

## Publications for Kristi Jones

### 2018

Oates, E., Jones, K., Donkervoort, S., Charlton, A., Brammah, S., Smith III, J., Ware, J., Yau, K., Swanson, L., Whiffin, N., Peduto, A., Bournazos, A., Waddell, L., Fitzsimons, R., O'Grady, G., Sandaradura, S., Ghaoui, R., Cooper, S., Clarke, N., et al (2018). Congenital titinopathy: Comprehensive characterisation and pathogenic insights. *Annals of Neurology*, 83(6), 1106-1124. <a href="http://dx.doi.org/10.1002/ana.25241">[More Information]</a>

Dagar, V., Hutchison, W., Muscat, A., Krishnan, A., Hoke, D., Buckle, A., Siswara, P., Amor, D., Mann, J., Jones, K., et al (2018). Genetic variation affecting DNA methylation and the human imprinting disorder, Beckwith-Wiedemann syndrome. *Clinical Epigenetics*, 10(1), 1-13. <a href="http://dx.doi.org/10.1186/s13148-018-0546-4">[More Information]</a>

Koczkowska, M., Chen, Y., Callens, T., Gomes, A., Sharp, A., Johnson, S., Hsiao, M., Chen, Z., Balasubramanian, M., Jones, K., et al (2018). Genotype-Phenotype Correlation in NF1: Evidence for a More Severe Phenotype Associated with Missense Mutations Affecting NF1 Codons 844-848. *American Journal of Human Genetics*, 102(1), 69-87. <a href="http://dx.doi.org/10.1016/j.ajhg.2017.12.001">[More Information]</a>

Jayanetti, V., Klistorner, A., Graham, S., Dexter, M., Flaherty, M., Jones, K., Billson, F., Wilson, M., North, K., Grigg, J., Fraser, C. (2018). Monitoring of optic nerve function in Neurofibromatosis 2 children with optic nerve sheath meningiomas using multifocal visual evoked potentials. *Journal of Clinical Neuroscience*, 50, 262-267. <a href="http://dx.doi.org/10.1016/j.jocn.2018.01.012">[More Information]</a>

Diaz-Manera, J., Fernandez-Torron, R., LLauger, J., James, M., Mayhew, A., Smith, F., Moore, U., Blamire, A., Carlier, P., Rufibach, L., Foster, S., Peduto, A., Jones, K., et al (2018). Muscle MRI in patients with dysferlinopathy: pattern recognition and implications for clinical trials. *Journal of Neurology, Neurosurgery and Psychiatry*, 89(10), 1071-1081. <a href="http://dx.doi.org/10.1136/jnnp-2017-317488">[More Information]</a>

Sandaradura, S., Bournazos, A., Mallawaarachchi, A., Cummings, B., Waddell, L., Jones, K., Troedson, C., Sudarsanam, A., Nash, B., Peters, G., Cooper, S., et al (2018). Nemaline myopathy and distal arthrogryposis associated with an autosomal recessive  $\frac{1}{2}$ TNNT3 $\frac{1}{2}$ splice variant. *Human Mutation*, 39(3), 383-388. <a href="http://dx.doi.org/10.1002/humu.23385">[More Information]</a>

Kariyawasam, D., Carey, K., Jones, K., Farrar, M. (2018). New and developing therapies in spinal muscular atrophy. *Paediatric Respiratory Reviews*, 28, 3-10. <a href="http://dx.doi.org/10.1016/j.prrv.2018.03.003">[More Information]</a>

Farrar, M., Teoh, H., Carey, K., Cairns, A., Forbes, R., Herbert, K., Holland, S., Jones, K., Menezes, M., Morrison, M., Webster, R., et al (2018). Nusinersen for SMA: expanded access programme. *Journal of Neurology, Neurosurgery and Psychiatry*, 89(9), 937-942. <a href="http://dx.doi.org/10.1136/jnnp-2017-317412">[More

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Fitzgerald, D., Abel, F., Jones, K., Farrar, M. (2018). Spinal muscular atrophy: A modifiable disease emerges. *Paediatric Respiratory Reviews*, 28, 1-2. <a href="http://dx.doi.org/10.1016/j.prrv.2018.07.001">[More Information]</a>

Kan, A., Butler, J., Hutchence, M., Jones, K., Widger, J., Doumit, M. (2018). Teaching manually assisted cough to caregivers of children with neuromuscular disease. *Respiratory Care*, 63(12), 1520-1527. <a href="http://dx.doi.org/10.4187/respcare.06213">[More Information]</a>

