

Publications for Lisa Riley

2019

Rius, R., Riley, L., Guo, Y., Menezes, M., Compton, A., Van Bergen, N., Gayevskiy, V., Cowley, M., Cummings, B., Adams, L., Ellaway, C., et al (2019). Cryptic intronic NBAS variant reveals the genetic basis of recurrent liver failure in a child. *Molecular Genetics and Metabolism*, 126(1), 77-82. [More Information]

2018

Balasubramaniam, S., Riley, L., Vasudevan, A., Cowley, M., Gayevskiy, V., Sue, C., Edwards, C., Edkins, E., Junckerstoff, R., Kiraly-Borri, C., Christodoulou, J., et al (2018). EPG5-Related Vici Syndrome: A Primary Defect of Autophagic Regulation with an Emerging Phenotype Overlapping with Mitochondrial Disorders. *JIMD Reports*, 42, 19-29. [More Information]

Coman, D., Vissers, L., Riley, L., Kwint, M., Hauck, R., Koster, J., Geuer, S., Hopkins, S., Hallinan, B., Sweetman, L., Christodoulou, J., et al (2018). Squalene Synthase Deficiency: Clinical, Biochemical, and Molecular Characterization of a Defect in Cholesterol Biosynthesis. *American Journal of Human Genetics*, 103(1), 125-130. [More Information]

Riley, L., Heeney, M., Rudinger-Thirion, J., Frugier, M., Campagna, D., Zhou, R., Hale, G., Hilliard, L., Kaplan, J., Kwiatkowski, J., Christodoulou, J., et al (2018). The phenotypic spectrum of germline YARS2 variants: from isolated sideroblastic anemia to mitochondrial myopathy, lactic acidosis and sideroblastic anemia 2. *Haematologica*, 103(12), 2008-2015. [More Information]

Nafisinia, M., Menezes, M., Gold, W., Riley, L., Hatch, J., Cardinal, J., Coman, D., Christodoulou, J. (2018). Tread carefully: A functional variant in the human NADPH oxidase 4 (NOX4) is not disease causing. *Molecular Genetics and Metabolism*, 123(3), 382-387. [More Information]

2017

Gold, W., Sobreira, N., Wiame, E., Marbaix, A., Van Schaftingen, E., Franzka, P., Riley, L., Worgan, L., Hubner, C., Christodoulou, J., Ades, L. (2017). A novel mutation in GMPPA in siblings with apparent intellectual disability, epilepsy, dysmorphism, and autonomic dysfunction. *American Journal of Medical Genetics, Part A*, 173(8), 2246-2250. [More Information]

Torraco, A., Bianchi, M., Verrigni, D., Gelmetti, V., Riley, L., Niceta, M., Martinelli, D., Montanari, A., Guo, Y., Rizza, T., Christodoulou, J., et al (2017). A novel mutation in NDUFB11 unveils a new clinical phenotype associated with lactic acidosis and sideroblastic anemia. *Clinical Genetics*, 91(3), 441-447. [More Information]

Riley, L., Cowley, M., Gayevskiy, V., Roscioli, T., Thorburn,

D., Prelog, K., Bahlo, M., Sue, C., Balasubramaniam, S., Christodoulou, J. (2017). A SLC39A8 variant causes manganese deficiency, and glycosylation and mitochondrial disorders. *Journal of Inherited Metabolic Disease*, 40(2), 261-269. [More Information]

Nafisinia, M., Riley, L., Gold, W., Bhattacharya, K., Broderick, C., Thorburn, D., Simons, C., Christodoulou, J. (2017). Compound heterozygous mutations in glycyI-tRNA synthetase (GARS) cause mitochondrial respiratory chain dysfunction. *PloS One*, 12(6), 1-12. [More Information]

Nafisinia, M., Sobreira, N., Riley, L., Gold, W., Uhlenberg, B., Weib, C., Boehm, C., Prelog, K., Ouvrier, R., Christodoulou, J. (2017). Mutations in RARS cause a hypomyelination disorder akin to Pelizaeus-Merzbacher disease. *European Journal of Human Genetics*, 25(10), 1134-1141. [More Information]

