

Publications for Kaustuv Bhattacharya

2018

Selvanathan, A., Ellaway, C., Wilson, C., Owens, P., Shaw, P., Bhattacharya, K. (2018). Effectiveness of Early Hematopoietic Stem Cell Transplantation in Preventing Neurocognitive Decline in Mucopolysaccharidosis Type II: A Case Series. *JIMD Reports*, 41, 81-89. [More Information]

Owens, P., Wong, M., Bhattacharya, K., Ellaway, C. (2018). Infantile-onset Pompe disease: A case series highlighting early clinical features, spectrum of disease severity and treatment response. *Journal of Paediatrics and Child Health*, 54(11), 1255-1261. [More Information]

2017

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

Schrover, R., Evans, K., Giugliani, R., Noble, I., Bhattacharya, K. (2017). Minimal clinically important difference for the 6-min walk test: Literature review and application to Morquio A syndrome. *Orphanet Journal of Rare Diseases*, 12(1), 1-11. [More Information]

Korula, S., Owens, P., Charlton, A., Bhattacharya, K. (2017). Rare Case of Hepatic Gaucheroma in a Child on Enzyme Replacement Therapy. *JIMD Reports*, 32, 101-104. [More Information]

2016

Montano, A., Lock-Hock, N., Steiner, R., Graham, B., Szlago, M., Greenstein, R., Pineda, M., Gonzalez-Meneses, A., Coker, M., Bhattacharya, K., et al (2016). Clinical course of sly syndrome (mucopolysaccharidosis type VII). *Journal of Medical Genetics*, 53(6), 403-418. [More Information]

Bhattacharya, K., Pontin, J., Thompson, S. (2016). Dietary Management of the Ketogenic Glycogen Storage Diseases. *Journal of Inborn Errors of Metabolism and Screening*, 4, 1-6. [More Information]

Menezes, M., Rahman, S., Bhattacharya, K., Clark, D., Christodoulou, J., Ellaway, C., Farrar, M., Pitt, M., Sampaio, H., Ware, T., Ouvrier, R., et al (2016). Neurophysiological profile of peripheral neuropathy associated with childhood mitochondrial disease. *Mitochondrion*, 30, 162-167. [More Information]

2015

Bhattacharya, K., Mundy, H., Lilburn, M., Champion, M.,

Morley, D., Maillot, F. (2015). A pilot longitudinal study of the use of waxy maize heat modified starch in the treatment of adults with glycogen storage disease type I: A randomized double-blind cross-over study. *Orphanet Journal of Rare Diseases*, 10(1), 1-11. [More Information]

Berrier, K., Kazi, Z., Prater, S., Bali, D., Goldstein, J., Stefanescu, M., Rehder, C., Botha, E., Ellaway, C., Bhattacharya, K., et al (2015). CRIM-negative infantile Pompe disease: characterization of immune responses in patients treated with ERT monotherapy. *Genetics in Medicine*, 17(11), 912-918. [More Information]

Choy, Y., Bhattacharya, K., Balasubramaniam, S., Fietz, M., Fu, A., Inwood, A., Jin, D., Kim, O., Kosuga, M., Kwun, Y., et al (2015). Identifying the need for a multidisciplinary approach for early recognition of mucopolysaccharidosis VI (MPS VI). *Molecular Genetics and Metabolism*, 115(1), 41-47. [More Information]

Bhattacharya, K. (2015). Investigation and management of the hepatic glycogen storage diseases. *Translational Pediatrics*, 4(3), 240-248.

Bhattacharya, K. (2015). Where will genetic research take us? *Translational Pediatrics*, 4(4), 318-319. [More Information]

2014

Estrella, J., Wilcken, B., Carpenter, K., Bhattacharya, K., Tchan, M., Wiley, V. (2014). Expanded newborn screening in New South Wales: missed cases. *Journal of Inherited Metabolic Disease*, 37(6), 881-887. [More Information]

Ho, G., Alexander, I., Bhattacharya, K., Dennison, B., Ellaway, C., Thompson, S., Wilcken, B., Christodoulou, J. (2014). The Molecular Bases of Phenylketonuria (PKU) in New South Wales, Australia: Mutation Profile and Correlation with Tetrahydrobiopterin (BH4) Responsiveness. *JIMD Reports*, 14, 55-65. [More Information]

Baker, P., Friederich, M., Swanson, M., Shaikh, T., Bhattacharya, K., Scharer, G., Aicher, J., Creardon-Swindell, G., Geiger, E., Maclean, K., Procopis, P., et al (2014). Variant non-ketotic hyperglycinemia is caused by mutations in LIAS, BOLA3 and the novel gene GLRX5. *Brain*, 137(2), 366-379. [More Information]

2012

Coman, D., Bhattacharya, K. (2012). Extended newborn screening: An update for the general paediatrician. *Journal of Paediatrics and Child Health*, 48(2), E68-E72. <a href="http://dx.doi.org/10.1111/j.1440-

1754.2011.02199.x">[More Information]

2011

Bhattacharya, K. (2011). Dietary dilemmas in the management of glycogen storage disease type I. *Journal of Inherited Metabolic Disease*, 34(3), 621-629. [More Information]

2010

Dagli, A., Lee, P., Correia, C., Rodriguez, C., Bhattacharya, K., Steinkrauss, L., Stanley, C., Weinstein, D. (2010). Pregnancy in glycogen storage disease type Ib: gestational care and report of first successful deliveries. *Journal of Inherited Metabolic Disease*, 33(Suppl 3), S151-S157. [More Information]

2009

Shanti, B., Silink, M., Bhattacharya, K., Howard, N., Carpenter, K., Fietz, M., Clayton, P., Christodoulou, J. (2009). Congenital disorder of glycosylation type Ia: Heterogeneity in the clinical presentation from multivisceral failure to hyperinsulinaemic hypoglycaemia as leading symptoms in three infants with phosphomannomutase deficiency. *Journal of Inherited Metabolic Disease*, Short Report #166 - online, 1-11. [More Information]

2006

Bhattacharya, K., Khalili, V., Wiley, V., Carpenter, K., Wilcken, B. (2006). Newborn screening may fail to identify intermediate forms of maple syrup urine disease. *Journal of Inherited Metabolic Disease*, 29(4), 586-586. [More Information]