# Clotilde Lagier-Tourenne

List of Publications by Year
in descending order



| 58 | 11,486 | 66343 42 | 133252 59 |
|---|---|---|---|
| papers | citations | h-index | g-index |

| 63 | 63 | 63 | 13043 |
|---|---|---|---|
| all docs | docs citations | times ranked | citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | Cytoplasmic FUS triggers early behavioral alterations linked to cortical neuronal hyperactivity and inhibitory synaptic defects. Nature Communications, 2021, 12, 3028. | 12.8 | 28 |
| 2 | Wild-type FUS corrects ALS-like disease induced by cytoplasmic mutant FUS through autoregulation. Molecular Neurodegeneration, 2021, 16, 61. | 10.8 | 9 |
| 3 | Loss of TDP-43 in male germ cells causes meiotic failure and impairs fertility in mice. Journal of Biological Chemistry, 2021, 297, 101231. | 3.4 | 8 |
| 4 | Reduced C9ORF72 function exacerbates gain of toxicity from ALS/FTD-causing repeat expansion in C9orf72. Nature Neuroscience, 2020, 23, 615-624. | 14.8 | 157 |
| 5 | Phenotypic Suppression of ALS/FTD-Associated Neurodegeneration Highlights Mechanisms of Dysfunction. Journal of Neuroscience, 2019, 39, 8217-8224. | 3.6 | 13 |
| 6 | FUS-mediated regulation of acetylcholine receptor transcription at neuromuscular junctions is compromised in amyotrophic lateral sclerosis. Nature Neuroscience, 2019, 22, 1793-1805. | 14.8 | 81 |
| 7 | Disruption of RNA Metabolism in Neurological Diseases and Emerging Therapeutic Interventions. Neuron, 2019, 102, 294-320. | 8.1 | 176 |
| 8 | Premature polyadenylation-mediated loss of stathmin-2 is a hallmark of TDP-43-dependent neurodegeneration. Nature Neuroscience, 2019, 22, 180-190. | 14.8 | 345 |
| 9 | Overriding FUS autoregulation in mice triggers gain-of-toxic dysfunctions in RNA metabolism and autophagy-lysosome axis. ELife, 2019, 8, . | 6.0 | 65 |
| 10 | Nuclear-Import Receptors Reverse Aberrant Phase Transitions of RNA-Binding Proteins with Prion-like Domains. Cell, 2018, 173, 677-692.e20. | 28.9 | 376 |
| 11 | Nuclear pores: the gate to neurodegeneration. Nature Neuroscience, 2018, 21, 156-158. | 14.8 | 34 |
| 12 | CUG initiation and frameshifting enable production of dipeptide repeat proteins from ALS/FTD C9ORF72 transcripts. Nature Communications, 2018, 9, 152. | 12.8 | 123 |
| 13 | Taking on the Elephant in the Tissue Culture Room: iPSC Modeling for Sporadic ALS. Cell Stem Cell, 2018, 23, 466-467. | 11.1 | 4 |
| 14 | Animal models of neurodegenerative diseases. Nature Neuroscience, 2018, 21, 1370-1379. | 14.8 | 358 |
| 15 | ALS/FTD-Linked Mutation in FUS Suppresses Intra-axonal Protein Synthesis and Drives Disease Without Nuclear Loss-of-Function of FUS. Neuron, 2018, 100, 816-830.e7. | 8.1 | 185 |
| 16 | Motor neuron intrinsic and extrinsic mechanisms contribute to the pathogenesis of FUS-associated amyotrophic lateral sclerosis. Acta Neuropathologica, 2017, 133, 887-906. | 7.7 | 111 |
| 17 | Polyglutamine-Expanded Huntingtin Exacerbates Age-Related Disruption of Nuclear Integrity and Nucleocytoplasmic Transport. Neuron, 2017, 94, 48-57.e4. | 8.1 | 190 |
| 18 | Novel autosomal dominant <scp>TNNT</scp>1 mutation causing nemaline myopathy. Molecular Genetics & Genomic Medicine, 2017, 5, 678-691. | 1.2 | 26 |

