Publications for Bridget Wilcken

2019

Martinez, N., Lipke, M., Robinson, J., Wilcken, B. (2019). Sialuria: Ninth patient described has a novel mutation in GNE. In edited by Eva Morava, Matthias Baumgartner, Marc Patterson, Shamima Rahman, Joha (Eds.), *JIMD Reports, Volume 44*, (pp. 17-21). Berlin: Springer. [More Information]

2018

Sampaio, H., Wilcken, B., Farrar, M. (2018). Screening for spinal muscular atrophy. *Medical Journal of Australia*, 209(4), 147-148. [More Information]

2017

Coffey, M., Whitaker, V., Gentin, N., Junek, R., Shalhoub, C., Nightingale, S., Hilton, J., Wiley, V., Wilcken, B., Gaskin, K., et al (2017). Differences in Outcomes between Early and Late Diagnosis of Cystic Fibrosis in the Newborn Screening Era. *The Journal of Pediatrics*, 181, 137-145. [More Information]

Coffey, M., Whitaker, V., Gentin, N., Junek, R., Shalhoub, C., Nightingale, S., Hilton, J., Wiley, V., Wilcken, B., Gaskin, K., et al (2017). Differences in Outcomes between Early and Late Diagnosis of Cystic Fibrosis in the Newborn Screening Era. *Obstetrical and Gynecological Survey*, 72(6), 328-330. [More Information]

Okun, J., Gan-Schreier, H., Ben-Omran, T., Schmidt, K., Fang-Hoffmann, J., Gramer, G., Abdoh, G., Shahbeck, N., Al Rifai, H., Al Khal, A., et al (2017). Newborn Screening for Vitamin B6 Non-responsive Classical Homocystinuria: Systematical Evaluation of a Two-Tier Strategy. In E. Morava, M. Baumgartner, M. Patterson, S. Rahman, J Zschocke & V Peters (Eds.), *JIMD Reports*, (pp. 87-94). Berlin Heidelberg: Springer. [More Information]

Wilcken, B. (2017). Therapeutic targets in homocystinuria due to cystathionine beta-synthase deficiency: new European guidelines. *Expert Opinion on Orphan Drugs*, 5(1), 1-3. [More Information]

2016

Wilcken, B. (2016). 3-Methylcrotonyl-CoA carboxylase deficiency: to screen or not to screen? *Journal of Inherited Metabolic Disease*, 39, 171-172. [More Information]

Lain, S., Bentley, J., Wiley, V., Roberts, C., Jack, M., Wilcken, B., Nassar, N. (2016). Association between borderline neonatal thyroid-stimulating hormone concentrations and educational and developmental outcomes: a population-based record-linkage study. *The Lancet Diabetes & Endocrinology*, 4(9), 756-765. [More Information]

Duley, J., Henman, M., Carpenter, K., Bamshad, M., Marshall, G., Ooi, C., Wilcken, B., Pinner, J. (2016). Elevated plasma dihydroorotate in Miller syndrome: Biochemical, diagnostic and clinical implications, and treatment with uridine. *Molecular Genetics and Metabolism*, 119, 83-90. [More Information]

2015

Wilcken, B., Wiley, V. (2015). Fifty years of newborn screening. *Journal of Paediatrics and Child Health*, 51(1), 103-107. [More Information]

Wilcken, B. (2015). Treatments for rare diseases: Molybdenum cofactor deficiency. *The Lancet*, 386(10007), 1924-1925. [More Information]

Lain, S., Roberts, C., Wilcken, B., Wiley, V., Jack, M., Nassar, N. (2015). Using record linkage to investigate perinatal factors and neonatal thyroid-stimulating hormone. *Journal of Paediatrics and Child Health*, 51(6), 620-625. [More Information]

2014

Lim, S., Smith, K., Stroud, D., Compton, A., Tucker, E., Dasvarma, A., Gandolfo, L., Marum, J., McKenzie, M., Peters, H., Procopis, P., Wilcken, B., Christodoulou, J., et al (2014). A Founder Mutation in PET100 Causes Isolated Complex IV Deficiency in Lebanese Individuals with Leigh Syndrome. *American Journal of Human Genetics*, 94(2), 209-222. [More Information]

Sudarsanam, A., Singh, H., Wilcken, B., Stormon, M., Arbuckle, S., Schmitt, B., Clayton, P., Earl, J., Webster, R. (2014). Cirrhosis Associated with Pyridoxal 5'-Phosphate Treatment of Pyridoxamine 5'-Phosphate Oxidase Deficiency. In Johannes Zschocke, K. Michael Gibson, Garry Brown, Eva Morava, Verena Peters (Eds.), *JIMD Reports Volume 17*, (pp. 67-70). Heidelberg, Germany: Springer.

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]

Estrella, J., Yee, G., Wilcken, B., Tchan, M., Talbot, M. (2014). Hyperammonemic encephalopathy complicating bariatric surgery: A case study and review of the literature. *Surgery for Obesity and Related Diseases*, 10(3), e35-e38. [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]

Wilcken, B. (2014). The ward round. *Journal of Paediatrics and Child Health*, 50(8), 660-660. [More Information]

2013

Wilcken, B. (2013). Congenital adrenal hyperplasia: One hundred years of data. *The Lancet Diabetes and Endocrinology*, 1(1), 4-5. [More Information]

Christie, L., Wotton, T., Bennetts, B., Wiley, V., Wilcken, B., Rogers, C., Boyle, J., Turner, C., Hansen, J., Hunter, M., et al (2013). Maternal attitudes to newborn screening for fragile X syndrome. *American Journal of Medical Genetics, Part A*, 161A (2), 301-311. [More Information]

