidate Morpholino Splice-Switching Oligonucleotides May Increase Exon-Skipping Efficacy in DMD. Molecular Therapy - Nucleic Acids, 2018, 13, 534-542.	5.1	7
3	Myoblasts and macrophages are required for therapeutic morpholino antisense oligonucleotide delivery to dystrophic muscle. Nature Communications, 2017, 8, 941.	12.8	44
4	Dropped head congenital muscular dystrophy caused by de novo mutations in LMNA. Brain and Development, 2017, 39, 361-364.	1.1	12
5	DMD genotypes and loss of ambulation in the CINRG Duchenne Natural History Study. Neurology, 2016, 87, 401-409.	1.1	119
6	Exome Sequencing Identifies DYNC1H1 Variant Associated With Vertebral Abnormality and Spinal Muscular Atrophy With Lower Extremity Predominance. Pediatric Neurology, 2015, 52, 239-244.	2.1	27
7	Novel mutations expand the clinical spectrum of DYNC1H1 -associated spinal muscular atrophy. Neurology, 2015, 84, 668-679.	1.1	106
8	Validation of genetic modifiers for Duchenne muscular dystrophy: a multicentre study assessingSPP1andLTBP4variants. Journal of Neurology, Neurosurgery and Psychiatry, 2015, 86, 1060-1065.	1.9	86
9	What Can We Learn From Clinical Trials of Exon Skipping for DMD?. Molecular Therapy - Nucleic Acids, 2014, 3, e152.	5.1	80
10	Discovery of serum protein biomarkers in the mdx mouse model and cross-species comparison to Duchenne muscular dystrophy patients. Human Molecular Genetics, 2014, 23, 6458-6469.	2.9	106
11	Mutations in GDP-Mannose Pyrophosphorylase B Cause Congenital and Limb-Girdle Muscular Dystrophies Associated with Hypoglycosylation of Î±-Dystroglycan. American Journal of Human Genetics, 2013, 93, 29-41.	6.2	197
12	Mutations in B3GALNT2 Cause Congenital Muscular Dystrophy and Hypoglycosylation of Î±-Dystroglycan. American Journal of Human Genetics, 2013, 92, 354-365.	6.2	172
13	ISPD gene mutations are a common cause of congenital and limb-girdle muscular dystrophies. Brain, 2013, 136, 269-281.	7.6	80
14	Dystromirs as Serum Biomarkers for Monitoring the Disease Severity in Duchenne Muscular Dystrophy. PLoS ONE, 2013, 8, e80263.	2.5	137
15	ISPD loss-of-function mutations disrupt dystroglycan O-mannosylation and cause Walker-Warburg syndrome. Nature Genetics, 2012, 44, 575-580.	21.4	212
16	Exon-skipping therapy for Duchenne muscular dystrophy â€“ Authors' reply. Lancet, The, 2012, 379, e10-e11.	13.7	2
17	Restoration of the Dystrophin-associated Glycoprotein Complex After Exon Skipping Therapy in Duchenne Muscular Dystrophy. Molecular Therapy, 2012, 20, 462-467.	8.2	99
18	Exon Skipping Quantification by Quantitative Reverse-Transcription Polymerase Chain Reaction in Duchenne Muscular Dystrophy Patients Treated with the Antisense Oligomer Eteplirsen. Human Gene Therapy Methods, 2012, 23, 336-345.	2.1	38

#	Article	IF	Citations
19	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. Lancet, The, 2011, 378, 595-605.	13.7	803
20	Zebrafish Fukutin family proteins link the unfolded protein response with dystroglycanopathies. Human Molecular Genetics, 2011, 20, 1763-1775.	2.9	72
21	Dystrophin quantification and clinical correlations in Becker muscular dystrophy: implications for clinical trials. Brain, 2011, 134, 3547-3559.	7.6	125
22	Transgenic Overexpression of LARGE Induces Î±-Dystroglycan Hyperglycosylation in Skeletal and Cardiac Muscle. PLoS ONE, 2010, 5, e14434.	2.5	42
23	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. Lancet Neurology, The, 2009, 8, 918-928.	10.2	617
24	A Homozygous Mutation in Human PRICKLE1 Causes an Autosomal-Recessive Progressive Myoclonus Epilepsy-Ataxia Syndrome. American Journal of Human Genetics, 2008, 83, 572-581.	6.2	199