

## Sterol and Isoprenoid Research Consortium

### Book Chapters

1. He M, Smith L, Vockley J. SC4MOL deficiency. In: Valle D, Beaudet A, Vogelstein B, et al., eds. *Online Metabolic and Molecular Bases of Inherited Disease*. New York, NY: McGraw-Hill Global Education Holdings, LLC; 2014.

### Journal Articles

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3. DeBarber AE, Connor WE, Pappu AS, Merkens LS, Steiner RD. ESI-MS/MS quantification of 7 $\alpha$ -hydroxy-4-cholesten-3-one facilitates rapid, convenient diagnostic testing for cerebrotendinous xanthomatosis. *Clin. Chim. Acta.* Jan 2010;411(1-2):43-48. PMID: 19808031
4. McLarren KW, Severson TM, du Souich C, Stockton DW, Kratz LE, Cunningham D, Hendson G, Morin RD, Wu D, Paul JE, An J, Nelson TN, Chou A, DeBarber AE, Merkens LS, Michaud JL, Waters PJ, Yin J, McGillivray B, Demos M, Rouleau GA, Grzeschik KH, Smith R, Tarpey PS, Shears D, Schwartz CE, Gecz J, Stratton MR, Arbour L, Hurlburt J, Van Allen MI, Herman GE, Zhao Y, Moore R, Kelley RI, Jones SJ, Steiner RD, Raymond FL, Marra MA, Boerkoel CF. Hypomorphic temperature-sensitive alleles of NSDHL cause CK syndrome. *Am. J. Hum. Genet.* Dec 10 2010;87(6):905-914. PMID: 21129721, PMCID: PMC2997364
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7. DeBarber AE, Sandler Y, Pappu AS, Merkens LS, Duell PB, Lear SR, Erickson SK, Steiner RD. Profiling sterols in cerebrotendinous xanthomatosis: utility of Girard derivatization and high resolution exact mass LC-ESI-MS(n) analysis. *J. Chromatogr. B Analyt. Technol. Biomed. Life. Sci.* May 15 2011;879(17-18):1384-1392. PMID: 21168372, PMCID: PMC3326078
8. Steiner LA, Ehrenkranz RA, Peterec SM, Steiner RD, Reyes-Mugica M, Gallagher PG. Perinatal onset mevalonate kinase deficiency. *Pediatr. Dev. Pathol.* Jul-Aug 2011;14(4):301-306. PMID: 21425920
9. Monson DM, DeBarber AE, Bock CJ, Anadiotis G, Merkens LS, Steiner RD, Stout AU. Cerebrotendinous xanthomatosis: a treatable disease with juvenile cataracts as a presenting sign. *Arch. Ophthalmol.* Aug 2011;129(8):1087-1088. PMID: 21825196, PMCID: PMC3366103
10. Hager EJ, Piganelli JD, Tse HM, Gibson KM. Aberrant expression of costimulatory molecules in splenocytes of the mevalonate kinase-deficient mouse model of human hyper-IgD syndrome (HIDS). *J. Inherit. Metab. Dis.* Jan 2012;35(1):159-168. PMID: 21607759, PMCID: PMC3654530

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22. Othman RA, Myrie SB, Mymin D, et al. Ezetimibe reduces plant sterol accumulation and favorably increases platelet count in sitosterolemia. *J. Pediatr.* Jan 2015;166(1):125-131. PMID: 25444527, PMCID: PMC4274192
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