Wilcken, B. (2013). Newborn screening: Gaps in the evidence. *Science*, 342(6155), 197-198. [More Information]

van de Kamp, J., Betsalel, O., Mercimek-Mahmutoglu, S., Abulhoul, L., Grunewald, S., Anselm, I., Azzouz, H., Bratkovic, D., de Brouwer, A., Hamel, B., et al (2013). Phenotype and genotype in 101 males with X-linked creatine transporter deficiency. *Journal of Medical Genetics*, 50(7), 463-472. [More Information]

Tchan, M., Westbrook, M., Wilcox, G., Cutler, R., Smith, N., Penman, R., Strauss, B., Wilcken, B. (2013). The Management of Pregnancy in Maple Syrup Urine Disease: Experience with Two Patients. *JIMD Reports*, 10, 113-117. [More Information]

Tchan, M., Wilcken, B., Christodoulou, J. (2013). The mild form of menkes disease: a 34 year progress report on the original case. *JIMD Reports*, 9, 81-84. [More Information]

2012

Grunert, S., Stucki, M., Morscher, R., Suormala, T., Burer, C., Burda, P., Christensen, E., Ficicioglu, C., Herwig, J., Kolker, S., et al (2012). 3-Methylcrotonyl-CoA Carboxylase Deficiency: Clinical, biochemical, enzymatic and molecular studies in 88 individuals. *Orphanet Journal of Rare Diseases*, 7(1), 1-24. [More Information]

Wilson, C., Kerruish, N., Wilcken, B., Wiltshire, E., Bendikson, K., Webster, D. (2012). Diagnosis of disorders of intermediary metabolism in New Zealand before and after expanded newborn screening: 2004-2009. *New Zealand Medical Journal*, 125(1348), 42-50. [More Information]

Alodaib, A., Carpenter, K., Wiley, V., Wotton, T., Christodoulou, J., Wilcken, B. (2012). Homocysteine Measurement in Dried Blood Spot for Neonatal Detection of Homocystinurias. *JIMD Reports*, 5, 1-6.

Alodaib, A., Carpenter, K., Wiley, V., Wotton, T., Christodoulou, J., Wilcken, B. (2012). Homocysteine Measurement in Dried Blood Spot for Neonatal Detection of Homocystinurias. In Society for the Study of Inborn Errors of Metabolism (Eds.), *JIMD Reports - Case and Research Reports, 2012/2*, (pp. 1-6). Heidelberg: Springer. [More Information]

Wilcken, B. (2012). Leukoencephalopathies Associated with Disorders of Cobalamin and Folate Metabolism. *Seminars in Neurology*, 32(1), 68-74. [More Information]

Fletcher, J., Wilcken, B. (2012). Neonatal screening for lysosomal storage disorders. *The Lancet*, 379(9813), 294-295. [More Information]

Bennett, M., Rinaldo, P., Wilcken, B., Pass, K., Watson, M., Wanders, R. (2012). Newborn Screening for Metabolic Disorders: How Are We Doing, and Where Are We Going? *Clinical Chemistry*, 58(2), 234-331. [More Information]

Wilcken, B. (2012). Screening for disease in the newborn: the evidence base for blood-spot screening. *Pathology*, 44(2), 73-79. [More Information]

2011

Alodaib, A., Carpenter, K., Wiley, V., Sim, K., Christodoulou, J., Wilcken, B. (2011). An improved ultra performance liquid chromatography-tandem mass spectrometry method for the determination of alloisoleucine and branched chain amino acids in dried blood samples. *Annals of Clinical Biochemistry*, 48(5), 468-470. [More Information]

Wilcken, B. (2011). Clinical practice and the development of evidence. *Journal of Inherited Metabolic Disease*, 35(1), 3-4. [More Information]

McHugh, D., Cameron, C., Abdenur, J., Abdulrahman, M., Adair, O., Nuaimi, S., Ahlman, H., Allen, J., Antonozzi, I., Archer, S., Wilcken, B., Wiley, V., et al (2011). Clinical validation of cutoff target ranges in newborn screening of metabolic disorders by tandem mass spectrometry: A worldwide collaborative project. *Genetics in Medicine*, 13(3), 230-254. [More Information]

Kolker, S., Christensen, E., Leonard, J., Greenberg, C., Boneh, A., Burlina, A., Burlina, A., Dixon, M., Duran, M., Cazorla, A., Wilcken, B., et al (2011). Diagnosis and management of glutaric aciduria type
I - revised recommendations. *Journal of Inherited Metabolic Disease*, 34(3), 677-694. [More Information]

Wilcken, B. (2011). Ethical issues in genetics. *Journal of Paediatrics and Child Health*, 47(9), 668-671. [More Information]

Wilcken, B., Wiley, V. (2011). Increased iodine deficiency in Victoria, Australia: analysis of neonatal thyroid-stimulating hormone data, 2001 to 2006. *Medical Journal of Australia*, 194(4), 209-210. [More

Information]

Gaskin, K., Wilcken, B. (2011). Long-term outcomes for patients with cystic fibrosis in Australia. *Medical Journal of Australia*, 195(7), 370-371. [More Information]

Bijarnia, S., Wilcken, B., Wiley, V. (2011). Newborn screening for congenital hypothyroidism
in very-low-birth-weight babies: the need for a second test. *Journal of Inherited Metabolic Disease*, 34(3), 827-833. [More Information]

Wilcken, B. (2011). Newborn screening: how are we travelling, and where should we be going? *Journal of Inherited Metabolic Disease*, 34(3), 569-574. [More Information]