Moore, U., Jacobs, M., Fernandez-Torron, R., Jang, J., James, M., Mayhew, A., Rufibach, L., Mittal, P., Eagle, M., Jones, K., et al (2018). Teenage exercise is associated with earlier symptom onset in dysferlinopathy: a retrospective cohort study. *Journal of Neurology, Neurosurgery and Psychiatry*, 89(11), 1224-1226. <a href="http://dx.doi.org/10.1136/jnnp-2017-317329">[More Information]</a>

Ravenscroft, G., Pannell, S., O'Grady, G., Ong, R., Ee, H., Faiz, F., Marns, L., Goel, H., Kumarasinghe, P., Sollis, E., et al (2018). Variants in ACTG2 underlie a substantial number of Australasian patients with primary chronic intestinal pseudo-obstruction. *Neurogastroenterology and Motility*, 30(9), 1-9. <a href="http://dx.doi.org/10.1111/nmo.13371">[More Information]</a>

### 2016

Harris, E., Bladen, C., Mayhew, A., James, M., Bettinson, K., Moore, U., Smith, F., Smith, F., Rufibach, L., Cnaan, A., Jones, K., et al (2016). The Clinical Outcome Study for dysferlinopathy: An international multicenter study. *Neurology: Genetics*, 2(4), 1-10. <a href="http://dx.doi.org/10.1212/NXG.000000000000089">[More Information]</a>

### 2015

Pandit, C., Waters, K., Jones, K., Young, H., Fitzgerald, D. (2015). Can daytime measures of lung function predict respiratory failure in children with neuromuscular disease? *Paediatric Respiratory Reviews*, 16(4), 241-245. <a href="http://dx.doi.org/10.1016/j.prrv.2015.08.001">[More Information]</a>

Ghaoui, R., Cooper, S., Lek, M., Jones, K., Corbett, A., Reddel, S., Needham, M., Liang, C., Waddell, L., Nicholson, G., O'Grady, G., Kaur, S., Sue, C., Clarke, N., et al (2015). Use of Whole-Exome Sequencing for Diagnosis of Limb-Girdle Muscular Dystrophy: Outcomes and Lessons Learned. *JAMA Neurology*, 72(12), 1424-1432. <a href="http://dx.doi.org/10.1001/jamaneurol.2015.2274">[More Information]</a>

### 2014

Bushby, K., Finkel, R., Wong, B., Barohn, R., Campbell, C., Comi, G., Connolly, A., Day, J., Flanigan, K., Goemans, N., Jones, K., et al (2014). Ataluren treatment of patients with nonsense mutation dystrophinopathy. *Muscle and Nerve*, 50(4), 477-487. <a href="http://dx.doi.org/10.1002/mus.24332">[More

## 2012

Menezes, M., Waddell, L., Evesson, F., Cooper, S., Webster, R., Jones, K., Mowat, D., Kiernan, M., Johnston, H., Corbett, A., North, K., Clarke, N., et al (2012). Importance and challenge of making an early diagnosis in LMNA-related muscular dystrophy. *Neurology*, 78(16), 1258-1263. <a href="http://dx.doi.org/10.1212/WNL.0b013e318250d839">[More Information]

## 2011

Rohrbach, M., Vandersteen, A., Yi, U., Serdaroglu, G., Ataman, E., Chopra, M., Garcia, S., Jones, K., Kariminejad, A., Kraenzlin, M., et al (2011). Phenotypic variability of the kyphoscoliotic type of Ehlers-Danlos syndrome (EDS VIA): clinical, molecular and biochemical delineation. *Orphanet Journal of Rare Diseases*, 6, Article 46 - 1-Article 46 - 9. <a href="http://dx.doi.org/10.1186/1750-1172-6-46">[More Information]

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Mehr, S., Jones, K., Singh-Grewal, D., Aksentijevich, I., Kakakios, A. (2010). Chronic urticaria of neonatal onset: A potential sign of autoinflammation. *Journal of Paediatrics and Child Health*, 46(10), 608-610. <a href="http://dx.doi.org/10.1111/j.1440-1754.2009.01688.x">[More Information]