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|---|---|---|---|
| 19 | Toxic gain of function from mutant <scp>FUS</scp> protein is crucial to trigger cell autonomous motor neuron loss. EMBO Journal, 2016, 35, 1077-1097. | 7.8 | 187 |
| 20 | Gain of Toxicity from ALS/FTD-Linked Repeat Expansions in C9ORF72 Is Alleviated by Antisense Oligonucleotides Targeting GGGGCC-Containing RNAs. Neuron, 2016, 90, 535-550. | 8.1 | 437 |
| 21 | C9ORF72 poly(GA) aggregates sequester and impair HR23 and nucleocytoplasmic transport proteins. Nature Neuroscience, 2016, 19, 668-677. | 14.8 | 268 |
| 22 | Exome sequencing in amyotrophic lateral sclerosis identifies risk genes and pathways. Science, 2015, 347, 1436-1441. | 12.6 | 823 |
| 23 | ALS-causative mutations in FUS/TLS confer gain and loss of function by altered association with SMN and U1-snRNP. Nature Communications, 2015, 6, 6171. | 12.8 | 205 |
| 24 | RNA-binding proteins in neurodegeneration: Seq and you shall receive. Trends in Neurosciences, 2015, 38, 226-236. | 8.6 | 97 |
| 25 | Novel clinical associations with specific C9ORF72 transcripts in patients with repeat expansions in C9ORF72. Acta Neuropathologica, 2015, 130, 863-876. | 7.7 | 104 |
| 26 | Cerebellar c9RAN proteins associate with clinical and neuropathological characteristics of C9ORF72 repeat expansion carriers. Acta Neuropathologica, 2015, 130, 559-573. | 7.7 | 89 |
| 27 | Translational profiling identifies a cascade of damage initiated in motor neurons and spreading to glia in mutant SOD1-mediated ALS. Proceedings of the National Academy of Sciences of the United States of America, 2015, 112, E6993-7002. | 7.1 | 165 |
| 28 | Direct conversion of patient fibroblasts demonstrates non-cell autonomous toxicity of astrocytes to motor neurons in familial and sporadic ALS. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, 829-832. | 7.1 | 296 |
| 29 | Exploring the Effect of Sequence Length and Composition on Allele-Selective Inhibition of Human Huntingtin Expression by Single-Stranded Silencing RNAs. Nucleic Acid Therapeutics, 2014, 24, 199-209. | 3.6 | 21 |
| 30 | Mutant Huntingtin promotes autonomous microglia activation via myeloid lineage-determining factors. Nature Neuroscience, 2014, 17, 513-521. | 14.8 | 274 |
| 31 | CFTR p.Arg117His associated with CBAVD and other CFTR-related disorders. Journal of Medical Genetics, 2013, 50, 220-227. | 3.2 | 31 |
| 32 | ALS-linked TDP-43 mutations produce aberrant RNA splicing and adult-onset motor neuron disease without aggregation or loss of nuclear TDP-43. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, E736-45. | 7.1 | 370 |
| 33 | Targeted degradation of sense and antisense C9orf72 RNA foci as therapy for ALS and frontotemporal degeneration. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, E4530-9. | 7.1 | 508 |
| 34 | Divergent roles of ALS-linked proteins FUS/TLS and TDP-43 intersect in processing long pre-mRNAs. Nature Neuroscience, 2012, 15, 1488-1497. | 14.8 | 628 |
| 35 | Misregulated RNA processing in amyotrophic lateral sclerosis. Brain Research, 2012, 1462, 3-15. | 2.2 | 150 |
| 36 | Long pre-mRNA depletion and RNA missplicing contribute to neuronal vulnerability from loss of TDP-43. Nature Neuroscience, 2011, 14, 459-468. | 14.8 | 1,050 |