2010

Wortmann, S., Kremer, B., Graham, A., Willemsen, M., Loupatty, F., Hogg, S., Engelke, U., Kluijtmans, L., Wanders, R., Illsinger, S., Wilcken, B., et al (2010). 3-Methylglutaconic Aciduria Type I Redefined: A Syndrome with Late-Onset Leukoencephalopathy. *Neurology*, 75(12), 1079-1083. [More Information]

Balasubramaniam, S., Rudduck, C., Bennetts, B., Peters, G., Wilcken, B., Ellaway, C. (2010). Contiguous gene deletion syndrome in a female with ornithine transcarbamylase deficiency. *Molecular Genetics and Metabolism*, 99(1), 34-41. [More Information]

Spiekerkoetter, U., Bastin, J., Gillingham, M., Morris, A., Wijburg, F., Wilcken, B. (2010). Current Issues Regarding Treatment of Mitochondrial Fatty Acid Oxidation Disorders. *Journal of Inherited Metabolic Disease*, 33(5), 555-561. [More Information]

Wilcken, B. (2010). Expanded newborn screening: reducing harm, assessing benefit. *Journal of Inherited Metabolic Disease*, 33, S205-S210. [More Information]

Wilcken, B. (2010). Fatty acid oxidation disorders: outcome and long-term prognosis. *Journal of Inherited Metabolic Disease*, 33(5), 501-506. [More Information]

Khalid, J., Oerton, J., Besley, G., Dalton, N., Downing, M., Green, A., Henderson, M., Krywawych, S., Wiley, V., Wilcken, B., et al (2010). Relationship of Octanoylcarnitine Concentrations to Age at Sampling in Unaffected Newborns Screened for Medium-Chain Acyl-CoA Dehydrogenase Deficiency. *Clinical Chemistry*, 56(6), 1015-1021. [More Information]

Warne, G., Armstrong, K., Faunce, T., Wilcken, B., Boneh, A., Geelhoed, E., Craig, M. (2010). The case for newborn screening for congenital adrenal hyperplasia in Australia. *Medical Journal of Australia*, 192(2), 107. [More Information]

Willemsen, M., Verbeek, M., Kamsteeg, E., de Rijk-van Andel, J., Aeby, A., Blau, N., Burlina, A., Donati, M., Geurtz, B., Grattan-Smith, P., Wilcken, B., et al (2010). Tyrosine hydroxylase deficiency: a treatable disorder of brain catecholamine biosynthesis. *Brain*, 133(6), 1810-1822. [More Information]

2009

Cunningham, S., Spinoulas, A., Carpenter, K., Wilcken, B., Kuchel, P., Alexander, I. (2009). AAV2/8-mediated Correction of OTC Deficiency Is Robust in Adult but Not Neonatal Spf(ash) Mice. *Molecular Therapy*, 17(8), 1340-1346. [More Information]

Wilcken, B. (2009). Cystic fibrosis: refining the approach to newborn screening. *The Journal of Pediatrics*, 155(5), 605-606. [More Information]

Norman, R., Haas, M., Chaplin, M., Joy, P., Wilcken, B. (2009). Economic evaluation of tandem mass spectrometry newborn screening in Australia. *Pediatrics*, 123(2), 451-457. [More Information]

Wilcken, B., Gaskin, K., Rizzotti, P., Sammon, A., Sands, D., Smyth, A., Sommerburg, O., Torresani, T., Travert, G., Vernooij, A., et al (2009). European best practice guidelines for cystic fibrosis neonatal screening. *Journal of Cystic Fibrosis*. [More Information]

Wilcken, B., Haas, M., Joy, P., Wiley, V., Bowling, F., Carpenter, K., Christodoulou, J., Cowley, D., Ellaway, C., Fletcher, J., et al (2009). Expanded Newborn Screening: Outcome in Screened and Unscreened Patients at Age 6 Years. *Pediatrics (English Edition)*, 124(2), e241-e248. [More Information]

Norman, R., Haas, M., Wilcken, B. (2009). International perspectives on the cost-effectiveness of tandem mass spectrometry for rare metabolic conditions. *Health Policy*, 89(3), 252-260. [More Information]

Joy, P., Black, C., Rocca, A., Haas, M., Wilcken, B. (2009). Neuropsychological functioning in children with medium chain acyl coenzyme a dehydrogenase deficiency (MCADD): The impact of early diagnosis and screening on outcome. *Child Neuropsychology*, 15(1), 8-20. [More Information]

2008

Wilcken, B. (2008). Community-wide screening for cystic fibrosis carriers could replace newborn screening for the diagnosis of cystic fibrosis. *Journal of Paediatrics and Child Health*, 44(4), 232-233; author reply 233. [More Information]

Wilcken, B. (2008). Disorders of the carnitine cycle and detection by newborn screening. *Annals of the Academy of Medicine, Singapore (AAMS)*, 37(12), 71-73. [More Information]

Bijarnia, S., Wiley, V., Carpenter, K., Christodoulou, J., Ellaway, C., Wilcken, B. (2008). Glutaric aciduria type I: outcome following detection by newborn screening. *Journal of Inherited Metabolic Disease*, 31(4), 503-507. [More Information]

Wilcken, B. (2008). Improving child health--newborn screening for all? *Annals of the Academy of Medicine, Singapore (AAMS)*, 37(12), 3. [More Information]

Wilcken, B. (2008). More on medium-chain acyl-coenzyme a dehydrogenase deficiency in a neonate. *New England Journal of Medicine*, 358(6), 647; author reply 647. [More Information]

Hackett, A., Gillard, J., Wilcken, B. (2008). n of 1 trial for an ornithine transcarbamylase deficiency carrier. *Molecular Genetics and Metabolism*, 94(2), 157-161. [More Information]

