

## Dr Elizabeth Wraige's publications

Clinical and genetic findings in a large cohort of patients with ryanadine receptor 1 gene-associated myopathies. Klein A, Lillis S, Muteanu I, Scoto M, Zhou H, Quinlivian R, Straub V, Manzur AY, Roper H, Jeannet PY, Rakowicz W, Jones DH, Jensen UB, **Wraige E**, Trump N, Schara U, Lochmuller H, Sarkozy a, Kingston H, Norwood F, Damina M, Kirschner J, Longman C, Roberts M, Auer-Grumbach M, Hughes I, Bushby K, Sewry C, Robb S, Abbs S, Jungbluth H, Muntoni F. *Hum Mutat* 2012 Jun;33(6):981-8

Generalized arterial calcification of infancy and pseudoxanthoma elasticum can be caused by mutations in either ENPP1 or ABCC6. Nitschke Y, Baujat G, Botschen U, Wittkamp T, du Moulin M, Stella J, Le Merrer M, Guest G, Lambot K, Tazarourte-Pinturier MF, Chassaing N, Roche O, Feenstra I, Loechner K, Deshpande C, Garber SJ, Chikarmane R, Steinmann B, Shahinyan T, Martorell L, Davies J, Smith WE, Kahler SG, McCulloch M, **Wraige E**, Loidi L, Höhne W, Martin L, Hadj-Rabia S, Terkeltaub R, Rutsch F. *Am J Hum Genet.* 2012 Jan 13; 90(1):25-29

Beneficial use of steroids in hereditary neuropathy with liability to pressure palsy. Heng HS, Tang SS, Goyal S, **Wraige EA**, Lim MJ. *Dev Med and Child Neurol.* 2012 feb;54(2):183-6

Muscle magnetic resonance imaging in congenital myopathies due to ryanodine receptor type 1 gene mutations. Klein A, Jungbluth H, Clement E, Lillis S, Abbs S, Munot P, Pane M, **Wraige E**, Schara U, Straub V, Mercuri E, Muntoni F. *Arch Neurol.* 2011 Sep;68(9):1171-9.

King-Denborough syndrome with and without mutations in the skeletal muscle ryanodine receptor gene. Dowling JJ, Lillis S, Amburgey K, Zhou H, Al-Sarraj S, Buk SJ, **Wraige E**, Chow G, Abbs S, Leber S, Lachlan K, Baralle D, Taylor A, Sewry C, Muntoni F, Jungbluth H. *Neuromuscul Disord.* 2011 Jun;21(6):420-7. Epub 2011 Apr 22.

Congenital lower brachial plexus palsy due to cervical ribs Desurkar A, Mills K, Pitt M, Jan W, Sinisi M, Male I, **Wraige E**. *Dev Med Child Neurol.* 2011 Feb;53(2):188-90. doi: 10.1111/j.1469-8749.2010.03841.x. Erratum in: *Dev Med Child Neurol.* 2011 May;53(5):478. Sinisi, Marco [corrected to Sinisi, Marco].

Infantile onset myofibrillar myopathy due to recessive CRYAB mutations Forrest KM, Al-Sarraj S, Sewry C, Buk S, Tan SV, Pitt M, Durward A, McDougall M, Irving M, Hanna MG, Matthews E, Sarkozy A, Hudson J, Barresi R, Bushby K, Jungbluth H, **Wraige E**. *Neuromuscul Disord.* 2011 Jan;21(1):37-40. Epub 2010 Dec 3.

Clinical and molecular characterisation of hereditary dopamine transporter deficiency syndrome: an observational cohort and experimental study Kurian MA, Li Y, Zhen J, Meyer E, Hai N, Christen HJ, Hoffmann GF, Jardine P, von

Moers A, Mordekar SR, O'Callaghan F, Wassmer E, **Wraige** E, Dietrich C, Lewis T, Hyland K, Heales S Jr, Sanger T, Gissen P, Assmann BE, Reith ME, Maher ER. *Lancet Neurol*. 2011 Jan;10(1):54-62. Epub 2010 Nov 25.

Basilar artery dolichoectasia in childhood: evidence of vascular compromise  
Forrest KM, Siddiqui A, Lim M, **Wraige** E. *Childs Nerv Syst*. 2011 Jan;27(1):193-6. Epub 2010 Aug 28

Congenital insensitivity to pain: novel SCN9A missense and in-frame deletion mutations  
Cox JJ, Sheynin J, Shorer Z, Reimann F, Nicholas AK, Zubovic L, Baralle M, **Wraige** E, Manor E, Levy J, Woods CG, Parvari R. *Hum Mutat*. 2010 Sep;31(9):E1670-86.

Charcot-Marie-Tooth (CMT) disease 1A with superimposed inflammatory polyneuropathy in children. Desurkar A, Lin JP, Mills K, Al-Sarraj S, Jan W, Jungbluth H, **Wraige** E. *Neuropediatrics*. 2009 Apr;40(2):85-8. Epub 2009 Oct 6. □

Generalized arterial calcification of infancy: phenotypic spectrum among three siblings including one case without obvious arterial calcifications. Dlamini N, Splitt M, Durkan A, Siddiqui A, Padayachee S, Hobbins S, Rutsch F, **Wraige** E. *Am J Med Genet A*. 2009 Mar;149A(3):456-60.

Interfamilial phenotypic heterogeneity in SMARD1. Joseph S, Robb SA, Mohammed S, Lillis S, Simonds A, Manzur AY, Walter S, **Wraige** E. *Neuromuscul Disord*. 2009 Mar;19(3):193-5. Epub 2009 Jan 20.

Surgery for scoliosis in Duchenne muscular dystrophy. Cheuk DK, Wong V, **Wraige** E, Baxter P, Cole A, N'Diaye T, Mayowe V. *Cochrane Database Syst Rev*. 2007 Jan 24;(1):CD005375. Review.

A proposed classification for subtypes of arterial ischaemic stroke in children. **Wraige** E, Pohl KR, Ganesan V. *Dev Med Child Neurol*. 2005 Apr;47(4):252-6.