Hoornaert, K., Vereecke, I., Dewinter, C., Rosenberg, T., Beemer, F., Leroy, J., Bendix, L., Bjork, E., Bonduelle, M., Boute, O., Jones, K., et al (2010). Stickler syndrome caused by COL2A1 mutations: genotype-phenotype correlation in a series of 100 patients. *European Journal of Human Genetics*, 18(8), 872-880. <a href="http://dx.doi.org/10.1038/ejhg.2010.23">[More Information]

## 2009

D'haene, B., Attanasio, C., Beysen, D., Dostie, J., Lemire, E., Bouchard, P., Field, M., Jones, K., Lorenz, B., Menten, B., et al (2009). Disease-Causing 7.4 kb Cis-Regulatory Deletion Disrupting Conserved Non-Coding Sequences and Their Interaction with the FOXL2 Promotor: Implications for Mutation Screening. *PLoS Genetics*, 5(6), 13. <a href="http://dx.doi.org/10.1371/journal.pgen.1000522">[More Information]

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Compton, A., Albrecht, D., Seto, J., Cooper, S., Ilkovski, B., Jones, K., Challis, D., Mowat, D., Ranscht, B., Bahlo, M., North, K., et al (2008). Mutations in Contactin-1, a Neural Adhesion and Neuromuscular Junction Protein, Cause a Familial Form of Lethal Congenital Myopathy. *American Journal of Human Genetics*, 83(6), 714-724. <a href="http://dx.doi.org/10.1016/j.ajhg.2008.10.022">[More Information]

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Peat, R., Baker, N., Jones, K., North, K., Lamande, S. (2007). Variable penetrance of COL6A1 null mutations: Implications for prenatal diagnosis and genetic counselling in Ullrich congenital muscular dystrophy families. *Neuromuscular Disorders*, 17(7), 547-557. <a href="http://dx.doi.org/10.1016/j.nmd.2007.03.017">[More Information]

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Peat, R., Baker, N., Jones, K., Lamande, S., North, K. (2006). Defining the aetiology of congenital muscular dystrophy in a large cohort. *XIth International Congress on Neuromuscular Diseases*, United Kingdom: Elsevier.

Geevasinga, N., Richards, F., Jones, K. (2006). Juvenile Huntington disease. *Journal of Paediatrics and Child Health*, 42(9), 552-4. <a href="http://dx.doi.org/10.1111/j.1440-1754.2006.00921.x">[More Information]

Gabbett, M., Jones, K., Cowell, C., Sillence, D., Wilson, M. (2006). Neonatal severe hyperparathyroidism: An important clue to the aetiology. *Journal of Paediatrics and Child Health*, 42(12), 813-816. <a href="http://dx.doi.org/10.1111/j.1440-1754.2006.00983.x">[More Information]

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Shaw, C., Stankiewicz, P., Christodoulou, J., Smith, E., Jones, K., Lupski, J. (2004). A Girl With Duplication 17p10-p12 Associated With A Dicentric Chromosome. *American Journal of Medical Genetics, Part A*, 124A (2), 173-178.

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Jones, K., North, K. (2003). Congenital Muscular Dystrophies. In H. R. Jones, D. C. De Vivo, B. T. Darras (Eds.), *Neuromuscular Disorders Of Infancy, Childhood, And Adolescence: A Clinicians Approach*, (pp. 633-647). Amsterdam: Butterworth Heinemann.

Jones, K., Compton, A., Yang, N., Mills, M., Peters, M., Mowat, D., Kunkel, L., Froehner, S., North, K. (2003). Deficiency of the syntrophins and alpha-dystrobrevin in patients with inherited myopathy. *Neuromuscular Disorders*, 13, 456-467. <a href="http://dx.doi.org/10.1016/S0960-8966(03)00066-X">[More Information]

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Jones, K., North, K., He, Y., Vignier, N., Morgan, G., Chevally, M., Barois, A., Estournet-Mathiaud, B., Hori, H., Mizuta, T., et al (2001). Congenital muscular dystrophy with primary partial laminin alpha-2 chain deficiency: Molecular study. *Neurology*, 57, 1319-1322. <a href="http://dx.doi.org/10.1212/WNL.57.7.1319">[More Information]

Jones, K., Ouvrier, R., North, K., Morgan, G., Johnston, H., Tobias, V., Wilkinson, I. (2001). The expanding phenotype of laminin alpha-2 chain (merosin) abnormalities: case series and review. *Journal of Medical Genetics*, 38, 649-657.