Wilcken, B., Wiley, V. (2008). Newborn screening. *Pathology*, 40(2), 104-115. [More Information]

Wilcken, B. (2008). Newborn screening for all identifiable disorders with tandem mass spectrometry is cost effective: the negative case. *Annals of the Academy of Medicine, Singapore (AAMS)*, 37(12), 36-33. [More Information]

McKay, K., Wilcken, B. (2008). Newborn screening for cystic fibrosis offers an advantage over symptomatic diagnosis for the long term benefit of patients: the motion for. *Paediatric Respiratory Reviews*, 9(4), 290-294. [More Information]

Wilcken, B. (2008). The consequences of extended newborn screening programmes: Do we know who needs treatment? *Journal of Inherited Metabolic Disease*, 31(2), 173-177. [More Information]

Gleeson, H., Wiley, V., Wilcken, B., Elliott, E., Cowell, C., Thonsett, M., Byrne, G., Ambler, G. (2008). Two-year pilot study of newborn screening for congenital adrenal hyperlasia in New South Wales compared with nationwide case surveillance in Australia. *Journal of Paediatrics and Child Health*, 44(10), 554-559. [More Information]

2007

Vyletal, P., Sokolova, J., Cooper, D., Kraus, J., Krawczak, M., Pepe, G., Rickards, O., Koch, H., Linnebank, M., Kluijtmans, L., Wilcken, B., et al (2007). Diversity of cystathionine beta-synthase haplotypes bearing the most common homocystinuria mutation c.833T>C: a possible role for gene conversion. *Human Mutation*, 28(3), 255-264. [More Information]

Chiong, M., Bennetts, B., Strasser, S., Wilcken, B. (2007). Fatal late-onset ornithine transcarbamylase deficiency after coronary artery bypass surgery. *Medical Journal of Australia*, 186(8), 418-419. [More Information]

Kolker, S., Christensen, E., Leonard, J., Greenberg, C., Burlina, A., Burlina, A., Dixon, M., Duran, M., Goodman, S., Koeller, D., Wilcken, B., et al (2007). Guideline for the diagnosis and management of glutaryl-CoA dehydrogenase deficiency (glutaric aciduria type I). *Journal of Inherited Metabolic Disease*, 30(1), 5-22. [More Information]

Haas, M., Chaplin, M., Joy, P., Wiley, V., Black, C., Wilcken, B. (2007). Healthcare use and costs of medium-chain acyl-CoA dehydrogenase deficiency in Australia: screening versus no screening. *The Journal of Pediatrics*, 151(2), 121-126. [More Information]

Wilcken, B. (2007), *Invited speaker. Expanded Newborn Screening. Groningen February 2007.*

Wilcken, B. (2007), *Invited speaker: Society for the Study of Inborn Errors of MetabolismHamburg, Germany, September 2007: Consequences of extended screening programmes*.

Wilcken, B. (2007), *Invited speaker; International Society for Neonatal Screening - Asia-Pacific regional Meeting, Singapore August 2007 . Keynote address; plus lecture on carnitine cycle disorders.*

Chiong, M., Procopis, P., Wilcken, B. (2007). Late-Onset Nonketotic Hyperglycinemia With Leukodystrophy and an Unusual Clinical Course. *Pediatric Neurology*, 37(4), 283-286. [More Information]

Wilcken, B., Gaskin, K. (2007). More evidence to favour newborn screening for cystic fibrosis. *The Lancet*, 369(9568), 1146-1147. [More Information]

Wilcken, B. (2007). Newborn screening for cystic fibrosis: Techniques and strategies. *Journal of Inherited Metabolic Disease*, 30(4), 537-543. [More Information]

Tan, E., Wiley, V., Carpenter, K., Wilcken, B. (2007). Non-Ketotic Hyperglycinemia is usually not detectable by tandem mass spectrometry newborn screening. *Molecular Genetics and Metabolism*, 90(4), 446-448. [More Information]

Wilcken, B., Haas, M., Joy, P., Wiley, V., Chaplin, M., Black, C., Fletcher, J., McGill, J., Boneh, A. (2007). Outcome of neonatal screening for medium-chain acyl-CoA dehydrogenase deficiency in Australia: a cohort study. *The Lancet*, 369(9555), 37-42. [More Information]

Cipolli, M., Castellani, C., Wilcken, B., Massie, J., McKay, K., Gruca, M., Tamanini, A., Assael, M., Gaskin, K. (2007). Pancreatic phenotype in cystic fibrosis patients identified by mutation screening. *Archives of Disease in Childhood*, 92(10), 842-846. [More Information]

Cipolli, M., Castellani, C., Wilcken, B., Massie, J., McKay, K., Gruca, M., Tamanini, A., Assael, M., Gaskin, K. (2007). Pancreatic phenotype in infants with cystic fibrosis identified by mutation screening. *Archives of Disease in Childhood*,

92(10), 842-846. [More Information]

Wilcken, B. (2007). Recent advances in newborn screening. *Journal of Inherited Metabolic Disease*, 30(2), 129-133. [More Information]

Schindeler, S., Ghosh-Jerath, S., Thompson, S., Rocca, A., Joy, P., Kemp, A., Rae, C., Green, K., Wilcken, B., Christodoulou, J. (2007). The effects of large neutral amino acid supplements in PKU: An MRS and neuropsychological study. *Molecular Genetics and Metabolism*, 91, 48-54. [More Information]

Wilson, C., Kerruish, N., Wilcken, B., Wiltshire, E., Webster, D. (2007). The failure to diagnose inborn errors of metabolism in New Zealand: the case for expanded newborn screening. *New Zealand Medical Journal*, 120(1262), U2727-U2727. [More Information]