Maas, R., Iwanicka-Pronicka, K., Kalkan Ucar, S., Alhaddad, B., AlSayed, M., Al-Owain, M., Al-Zaidan, H., Balasubramaniam, S., Baric, I., Bubshait, D., Christodoulou, J., Riley, L., et al (2017). Progressive deafness-dystonia due to SERAC1 mutations - a study of 67 cases. *Annals of Neurology*, 82(6), 1004-1015. [More Information]

Balasubramaniam, S., Riley, L., Bratkovic, D., Ketteridge, D., Manton, N., Cowley, M., Gayevskiy, V., Roscioli, T., Mohamed, M., Gardeitchik, T., Christodoulou, J., et al (2017). Unique presentation of cutis laxa with Leigh-like syndrome due to ECHS1 deficiency. *Journal of Inherited Metabolic Disease*, 40(5), 745-747. [More Information]

Nafisinia, M., Guo, Y., Dang, X., Li, J., Chen, Y., Zhang, J., Lake, N., Gold, W., Riley, L., Thornburn, D., Christodoulou, J., et al (2017). Whole Exome Sequencing Identifies the Genetic Basis of Late-Onset Leigh Syndrome in a Patient with MRI but Little Biochemical Evidence of a Mitochondrial Disorder. *JIMD Reports*, 32, 117-124. [More Information]

Alodaib, A., Sobreira, N., Gold, W., Riley, L., Van Bergen, N., Wilson, M., Bennetts, B., Thorburn, D., Boehm, C., Christodoulou, J. (2017). Whole-exome sequencing identifies novel variants in PNPT1 causing oxidative phosphorylation defects and severe multisystem disease. *European Journal of Human Genetics*, 25(1), 79-84. [More Information]

2016

Heimer, G., Keratar, J., Riley, L., Balasubramaniam, S., Eyal, E., Pietikainen, L., Hiltunen, J., Marek-Yagel, D., Christodoulou, J., et al (2016). MECR Mutations Cause Childhood-Onset Dystonia and Optic Atrophy, a Mitochondrial Fatty Acid Synthesis Disorder. *American Journal of Human Genetics*, 99(6), 1229-1244. [More Information]

Information]

Amon, K., Paxton, K., Klineberg, E., Riley, L., Hazell, P., Skinner, R., Hawke, C., Steinbeck, K. (2016). Recruiting a young adolescent rural cohort: costs and lessons learnt. *Advances in Pediatric Research*, 3, 1-9. [More Information]

2015

Riley, L., Rudinger-Thirion, J., Schmitz-Abe, K., Thorburn, D., Davis, R., Teo, J., Arbuckle, S., Cooper, S., Campagna, D., Frugier, M., Sue, C., Christodoulou, J., et al (2015). LARS2 Variants Associated with Hydrops, Lactic Acidosis, Sideroblastic Anemia, and Multisystem Failure. *JIMD Reports*, 28, 49-57. [More Information]

Menezes, M., Guo, Y., Zhang, J., Riley, L., Cooper, S., Thorburn, D., Li, J., Dong, D., Li, Z., Glessner, J., Davis, R., Sue, C., Alexander, S., Christodoulou, J., et al (2015). Mutation in mitochondrial ribosomal protein S7 (MRPS7) causes congenital sensorineural deafness, progressive hepatic and renal failure and lactic acidemia. *Human Molecular Genetics*, 24(8), 2297-2307. [More Information]

2014

Miller, D., Menezes, M., Simons, C., Riley, L., Cooper, S., Grimmond, S., Thorburn, D., Christodoulou, J., Taft, R. (2014). Rapid identification of a novel complex I MT-ND3 m.10134C>A mutation in a Leigh syndrome patient. *PloS One*, 9(8), 1-6. [More Information]

