Thomas O Crawford

List of Publications by Year
in descending order

58 papers	5,460 citations	126858 33 h-index	143943 57 g-index
60 all docs	60 docs citations	60 times ranked	5053 citing authors

#	Article	IF	Citations
1	Clinical and genetic spectrum of a large cohort of patients with Î´-sarcoglycan muscular dystrophy. Brain, 2022, 145, 596-606.	3.7	11
2	<scp>TRPV4</scp> mutations causing mixed neuropathy and skeletal phenotypes result in severe gain of function. Annals of Clinical and Translational Neurology, 2022, 9, 375-391.	1.7	9
3	Early treatment is a lifeline for infants with SMA. Nature Medicine, 2022, 28, 1348-1349.	15.2	5
4	Retrospective Diagnosis of Ataxia-Telangiectasia in an Adolescent Patient With a Remote History of T-Cell Leukemia. Journal of Pediatric Hematology/Oncology, 2021, 43, e138-e140.	0.3	2
5	Detection of SMN1 to SMN2 gene conversion events and partial SMN1 gene deletions using array digital PCR. Neurogenetics, 2021, 22, 53-64.	0.7	14
6	Impaired prenatal motor axon development necessitates early therapeutic intervention in severe SMA. Science Translational Medicine, 2021, 13, .	5.8	56
7	Safety and efficacy of nusinersen in spinal muscular atrophy: The <scp>EMBRACE</scp> study. Muscle and Nerve, 2021, 63, 668-677.	1.0	56
8	Growth in ataxia telangiectasia. Orphanet Journal of Rare Diseases, 2021, 16, 123.	1.2	9
9	Onasemnogene abeparvovec gene therapy for symptomatic infantile-onset spinal muscular atrophy in patients with two copies of SMN2 (STR1VE): an open-label, single-arm, multicentre, phase 3 trial. Lancet Neurology, The, 2021, 20, 284-293.	4.9	227
10	Improving the efficacy of exome sequencing at a quaternary care referral centre: novel mutations, clinical presentations and diagnostic challenges in rare neurogenetic diseases. Journal of Neurology, Neurosurgery and Psychiatry, 2021, 92, 1186-1196.	0.9	9
11	Multidisciplinary Management of Ataxia Telangiectasia: Current Perspectives. Journal of Multidisciplinary Healthcare, 2021, Volume 14, 1637-1643.	1.1	13
12	Assuring long-term safety of highly effective gene-modulating therapeutics for rare diseases. Journal of Clinical Investigation, 2021, 131, .	3.9	6
13	Association of Variants in the SPTLC1 Gene With Juvenile Amyotrophic Lateral Sclerosis. JAMA Neurology, 2021, 78, 1236.	4.5	46
14	Revised Recommendations for the Treatment of Infants Diagnosed with Spinal Muscular Atrophy Via Newborn Screening Who Have 4 Copies of SMN2. Journal of Neuromuscular Diseases, 2020, 7, 97-100.	1.1	89
15	The GENDULF algorithm: mining transcriptomics to uncover modifier genes for monogenic diseases. Molecular Systems Biology, 2020, 16, e9701.	3.2	2
16	Chromosome instability syndromes. Nature Reviews Disease Primers, 2019, 5, 64.	18.1	123
17	Nusinersen initiated in infants during the presymptomatic stage of spinal muscular atrophy: Interim efficacy and safety results from the Phase 2 NURTURE study. Neuromuscular Disorders, 2019, 29, 842-856.	0.3	401
18	Description of Restrictively Defined Acute Flaccid Myelitisâ€"Reply. JAMA Pediatrics, 2019, 173, 702.	3.3	1

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19	Neurofilament as a potential biomarker for spinal muscular atrophy. Annals of Clinical and Translational Neurology, 2019, 6, 932-944.	1.7	137
20	066â€…Avxs-101 gene-replacement therapy (GRT) for spinal muscular atrophy type 1 (SMA1): pivotal phase 3 study (STR1VE) update. Journal of Neurology, Neurosurgery and Psychiatry, 2019, 90, A22.1-A22.	0.9	3
21	Clinical Subpopulations in a Sample of North American Children Diagnosed With Acute Flaccid Myelitis, 2012-2016. JAMA Pediatrics, 2019, 173, 134.	3.3	51
22	Age-dependent SMN expression in disease-relevant tissue and implications for SMA treatment. Journal of Clinical Investigation, 2019, 129, 4817-4831.	3.9	106
23	Onasemnogene Abeparvovec Gene-Replacement Therapy (GRT) for Spinal Muscular Atrophy Type 1 (SMA1): Pivotal Phase 3 Study (STR1VE) Update. , 2019, 50, .		1
24	Diagnosis and management of spinal muscular atrophy: Part 1: Recommendations for diagnosis, rehabilitation, orthopedic and nutritional care. Neuromuscular Disorders, 2018, 28, 103-115.	0.3	584
25	Treatment Algorithm for Infants Diagnosed with Spinal Muscular Atrophy through Newborn Screening. Journal of Neuromuscular Diseases, 2018, 5, 145-158.	1.1	148
26	Fifteenâ€year longitudinal followâ€up of a patient with severe earlyâ€onset Charcotâ€Marieâ€Tooth disease type 2A. Muscle and Nerve, 2018, 57, E126-E128.	1.0	0
27	Clinical trial of Lâ€Carnitine and valproic acid in spinal muscular atrophy type I. Muscle and Nerve, 2018, 57, 193-199.	1.0	23
28	Diagnosis and management of spinal muscular atrophy: Part 2: Pulmonary and acute care; medications, supplements and immunizations; other organ systems; and ethics. Neuromuscular Disorders, 2018, 28, 197-207.	0.3	421
29	Duchenne Muscular Dystrophy Newborn Screening, a Case Study for Examining Ethical and Legal Issues for Pilots for Emerging Disorders: Considerations and Recommendations. International Journal of Neonatal Screening, 2018, 4, 6.	1.2	6
30	Charcotâ€“Marieâ€“Tooth Disease type 4C: Novel mutations, clinical presentations, and diagnostic challenges. Muscle and Nerve, 2018, 57, 749-755.	1.0	12
31	Two breakthrough gene-targeted treatments for spinal muscular atrophy: challenges remain. Journal of Clinical Investigation, 2018, 128, 3219-3227.	3.9	78
32	Establishing a reference dataset for the authentication of spinal muscular atrophy cell lines using STR profiling and digital PCR. Neuromuscular Disorders, 2017, 27, 439-446.	0.3	15
33	Assessment of impaired coordination between respiration and deglutition in children and young adults with ataxia telangiectasia. Developmental Medicine and Child Neurology, 2016, 58, 1069-1075.	1.1	9
34	KIF5A mutations cause an infantile onset phenotype including severe myoclonus with evidence of mitochondrial dysfunction. Annals of Neurology, 2016, 80, 633-637.	2.8	47
35	Ataxia telangiectasia: a review. Orphanet Journal of Rare Diseases, 2016, 11, 159.	1.2	419
36	SMN1 and SMN2 copy numbers in cell lines derived from patients with spinal muscular atrophy as measured by array digital PCR. Molecular Genetics & Genomic Medicine, 2015, 3, 248-257.	0.6	56