Cliffe, S., Wong, M., Taylor, P., Ruga, E., Wilcken, B., Lindeman, R., Buckley, M., Roscioli, T. (2007). The first prenatal diagnosis for veno-occlusive disease and immunodeficiency syndrome, an autosomal recessive condition associated with mutations in SP110. *Prenatal Diagnosis*, 27(7), 674-676. [More Information]

2006

Dionisi-Vici, C., Deodato, F., Roschinger, W., Rhead, W., Wilcken, B. (2006). 'Classical' organic acidurias, propionic aciduria, methylmalonic aciduria and isovaleric aciduria: Long-term outcome and effects of expanded newborn screening using tandem mass spectrometry. *Journal of Inherited Metabolic Disease*, 29(2-3), 383-389. [More Information]

Wopereis, S., Abd Hamid, U., Critchley, A., Royle, L., Dwek, R., Morava, E., Leroy, J., Wilcken, B., Lagerwerf, A., Huijben, K., et al (2006). Abnormal glycosylation with hypersialylated O-glycans in patients with Sialuria. *Biochimica et Biophysica Acta*, 1762 (6), 598-607. [More Information]

Wilcken, D., Wang, J., Sim, A., Green, K., Wilcken, B. (2006). Asymmetric dimethylarginine in homocystinuria due to cystathionine beta-synthase deficiency: relevance of renal function. *Journal of Inherited Metabolic Disease*, 29(1), 30-37. [More Information]

Wilcken, B. (2006). Disorders of Sulfur Amino Acid Metabolism. In Nenad Blau, Georg F. Hoffmann, James Leonard, Joe T.R. Clarke (Eds.), *Physician's Guide to the Treatment and Follow-up of Metabolic Diseases*, (pp. 105-116). Berlin: Springer.

Wilcken, B. (2006), *Invited plenary speaker, International Congress on Human Genetics, Chiba, Japan September 2006*.

Wilcken, B. (2006), *Invited speaker "Genomics and Public Health" Montreal June 3rd - 6th. "Newborn screening, achievements and future directions"*.

Wilcken, B. (2006), *Invited speaker International Society of Neonatal Screening , Tokushima, Japan September 2006*.

Travers, C., Guttikonda, K., Norton, C., Lewis, P., Mollart, L., Wiley, V., Wilcken, B., Eastman, C., Boyages, S. (2006). Iodine status in pregnant women and their newborns: are our babies at risk of iodine deficiency? *Medical Journal of Australia*, 184(12), 617-620. [More Information]

Wilcken, B. (2006). Lactation conference NSW College of Nursing. Invited speaker "Medium-chain acyl-CoA dehydrogenase deficiency". *Lactation conference*, Not Published: Not Published.

Waddell, L., Wiley, V., Carpenter, K., Bennetts, B., Angel, L., Andresen, B., Wilcken, B. (2006). Medium-chain acyl-CoA dehydrogenase deficiency: genotype-biochemical phenotype correlations. *Molecular Genetics and Metabolism*, 87(1), 32-39. [More Information]

Wilcken, B. (2006). Mini-Symposium: Newborn screening for inborn errors of metabolism-Clinical effectiveness. *Journal of Inherited Metabolic Disease*, 29, 366-369. [More Information]

Wilcken, B. (2006). Newborn Screening for Inborn Errors of Metabolism. In J. Fernandes, J-.M. Saudubray, G. van den Berghe, J.H. Walter (Eds.), *Inborn Metabolic Disease â€“ Diagnosis and Treatment 4th Ed*, (pp. 49-58). Germany: Springer.

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]

Wilcken, B. (2006). Short Course in Genetics; (invited speaker). March 17-18th. *Short Course in Genetics*, Not Published: Not Published.

Wilcken, B. (2006). Systematic Screening. In John Fernandes, J.-M. Saudubray, G. van den Berghe, J.H. Walter (Eds.), *Inborn Metabolic Disease â€“ Diagnosis and Treatment*, (pp. 50-57). Springer.

Wilcken, B. (2006). Wyeth conference: "Update in paediatrics"; invited speaker. May 4th. *Wyeth conference*, Not Published: Not Published.

2005

Bayliss, U., Cowell, C., Hong, J., Wiley, V., Wilcken, B. (2005). Acute presentation of childhood hypothyroidism. *Medical Journal of Australia*, 182(4), 200-200. [More Information]

Wilcken, B. (2005). Disorders of sulphur amino acid metabolism. In Nenad Blau, Georg F. Hoffmann, James Leonard, Joe T.R. Clarke (Eds.), *Physician's Guide to the Treatment and Follow-up of Metabolic Diseases*, (pp. 105-115). Berlin: Springer.

Wilcken, B. (2005). Halpern Symposium: Wollongong University, Department of Chemistry. "A very fruitful collaboration" November; invited speaker. *Halpern Symposium*, Not Published: Not Published.

Wilcken, B. (2005), *Invited speaker: Fatty Acid Oxidation Symposium: "Newborn screening for fatty acid oxidation defects" June. Amsterdam*.

Wilcken, B. (2005), *Invited speaker: Taiwan Rare disorders*

conference, Taipeh December.

Wilcken, B. (2005), *Invited to organize and speak at a symposium "Clinical effectiveness of newborn screening" at the Society for the Study of Inborn Errors of metabolism annual scientific meeting. Paris. September*.

McElduff, A., McElduff, P., Wiley, V., Wilcken, B. (2005). Neonatal thyrotropin as measured in a congenital hypothyroidism screening program: influence of the mode of delivery. *The Journal of Clinical Endocrinology and Metabolism*, 90(12), 6361-6363. [More Information]

Wilcken, B. (2005). Symposium: "Embryo, fetus and newborn: from experimental to clinical practice in the Antipodes" RHW Sydney May. *RHW Symposium*, Not Published: Not Published.

