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	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
2	The Neurobiology of Childhood Spinal Muscular Atrophy. Neurobiology of Disease, 1996, 3, 97-110.	2.1	461
3	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
4	Ataxia telangiectasia: a review. Orphanet Journal of Rare Diseases, 2016, 11, 159.	1.2	419
5	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
6	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
7	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
8	Exome Sequence Analysis Suggests that Genetic Burden Contributes to Phenotypic Variability and Complex Neuropathy. Cell Reports, 2015, 12, 1169-1183.	2.9	211
9	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
10	Treatment Algorithm for Infants Diagnosed with Spinal Muscular Atrophy through Newborn Screening. Journal of Neuromuscular Diseases, 2018, 5, 145-158.	1.1	148
11	Neurofilament as a potential biomarker for spinal muscular atrophy. Annals of Clinical and Translational Neurology, 2019, 6, 932-944.	1.7	137
12	Consequences of the Delayed Diagnosis of Ataxia-Telangiectasia. Pediatrics, 1998, 102, 98-100.	1.0	130
13	Chromosome instability syndromes. Nature Reviews Disease Primers, 2019, 5, 64.	18.1	123
14	Oropharyngeal dysphagia and aspiration in patients with ataxia-telangiectasia. Journal of Pediatrics, 2000, 136, 225-231.	0.9	113
15	Age-dependent SMN expression in disease-relevant tissue and implications for SMA treatment. Journal of Clinical Investigation, 2019, 129, 4817-4831.	3.9	106
16	Astrocytes influence the severity of spinal muscular atrophy. Human Molecular Genetics, 2015, 24, 4094-4102.	1.4	103
17	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
18	Abnormal fatty acid metabolism in childhood spinal muscular atrophy. Annals of Neurology, 1999, 45, 337-343.	2.8	84

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19	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
20	Ocular motor abnormalities in ataxia telangiectasia. Annals of Neurology, 1999, 46, 287-295.	2.8	82
21	Two breakthrough gene-targeted treatments for spinal muscular atrophy: challenges remain. Journal of Clinical Investigation, 2018, 128, 3219-3227.	3.9	78
22	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
23	Evaluation and management of pulmonary disease in ataxiaâ€telangiectasia. Pediatric Pulmonology, 2010, 45, 847-859.	1.0	67
24	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
25	Impaired prenatal motor axon development necessitates early therapeutic intervention in severe SMA. Science Translational Medicine, 2021, 13, .	5.8	56
26	Safety and efficacy of nusinersen in spinal muscular atrophy: The <scp>EMBRACE</scp> study. Muscle and Nerve, 2021, 63, 668-677.	1.0	56
27	Clinical Subpopulations in a Sample of North American Children Diagnosed With Acute Flaccid Myelitis, 2012-2016. JAMA Pediatrics, 2019, 173, 134.	3.3	51
28	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
29	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
30	Association of Variants in the SPTLC1 Gene With Juvenile Amyotrophic Lateral Sclerosis. JAMA Neurology, 2021, 78, 1236.	4.5	46
31	Stimulation single-fiber EMG in infant botulism. Muscle and Nerve, 1999, 22, 1698-1703.	1.0	40
32	Concerns about the design of clinical trials for spinal muscular atrophy. Neuromuscular Disorders, 2004, 14, 456-460.	0.3	39
33	Robust quantification of the SMN gene copy number by real-time TaqMan PCR. Neurogenetics, 2007, 8, 271-278.	0.7	34
34	Safety and caregiver satisfaction with gastrostomy in patients with Ataxia Telangiectasia. Orphanet Journal of Rare Diseases, 2011, 6, 23.	1.2	33
35	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
36	Thermal sensitivity in demyelinating neuropathy. Muscle and Nerve, 1993, 16, 301-306.	1.0	30

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37	Brain glucose metabolism in adults with ataxia-telangiectasia and their asymptomatic relatives. Brain, 2014, 137, 1753-1761.	3.7	29
38	Clinical trial of Lâ€Carnitine and valproic acid in spinal muscular atrophy type I. Muscle and Nerve, 2018, 57, 193-199.	1.0	23
39	High-Dose Glucocorticoid Therapy in the Management of Seizures in Neonatal Incontinentia Pigmenti. Journal of Child Neurology, 2015, 30, 100-106.	0.7	21
40	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
41	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
42	Multidisciplinary Management of Ataxia Telangiectasia: Current Perspectives. Journal of Multidisciplinary Healthcare, 2021, Volume 14, 1637-1643.	1.1	13
43	Charcotâ€“Marieâ€“Tooth Disease type 4C: Novel mutations, clinical presentations, and diagnostic challenges. Muscle and Nerve, 2018, 57, 749-755.	1.0	12
44	Clinical and genetic spectrum of a large cohort of patients with Î´-sarcoglycan muscular dystrophy. Brain, 2022, 145, 596-606.	3.7	11
45	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
46	Growth in ataxia telangiectasia. Orphanet Journal of Rare Diseases, 2021, 16, 123.	1.2	9
47	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
48	<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
49	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
50	Assuring long-term safety of highly effective gene-modulating therapeutics for rare diseases. Journal of Clinical Investigation, 2021, 131, .	3.9	6
51	Early treatment is a lifeline for infants with SMA. Nature Medicine, 2022, 28, 1348-1349.	15.2	5
52	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
53	Susceptibility-Weighted Imaging for Calcification in Cockayne Syndrome. Journal of Pediatrics, 2014, 165, 416-416.e1.	0.9	2
54	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

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55	The GENDULF algorithm: mining transcriptomics to uncover modifier genes for monogenic diseases. Molecular Systems Biology, 2020, 16, e9701.	3.2	2
56	Description of Restrictively Defined Acute Flaccid Myelitisâ€"Reply. JAMA Pediatrics, 2019, 173, 702.	3.3	1
57	Onasemnogene Abeparvovec Gene-Replacement Therapy (GRT) for Spinal Muscular Atrophy Type 1 (SMA1): Pivotal Phase 3 Study (STR1VE) Update. , 2019, 50, .		1
58	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