Morgan, A., Riley, L., Sheehy, P., Wynn, P. (2014). The influence of protein fractions from bovine colostrum digested in vivo and in vitro on human intestinal epithelial cell proliferation. *Journal of Dairy Research*, 81(1), 73-81. [More Information]

2013

Riley, L., Menezes, M., Rudinger-Thirion, J., Duff, R., de Lonlay, P., Rotig, A., Tchan, M., Davis, M., Cooper, S., Christodoulou, J. (2013). Phenotypic variability and identification of novel YARS2 mutations in YARS2 mitochondrial myopathy, lactic acidosis and sideroblastic anaemia. *Orphanet Journal of Rare Diseases*, 8(1), 1-11. [More Information]

2010

Riley, L., Gardiner-Garden, M., Thomson, P., Wynn, P., Williamson, P., Raadsma, H., Sheehy, P. (2010). The influence of extracellular matrix and prolactin on global gene expression profiles of primary bovine mammary epithelial cells in vitro. *Animal Genetics*, 41(1), 55-63. [More Information]

2009

Sheehy, P., Riley, L., Raadsma, H., Williamson, P., Wynn, P. (2009). A Functional Genomics approach to evaluate Candidate Genes located in a QTL interval for Milk production Traits on BTA6. *Animal Genetics*, 40(4), 492-498. [More Information]

Information]

2008

Riley, L., Williamson, P., Raadsma, H., Wynn, P., Sheehy, P. (2008). Elf5 Expression In the Bovine Mammary Gland During the Lactation Cycle and its Role in Milk Protein Gene Expression In Vitro. *5th International Symposium on Milk Genomics and Human Health 2008*, United States: California Dairy Research Foundation.

Riley, L., Williamson, P., Wynn, P., Sheehy, P. (2008). Lactoferrin decreases primary bovine mammary epithelial cell viability and casein expression. *Journal of Dairy Research*, 75, 135-141. [More Information]

Riley, L., Wynn, P., Williamson, P., Sheehy, P. (2008). The Role of Native Bovine α -lactalbumin in Bovine Mammary Epithelial Cell Apoptosis and Casein Expression. *Journal of Dairy Research*, 75, 319-325. [More Information]

2007

Wynn, P., Nicholas, L., Riley, L., Sheehy, P. (2007). Comparative Genomics And Lactation: Making More Money From Milk. *Pakistan Congress of Zoology*, Pakistan: Islamabad : The Society.

Wynn, P., Riley, L., Nicholas, K., Sheehy, P. (2007). Exploiting mammalian lactational strategies to boost commercial milk production. *XVI Conference of the Society of Animal Physiologists of India (SAPI)*, India: Society of Animal Physiologists of India.

2006

Riley, L., Roufogalis, B., Li, G., Weiss, A. (2006). A radioassay for synaptic core complex assembly: Screening of herbal extracts for effectors. *Analytical Biochemistry*, 357(1), 50-57. [More Information]

Wei, J., Cavanagh, J., Riley, L., Geale, P., Sheehy, P., Wynn, P., Raadsma, H., Williamson, P. (2006). Analysis of ABCG2 Function in the Mammary Gland. *Queenstown Molecular Biology Meeting 2006*.

Riley, L., Zubair, M., Thomson, P., Holt, M., Xavier, S., Wynn, P., Sheehy, P. (2006). Lactational performance of Quackenbush Swiss line 5 mice. *Journal of Animal Science*, 84(8), 2118-2125. [More Information]

2005

Wei, J., Cavanagh, J., Riley, L., Mathews, K., Sheehy, P., Wynn, P., Raadsma, H., Williamson, P. (2005). Analysis of ABCG2, a selected candidate gene associated with dairy traits. *Dairy research Foundation Symposium 2005*, Sydney: University of Sydney.

2004

Morgan, A., Riley, L., Sheehy, P., Wynn, P. (2004). Identification of Novel Bioactives in Bovine Colostrum: Regulation of Gut Development and Pituitary Hormone Secretion - in Current Topics in Dairy Production. *Dairy research Foundation Symposium 2004*, Sydney, Australia: University of Sydney.