Mitchell, J., Wilcken, B., Alexander, I., Ellaway, C., O'Grady, H., Wiley, V., Earl, J., Christodoulou, J. (2005). Tetrahydrobiopterin-responsive phenylketonuria: The New South Wales experience. *Molecular Genetics and Metabolism*, 86(Suppl 1), S81-S85. [More Information]

Dobrowolski, S., McKinney, J., Amat di San Filippo, C., Sim, K., Wilcken, B., Longo, N. (2005). Validation of dye-binding/high-resolution thermal denaturation for the identification of mutations in the SLC22A5 gene. *Human Mutation*, 25(3), 306-313. [More Information]

2004

Christodoulou, J., Wilcken, B. (2004). Biochemical Genetic Emergencies. In Henry Kilham & David Isaacs (Eds.), *The Childrens Hospital at Westmead Handbook: Clinical practice guidelines for paediatrics*, (pp. 104-111). Sydney: McGraw-Hill Education.

Wilcken, B. (2004). Invited speaker GP weekend. CHW. "Update on Newborn Screening". *GP weekend. CHW*, Not Published: Not Published.

Wilcken, B. (2004). Invited speaker, HGSA, Perth August. "Tandem mass spectrometry". *HGSA*, Not Published: Not Published.

Wilcken, B. (2004), *Invited speaker: 5th Asian-Pacific Meeting of the International Society for Neonatal Screening, Shanghai. "The future of newborn screening" and "Neonatal screening in Australia"*.

Wilcken, B. (2004). Invited speaker: Royal Australasian College of Physicians Annual Scientific Meeting, Canberra. "Newborn screening by tandem mass spectrometry: Ethical issues uncovered". *Royal Australasian College of Physicians Annual Scientific Meeting*. British Medical Journal Publishing Group.

Wilcken, B. (2004), *Invited speaker: Society for the Study of Inborn Errors of Metabolism, Amsterdam, September. "Newborn screening by tandem mass spectrometry, quality assurance"*.

Christodoulou, J., Wilcken, B. (2004). Perimortem Laboratory Investigation Of Genetic Metabolic Disorders. *Seminars In Neonatology*, 9(4), 275-280. [More Information]

Wilcken, B. (2004). Problems in the management of urea cycle disorders. *Molecular Genetics and Metabolism*, 81, S86-S91. [More Information]

Wilcken, B. (2004). Screening of Newborns for Metabolic Disorders With Mass Spectrometry. *JAMA - Journal of the American Medical Association*, 291(12), 1444-1445.

2003

Wilcken, B. (2003). An Introduction to Nutritional Treatment in Inborn Errors of Metabolism - different disorders, different approaches. *Southeast Asian Journal of Tropical Medicine and Public Health*, 34(S3), 198-201.

Wilcken, B. (2003). Chair of Organising Committee: XIth International Congress on Inborn Errors of Metabolism: Brisbane. *IXth International Congress on Inborn Errors of Metabolism*, Published: Not Published.

Wilcken, B. (2003). Does every baby get a newborn screening test? *Medical Journal of Australia*, 11, 400-401.

Wilcken, B. (2003). Ethical issues in newborn screening and the impact of new technologies. *European Journal of Pediatrics*, 162(Supplement 1), S62-S66.

Wilcken, B. (2003). Evaluating outcomes of newborn screening programs. *Southeast Asian Journal of Tropical Medicine and Public Health*, 34(3), 13-18.

Urwin, R., Bennetts, B., Wilcken, B., Lampropoulos, B., Beumont, P., Russell, J., Tanner, S., Nunn, K. (2003). Gene-gene interaction between the monoamine oxidase A gene and solute carrier family 6 (neurotransmitter transporter, noradrenalin) member 2 gene in anorexia nervose (restrictive subtype). *European Journal of Human Genetics*, 11(12), 945-950. [More Information]

Wilcken, B., Bamforth, F., Li, Z., Zhu, H., Ritvanen, A., Redlund, M., Stoll, C., Alembik, Y., Dott, B., Czeizel, A., et al (2003). Geographical and ethnic variation of the 677C>T allele of 5, 10 methylenetetrahydrofolate reductase (MTHFR): findings from over 7000 newborns from 16 areas world wide. *Journal of Medical Genetics*, 40(8), 619-625.

Therrell, B., Wilcken, B., Naruse, H. (2003). History of the International Society for Neonatal Screening. *Southeast Asian Journal of Tropical Medicine and Public Health*, 34(Supplement 3), 3-5.

Wiley, V., Carpenter, K., Bennetts, B., Wilcken, B. (2003). Information overload - new technologies, can we store the data? *Southeast Asian Journal of Tropical Medicine and Public Health*, 34(S3), 59-62.

Urwin, R., Bennetts, B., Wilcken, B., Beumont, P., Russell, J., Nunn, K. (2003). Investigation of epistasis between the serotonin transporter and norepinephrine transproter genes in anorexia nervosa. *Neuropsychopharmacology*, 28(7), 1351-1355. [More Information]

Wilcken, B. (2003), *Invited discussant Centre for Diseases Control Workshop on Neonatal Screening for Cystic Fibrosis: Atlanta, Georgia, USA; November 2003*.

Wilcken, B. (2003). Invited speaker, STEP lectures, School of Public Health, Sydney University "Newborn screening: evaluating candidate disorders". *STEP lectures, School of Public Health*.