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37	Astrocytes influence the severity of spinal muscular atrophy. Human Molecular Genetics, 2015, 24, 4094-4102.	1.4	103
38	Exome Sequence Analysis Suggests that Genetic Burden Contributes to Phenotypic Variability and Complex Neuropathy. Cell Reports, 2015, 12, 1169-1183.	2.9	211
39	Non-Aggregating Tau Phosphorylation by Cyclin-Dependent Kinase 5 Contributes to Motor Neuron Degeneration in Spinal Muscular Atrophy. Journal of Neuroscience, 2015, 35, 6038-6050.	1.7	33
40	High-Dose Glucocorticoid Therapy in the Management of Seizures in Neonatal Incontinentia Pigmenti. Journal of Child Neurology, 2015, 30, 100-106.	0.7	21
41	Brain glucose metabolism in adults with ataxia-telangiectasia and their asymptomatic relatives. Brain, 2014, 137, 1753-1761.	3.7	29
42	Susceptibility-Weighted Imaging for Calcification in Cockayne Syndrome. Journal of Pediatrics, 2014, 165, 416-416.e1.	0.9	2
43	SMN Is Essential for the Biogenesis of U7 Small Nuclear Ribonucleoprotein and 3â€²-End Formation of Histone mRNAs. Cell Reports, 2013, 5, 1187-1195.	2.9	76
44	Safety and caregiver satisfaction with gastrostomy in patients with Ataxia Telangiectasia. Orphanet Journal of Rare Diseases, 2011, 6, 23.	1.2	33
45	Evaluation and management of pulmonary disease in ataxiaâ€telangiectasia. Pediatric Pulmonology, 2010, 45, 847-859.	1.0	67
46	Neuronal SMN expression corrects spinal muscular atrophy in severe SMA mice while muscle-specific SMN expression has no phenotypic effect. Human Molecular Genetics, 2008, 17, 1063-1075.	1.4	199
47	Ataxiaâ€telangiectasia: Mild neurological presentation despite null ATM mutation and severe cellular phenotype. American Journal of Medical Genetics, Part A, 2007, 143A, 1827-1834.	0.7	49
48	Robust quantification of the SMN gene copy number by real-time TaqMan PCR. Neurogenetics, 2007, 8, 271-278.	0.7	34
49	Concerns about the design of clinical trials for spinal muscular atrophy. Neuromuscular Disorders, 2004, 14, 456-460.	0.3	39
50	Oropharyngeal dysphagia and aspiration in patients with ataxia-telangiectasia. Journal of Pediatrics, 2000, 136, 225-231.	0.9	113
51	Abnormal fatty acid metabolism in childhood spinal muscular atrophy. Annals of Neurology, 1999, 45, 337-343.	2.8	84
52	Ocular motor abnormalities in ataxia telangiectasia. Annals of Neurology, 1999, 46, 287-295.	2.8	82
53	Stimulation single-fiber EMG in infant botulism. Muscle and Nerve, 1999, 22, 1698-1703.	1.0	40
54	Consequences of the Delayed Diagnosis of Ataxia-Telangiectasia. Pediatrics, 1998, 102, 98-100.	1.0	130

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55	The Neurobiology of Childhood Spinal Muscular Atrophy. Neurobiology of Disease, 1996, 3, 97-110.	2.1	461
56	A novel cDNA detects homozygous microdeletions in greater than 50% of type I spinal muscular atrophy patients. Nature Genetics, 1995, 9, 56-62.	9.4	83
57	Thermal sensitivity in demyelinating neuropathy. Muscle and Nerve, 1993, 16, 301-306.	1.0	30
58	Cerebral white matter changes in acquired immunodeficiency syndrome dementia: Alterations of the blood-brain barrier. Annals of Neurology, 1993, 34, 339-350.	2.8	345