

References for *BTB*

November 3, 2009

- Gordon, A. (1996). "Biotinidase mutational 'hotspot'." Nat Genet **13**(2): 144-5.
- Hymes, J., C. M. Stanley, et al. (2001). "Mutations in BTB causing biotinidase deficiency." Hum Mutat **18**(5): 375-81.
- Muhl, A., D. Moslinger, et al. (2001). "Molecular characterisation of 34 patients with biotinidase deficiency ascertained by newborn screening and family investigation." Eur J Hum Genet **9**(4): 237-43.
- Norrgard, K. J., R. J. Pomponio, et al. (1997). "Mutation (Q456H) is the most common cause of profound biotinidase deficiency in children ascertained by newborn screening in the United States." Biochem Mol Med **61**(1): 22-7.
- Norrgard, K. J., R. J. Pomponio, et al. (1998). "Double mutation (A171T and D444H) is a common cause of profound biotinidase deficiency in children ascertained by newborn screening the the United States. Mutations in brief no. 128. Online." Hum Mutat **11**(5): 410.
- Norrgard, K. J., R. J. Pomponio, et al. (1999). "Mutations causing profound biotinidase deficiency in children ascertained by newborn screening in the United States occur at different frequencies than in symptomatic children." Pediatr Res **46**(1): 20-7.
- Pomponio, R. J., T. R. Reynolds, et al. (1995). "Mutational hotspot in the human biotinidase gene causes profound biotinidase deficiency." Nat Genet **11**(1): 96-8.
- Pomponio, R. J., K. J. Norrgard, et al. (1997). "Arg538 to Cys mutation in a CpG dinucleotide of the human biotinidase gene is the second most common cause of profound biotinidase deficiency in symptomatic children." Hum Genet **99**(4): 506-12.
- Pomponio, R. J., J. Hymes, et al. (1997). "Mutations in the human biotinidase gene that cause profound biotinidase deficiency in symptomatic children: molecular, biochemical, and clinical analysis." Pediatr Res **42**(6): 840-8.
- Suormala, T. M., E. R. Baumgartner, et al. (1990). "Comparison of patients with complete and partial biotinidase deficiency: biochemical studies." J Inherit Metab Dis **13**(1): 76-92.
- Wolf, B., G. S. Heard, et al. (1985). "Biotinidase deficiency: initial clinical features and rapid diagnosis." Ann Neurol **18**(5): 614-7.
- Wolf, B. (1991). "Worldwide survey of neonatal screening for biotinidase deficiency." J Inherit Metab Dis **14**(6): 923-7.
- Wolf, B., K. Norrgard, et al. (1997). "Profound biotinidase deficiency in two asymptomatic adults." Am J Med Genet **73**(1): 5-9.