Wilcken, B. (2003), *Invited speaker- Society for Inherited Metabolic Disease and Conference on Tandem Mass Spectrometry: Berkeley, CA, USA; January 2003. I was one of only two non-US people invited. "Confirmatorty testing" and*

"Evaluation of MSMS Screening".

Wilcken, B. (2003). Invited speaker: Annual Scientific Meeting Human Genetics Society of Australasia, Adelaide, oral presentation on tandem mass spectrometry screening. *Annual Scientific Meeting Human Genetics Society of Australasia*.

Wilcken, B. (2003). Invited speaker: Annual Scientific Meeting of the RCPA Sydney March 2003. "Perimortem investigation of inborn errors of metabolism". *Annual Scientific Meeting of the RCPA*.

Wilcken, B. (2003). Invited speaker: ANZ Childrenâ€™s Cancer Study Group- Sydney May 2003 â€“ "Prevention: neonatal and prenatal screening". *ANZ Children's Cancer Study Group*.

Wilcken, B. (2003). Invited speaker: ANZ Cystic Fibrosis Conference, Melbourne, July 2003."Update on screening for cystic fibrosis". *ANZ Cystic Fibrosis Conference*.

Wilcken, B. (2003). Invited speaker: Festshrift for Professor Kamath. Sydney, August 2003. "Inborn errors and liver disease". *Festshrift for Professor Kamath*.

Wilcken, B. (2003). Invited speaker: New developments in Urea Cycle Disease, (Satellite meeting of the ICIEM). Manly August 2003. "Problems in clinical management of urea cycle disorders". *New developments in Urea Cycle Disease, (Satellite meeting of the ICIEM)*, Not Published: Not Published.

Nga Ly, T., Peters, V., Gibson, K., Liesert, M., Buckel, W., Wilcken, B., Carpenter, K., Ensenauer, R., Hoffmann, G., Mack, M., et al (2003). Mutations in the AUH gene cause 3-Methylglutoconic aciduria Type I. *Human Mutation*, 21(4), 401-407. [More Information]

Wiley, V., Carpenter, K., Bayliss, U., Wilcken, B. (2003). Newborn screening - is it really that simple? *Southeast Asian Journal of Tropical Medicine and Public Health*, 34(S3), 107-110.

Wilcken, B., Wiley, V. (2003). Newborn screening methods for cystic fibrosis. *Paediatric Respiratory Reviews*, 11, 272-277. [More Information]

Chan, S., Wilcken, B., Wiley, V., McElduff, A. (2003). Postpartum maternal iodine status and the relationship to neonatal thryroid function. *Thyroid*, 13(9), 873-876. [More Information]

Wilcken, B., Wiley, V., Hammond, J., Carpenter, K. (2003). Screening newborns for inborn errors of metabolism by tandem mass spectrometry. *New England Journal of Medicine*, 348(5), 2304-2312. [More Information]

2002

Pitt, J., Carpenter, K., Wilcken, B., Boneh, A. (2002). 3-hydroxyglutarate excretion is increased in keototic patients: implications for glutaryl-CoA dehydrogenase deficiency testing. *Journal of Inherited Metabolic Disease*, 25(2), 83-88. [More Information]

Sim, K., Carpenter, K., Hammond, J., Christodoulou, J., Wilcken, B. (2002). Acylcarnitine profiles in fibroblasts from patients with respiratory chain defects can resemble those from patients with mitochondrial fatty acid Oxidation disorders. *Metabolism: Clinical and Experimental*, 51(3), 366-371. [More Information]

Urwin, R., Bennetts, B., Wilcken, B., Lampropolous, B., Beumont, P., Clarke, S., Russell, J., Tanner, S., Nunn, K. (2002). Anorexia nervosa (restrictive subtype) is associated with a polymorphism in the novel noreprinephrine transporter gene promoter polymorphic region. *Molecular Psychiatry*, 7(6), 652-657. [More Information]

Potter, S., Lu, A., Wilcken, B., Green, A., Rasko, J. (2002). Hartnup disorder: Polymorphisms identified in the neutral amino acid transporter SLC1A5. *Journal of Inherited Metabolic Disease*, 25(6), 437-448. [More Information]

Wilcken, B. (2002), *Invited debate moderator: Society for the Study of Inborn Errors of Metabolism, Dublin Ireland "Treatment should be available to all"*.

Wilcken, B. (2002). Invited speaker Royal College of Pathologists ASM â€“ Tandem mass spectrometry in the diagnosis of metabolic disease. *Royal College of Pathologists ASM*.

Wilcken, B. (2002), *Invited speaker, International Child Neurology Congress Beijing "Screening for neurogenetic disorders"*.

Wilcken, B. (2002), *Invited speaker: 5th Annual Scientific Meeting, The International Society for Neonatal Screening. Genoa. "Heterozygote detection"*.

Wilcken, B. (2002). Invited speaker: Australian Institute of Medical Scientists Annual Scientific meeting Sydney. "Hyperhomocysteinaemia". *Australian Institute of Medical Scientists Annual Scientific meeting*.

Wilcken, B., Carpenter, K., Wiley, V. (2002). Neonatal screening for medium-chain acyl-CoA dehydrogenase deficiency. *The Lancet (North American Edition)*, 359(9306), 627-628. [More Information]

McElduff, A., McElduff, P., Gunton, J., Hams, G., Wiley, V., Wilcken, B. (2002). Neonatal thyroid-stimulating hormone concentrations in northern Sydney: further indications of mild iodine deficiency. *Medical Journal of Australia*, 176(7), 317-320.

