

Dr Helen Mundy's publications

ELAC2 mutations cause a mitochondrial RNA processing defect associated with hypertrophic cardiomyopathy.

Haack TB, Kopajtich R, Freisinger P, Wieland T, Rorbach J, Nicholls TJ, Baruffini E, Walther A, Danhauser K, Zimmermann FA, Husain RA, Schum J, **Mundy H**, Ferrero I, Strom TM, Meitinger T, Taylor RW, Minczuk M, Mayr JA, Prokisch H.

Am J Hum Genet. 2013 Aug 8;93(2):211-23. doi: 10.1016/j.ajhg.2013.06.006. Epub 2013 Jul 11.

Thirteen new patients with guanidinoacetate methyltransferase deficiency and functional characterization of nineteen novel missense variants in the GAMT gene.

Mercimek-Mahmutoglu S, Ndika J, Kanhai W, de Villemeur TB, Cheillan D, Christensen E, Dorison N, Hannig V, Hendriks Y, Hofstede FC, Lion-Francois L, Lund AM, Mundy H, Pitelet G, Raspall-Chaure M, Scott-Schworer JA, Szakszon K, Valayannopoulos V, Williams M, Salomons GS. Hum Mutat. 2014 Apr;35(4):462-9. doi: 10.1002/humu.22511. Epub 2014 Mar 6

Molybdenum cofactor deficiency presenting with a parkinsonism-dystonia syndrome. Alkufri F, Harrower T, Rahman Y, Hughes E, Mundy H, Knibb JA, Moriarty J, Connor S, Samuel M. Mov Disord. 2013 Mar;28(3):399-401.

Liver transplantation for propionic acidemia in children. Vara R, Turner C, Mundy H, Heaton ND, Rela M, Mieli-Vergani G, Champion M, Hadzic N. Liver Transpl. 2011 Jun;17(6):661-7.

Adenylosuccinate lyase deficiency in the United Kingdom pediatric population: first three cases. Lundy CT, Jungbluth H, Pohl KR, Siddiqui A, Marinaki AM, Mundy H, Champion MP. Pediatr Neurol. 2010 Nov;43(5):351-4.

The pathogenic m.3243A>T mitochondrial DNA mutation is associated with a variable neurological phenotype. Alston CL, Bender A, Hargreaves IP, Mundy H, Deshpande C, Klopstock T, McFarland R, Horvath R, Taylor RW. Neuromuscul Disord. 2010 Jun;20(6):403-6.

Glucose transporter-1 deficiency syndrome: the expanding clinical and genetic spectrum of a treatable disorder. Leen WG, Klepper J, et al. Brain. 2010 Mar;133(Pt 3):655-70.

A novel mitochondrial MTND5 frameshift mutation causing isolated complex I deficiency, renal failure and myopathy. Alston CL, Morak M, Reid C, Hargreaves IP, Pope SA, Land JM, Heales SJ, Horvath R, Mundy H, Taylor RW. Neuromuscul Disord. 2010 Feb;20(2):131-5.

Reduction in bone mineral density in glycogenosis type III may be due to a mixed muscle and bone deficit. Mundy HR, Williams JE, Lee PJ, Fewtrell MS. J Inherit Metab Dis. 2008 Jun;31(3):418-23. Epub 2008 Apr 4.

A novel starch for the treatment of glycogen storage diseases. Bhattacharya K, Orton RC, Qi X, Mundy H, Morley DW, Champion MP, Eaton S, Tester RF, Lee PJ. J Inherit Metab Dis. 2007 Jun;30(3):350-7. Epub 2007 May 19

The effect of L-alanine therapy in a patient with adult onset glycogen storage disease type II. Mundy HR, Williams JE, Cousins AJ, Lee PJ. J Inherit Metab Dis. 2006 Feb;29(1):226-9.

Exercise capacity and biochemical profile during exercise in patients with glycogen storage disease type I. Mundy HR, Hindmarsh PC, Matthews DR, Leonard JV, Lee PJ Clin Endocrinol (Oxf). 2003 Mar;58(3):332-9.

The regulation of growth in glycogen storage disease type 1. Mundy HR, Hindmarsh PC, Matthews DR, Leonard JV, Lee PJ. Clin Endocrinol (Oxf). 2003 Mar;58(3):332-9.

Images in cardiovascular medicine. Myocardial fibrosis in glycogen storage disease type III. Moon JC, Mundy HR, Lee PJ, Mohiaddin RH, Pennell DJ. Circulation. 2003 Feb 25;107(7):e47.

Dietary control of phenylketonuria. Mundy H, Lilburn M, Cousins A, Lee P. Lancet. 2002 Dec 21-28;360(9350):2076

A child presenting with disordered consciousness, hallucinations, screaming episodes and abdominal pain. Hussain K, Mundy H, Aynsley-Green A, Champion M. Eur J Pediatr. 2002 Feb;161(2):127-9.