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|---|---|---|---|
| 37 | Molecular diagnosis of known recessive ataxias by homozygosity mapping with SNP arrays. Journal of Neurology, 2011, 258, 56-67. | 3.6 | 14 |
| 38 | An expansion in ALS genetics. Nature, 2010, 466, 1052-1053. | 27.8 | 26 |
| 39 | Variant CCG and GGC repeats within the CTG expansion dramatically modify mutational dynamics and likely contribute toward unusual symptoms in some myotonic dystrophy type 1 patients. Human Molecular Genetics, 2010, 19, 1399-1412. | 2.9 | 139 |
| 40 | Reactive oxygen species, oxidative stress, and cell death correlate with level of CoQ <sub>10</sub> deficiency. FASEB Journal, 2010, 24, 3733-3743. | 0.5 | 142 |
| 41 | Rundataxin, a novel protein with RUN and diacylglycerol binding domains, is mutant in a new recessive ataxia. Brain, 2010, 133, 2439-2447. | 7.6 | 46 |
| 42 | ALS-associated mutations in TDP-43 increase its stability and promote TDP-43 complexes with FUS/TLS. Proceedings of the National Academy of Sciences of the United States of America, 2010, 107, 13318-13323. | 7.1 | 391 |
| 43 | TDP-43 and FUS/TLS: emerging roles in RNA processing and neurodegeneration. Human Molecular Genetics, 2010, 19, R46-R64. | 2.9 | 840 |
| 44 | SPG11 spastic paraplegia. Journal of Neurology, 2009, 256, 104-108. | 3.6 | 96 |
| 45 | Use of SNP array analysis to identify a novel TRIM32 mutation in limb-girdle muscular dystrophy type 2H. Neuromuscular Disorders, 2009, 19, 255-260. | 0.6 | 29 |
| 46 | Rethinking ALS: The FUS about TDP-43. Cell, 2009, 136, 1001-1004. | 28.9 | 516 |
| 47 | ADCK3, an Ancestral Kinase, Is Mutated in a Form of Recessive Ataxia Associated with Coenzyme Q10 Deficiency. American Journal of Human Genetics, 2008, 82, 661-672. | 6.2 | 290 |
| 48 | A new form of childhood onset, autosomal recessive spinocerebellar ataxia and epilepsy is localized at 16q21-q23. Brain, 2007, 130, 1921-1928. | 7.6 | 46 |
| 49 | Homozygous Mutation in SPATA16 Is Associated with Male Infertility in Human Globozoospermia. American Journal of Human Genetics, 2007, 81, 813-820. | 6.2 | 273 |
| 50 | Human mitochondrial pyrophosphatase: cDNA cloning and analysis of the gene in patients with mtDNA depletion syndromes. Genomics, 2006, 87, 410-416. | 2.9 | 22 |
| 51 | AHI1gene mutations cause specific forms of Joubert syndrome-related disorders. Annals of Neurology, 2006, 59, 527-534. | 5.3 | 125 |
| 52 | The gene disrupted in Marinesco-SjÃ¶gren syndrome encodes SIL1, an HSPA5 cochaperone. Nature Genetics, 2005, 37, 1309-1311. | 21.4 | 204 |
| 53 | Thymidine phosphorylase mutations cause instability of mitochondrial DNA. Gene, 2005, 354, 152-156. | 2.2 | 49 |
| 54 | Homozygosity mapping of a third Joubert syndrome locus to 6q23. Journal of Medical Genetics, 2004, 41, 273-277. | 3.2 | 37 |

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|---|---|---|---|
| 55 | Two cousins with partial trisomy 12q and monosomy 12p recombinants of a familial pericentric inversion of the chromosome 12. American Journal of Medical Genetics Part A, 2004, 125A, 77-85. | 2.4 | 12 |
| 56 | Characterisation of mutations in 77 patients with X-linked myotubular myopathy, including a family with a very mild phenotype. Human Genetics, 2003, 112, 135-142. | 3.8 | 113 |
| 57 | Homozygosity mapping of Marinescoâ€“SjÃ¶gren syndrome to 5q31. European Journal of Human Genetics, 2003, 11, 770-778. | 2.8 | 53 |
| 58 | Linkage to 18qter differentiates two clinically overlapping syndromes: congenital cataracts-facial dysmorphism-neuropathy (CCFDN) syndrome and Marinesco-Sjogren syndrome. Journal of Medical Genetics, 2002, 39, 838-843. | 3.2 | 20 |