Ellaway, C., Wilcken, B., Christodoulou, J. (2002). Neonatology for the Generalist: Clinical approach to inborn errors of metabolism presenting in the newborn period. *Journal of Paediatrics and Child Health*, 38(5), 511-517. [More Information]

Sim, K., Carpenter, K., Hammond, J., Christodoulou, J., Wilcken, B. (2002). Quantitative fibroblasts acylcarnitine profiles in mitochondrial fatty acid beta-oxidation defects: phenotype/metabolite correlations. *Molecular Genetics and Metabolism*, 76(4), 327-334. [More Information]

Sim, K., Hammond, J., Wilcken, B. (2002). Strategies for the diagnosis of mitochondrial fatty acid beta-oxidation disorders. *Clinica Chimica Acta*, 323(1-2), 37-58. [More Information]

Gaustadnes, M., Wilcken, D., Oliveriusova, J., McGill, J., Fletcher, J., Kraus, J., Wilcken, B. (2002). The molecular basis of cystathionine Beta-synthase deficiency in Australian patients: Genotype-Phenotype correlations and response to treatment. *Human Mutation*, 20(2), 117-126. [More

Information]

Grattan-Smith, P., Wevers, R., Steenbergen-Spanjers, G., Fung, V., Earl, J., Wilcken, B. (2002). Tyrosine hydroxylase deficiency: Clinical manifestations of catecholamine insufficiency in infancy. *Movement Disorders*, 17(2), 354-359. [More Information]

2001

McMaster,, P., Hunt,, R., Wojtulewicz, J., Wilcken, B. (2001). An unusual cause of hepatitis. *Journal of Paediatrics and Child Health*, 37(6), 587-588.

Sim, K., Wilcken, B., Wiley, K., Carpenter, K. (2001). Carnitine palmitoyltransferase I deficiency in neonate identified by dried blood spot free carnitine and acylcarnitine profile. *Journal of Inherited Metabolic Disease*, 24, 51-59.

Wilcken, B., Sim, K., Wiley, V., Carpenter, K. (2001). Carnitine transporter defect diagnosed by newborn screening with electrospray tandem mass spectrometry. *The Journal of Pediatrics*, 138, 581-584.

Sim, K., Wilcken, B., Carpenter, K., Wiley, V. (2001). Evaluation of newborn screening for medium chain acyl-CoA dehydrogenase deficiency in 275,000 babies. *Archives of Disease in Childhood*, 85.

Wilcken, D., Wilcken, B. (2001). Homocysteine: Historical overview and current issues. In Ralph Carmel and Donald W. Jacobsen (Eds.), *Homocysteine in Health and Disease*. United States: Cambridge University Press.

Wilcken, B. (2001). Internal Medicine Society of Australia and New Zealand, Sydney, "Hyperhomocysteinaemia". *Internal Medicine Society of Australia and New Zealand*, Not Published: Not Published.

Massie, J., Poplawski, N., Wilcken, B., Robertson, C., Byrnes, C., Goldblatt, J. (2001). Intron-8 polythymidine sequence in Australasian individuals with CF mutations R117H and R117C. *European Respiratory Journal*, 17, 1195-1200.

Wilcken, B. (2001), *Invited speaker, 4th Asia-Pacific Meeting of the International Society for Neonatal Screening. "Evaluating Outcomes in Newborn Screening" and "Nutrition in the treatment of inborn errors of metabolism" Manila, Philippines*.

Wilcken, B. (2001), *Invited speaker, International symposium: The Early Diagnosis of Inherited Metabolic Disease. Fulda, Germany: "Ethics in Screening"*.

Wilcken, B. (2001), *Invited speaker, South African Paediatric Association Annual Scientific Meeting Sun City, SA. "Place of Newborn Screening in Health Delivery"*.

Wilcken, B. (2001). Invited speaker: Royal Australasian College of Physicians: May: Human Genome Project - prevention. *Royal Australasian College of Physicians Conference*.

Sim, K., Green, A., Wilcken, B., Potter, M., Hammond, J. (2001). Ornithine carbamoyltransferase deficiency: Improved sensitivity of testing for protein tolerance in the diagnosis of heterozygotes. *Journal of Inherited Metabolic Disease*, 24, 5-14.

Hogema, B., Akaboshi, S., Taylor, M., Salomons, G., Jakobs, C., Schutgens, R., Wilcken, B., Worthington, S., Maropoulos, G., Grompe, M., et al (2001). Prenatal Diagnosis of Succinic Semialdehyde Dehydrogenase Deficiency: Increased Accuracy Employing DNA, Enzyme, and Metabolite Analyses. *Molecular Genetics and Metabolism*, 72, 218-222.

Wilcken, B. (2001). Rare diseases and the assessment of intervention: What sorts of clinical trials can we use? *Journal of Inherited Metabolic Disease*, 24, 291-298.

Yap, S., Boers, G., Wilcken, B., Wilcken, D., Brenton, D., Lee, K., Walter, J., Howard, P., Naughten, E. (2001). Vascular outcome in patients with homocystinuria due to cystathionine beta-synthase deficiency treated chronically: a multicenter observational study. *Arteriosclerosis, Thrombosis, and Vascular Biology*, 21, 2080-2085.

2000

Carpenter, K., Wilcken, B., Christodoulou, J., Thorburn, D. (2000). Holocarboxylase synthetase deficiency: Urinary metabolites masked by gross ketosis. *Journal of Inherited Metabolic Disease*, 23(8), 845-846.

Massie, J., Wilcken, B., Van Asperen, P., Dorney, S., Gruca, M., Wiley, V., Gaskin, K. (2000). Pancreatic function and extended mutation analysis in ?F508 heterozygous infants with ane levated immunoreactive trypsinogen but normal sweat electrolyte levels. *The Journal of Pediatrics*, 137(2), 214-220.