Publications for Janine Smith

2017

Shi, H., Enriquez, A., Rapadas, M., Martin, E., Wang, R., Moreau, J., Lim, C., Szot, J., Ip, E., Hughes, J., Smith, J., Collins, F., Sillence, D., Winlaw, D., et al (2017). NAD deficiency, congenital malformations, and niacin supplementation. *New England Journal of Medicine*, 377(6), 544-552. [More Information]

2016

Eggers, S., Sadedin, S., van den Bergen, J., Robevska, G., Ohnesorg, T., Hewitt, J., Lambeth, L., Bouty, A., Knarston, I., Smith, G., Smith, J., Ewans, L., Shalhoub, C., Cowell, C., et al (2016). Disorders of sex development: insights from targeted gene sequencing of a large international patient cohort. *Genome Biology*, 17(1), 1-21. [More Information]

2009

Meyer-Rochow, G., Smith, J., Richardson, A., Marsh, D., Sidhu, S., Robinson, B., Benn, D. (2009). Denaturing High Performance Liquid Chromatography Detection of SDHB, SDHD, and VHL Germline Mutations in Pheochromocytoma. *Journal of Surgical Research*, 157, 55-62. [More Information]

2008

Rodriguez-Ballestero, M., Reynoso, R., Olarte, M., Villamar, M., Morera, C., Santarelli, R., Arslan, E., Meda, C., Smith, J., et al (2008). A multicenter study on the prevalence and spectrum of mutations in the otoferlin gene (OTOF) in subjects with nonsyndromic hearing impairment and auditory neuropathy. *Human Mutation*, 29(6), 823-831. [More Information]

Clement, E., Mercuri, E., Godfrey, C., Smith, J., Robb, S., Kinali, M., Straub, V., Bushby, K., Manzur, A., Talim, B., North, K., et al (2008). Brain involvement in muscular dystrophies with defective dystroglycan glycosylation. *Annals of Neurology*, 64(5), 573-582. [More Information]

Peat, R., Smith, J., Compton, A., Baker, N., Pace, R., Burkin, D., Kaufman, S., Lamande, S., North, K. (2008). The diagnosis and etiology of congenital muscular dystrophy. *Neurology*, 71(5), 312-321. [More Information]

2007

Godfrey, C., Clement, E., Mein, R., Brockington, M., Smith, J., Talim, B., Straub, V., Robb, S., Quinlivan, R., Feng, L., North, K., et al (2007). Refining genotype phenotype correlations in muscular dystrophies with defective glycosylation of dystroglycan. *Brain*, 130(10), 2725-2735. [More Information]

2005

Bajaj, R., Smith, J., Trochet, D., Pitkin, J., Ouvrier, R., Graf, N., Sillence, D., Kluckow, M. (2005). Congenital central hypoventilation syndrome and Hirschsprung's disease in an extremely preterm infant. *Pediatrics*, 115(6), e737-e738. [More Information]

Senderek, J., Krieger, M., Stendel, C., Bergmann, C., Moser, M., Breitbach-Faller, N., Rudnik-SchÃ¶neborn, S., Blaschek, A., Wolf, N., et al, North, K., Smith, J. (2005). Mutations in SIL1 cause Marinesco-SjÃ¶gren syndrome, a cerebellar ataxia with cataract and myopathy. *Nature Genetics*, 37(12), 1312-1314. [More Information]

Neas, K., Smith, J., Chia, N., Huseyin, S., St Heaps, L., Sholler, G., Tzioumi, D., Sillence, D., Mowat, D. (2005). Three patients with terminal deletions within the subtelomeric region of chromosome 9q. *American Journal of Medical Genetics, Part A*, 132(4), 425-30. [More Information]

2004

Kirk, E., Smith, J., Field, M., Marshall, G., Marsh, D. (2004). Diagnosis of Proteus Syndrome was correct. *American Journal of Medical Genetics, Part A*, 130A (2), 214-215. [More Information]