Leukonet publication list

Cerebral gray and white matter changes and clinical course in metachromatic leukodystrophy
Groeschel S, í Dali C, Clas P, Böhringer J, Duno M, Krarup C, Kehrer C, Wilke M, Krägeloh-Mann I. Neurology. 2012 Oct 16;79(16):1662-70. doi: 10.1212/WNL.0b013e31826e9ad2. Epub 2012 Sep 19.

Juvenile metachromatic leukodystrophy 10 years post-transplant compared to a non-transplanted cohort
Krägeloh-Mann I, Groeschel S, Kehrer C, Opherk K, Nägele T, Handgretinger R, Müller I (2012), BMT in press

Systematic approaches to central nervous system myelin. Cellular and Molecular Life Sciences
de Monasterio-Schrader P, Jahn O, Tenzer S, Wichert SP, Patzig J, Werner HB (2012) Cell Mol Life Sci. 2012 Sep;69(17):2879-94. doi: 10.1007/s00018-012-0958-9. Epub 2012 Mar 23

Assessment of Myelination in Hypomyelinating Disorders by Quantitative MRI
Dreha-Kulaczewski S, Brockmann K, Henneke M, Dechent P, Gärtner J, Helms G (2012). J Magn Reson Imaging. 2012 Dec;36(6):1329-38. doi: 10.1002/jmri.23774. Epub 2012 Aug 21.

Pelizaeus-Merzbacher-like disease" presenting as complicated hereditary spastic paraplegia.
Zittel, S., M. Nickel, N. I. Wolf, G. Uyanik, D. Glaser, C. Ganos, C. Gerloff, A. Munchau and A. Kohlschutter J Neurol. 2012 Nov;259(11):2498-500. doi: 10.1007/s00415-012-6617-0. Epub 2012 Jul 26

L-2 hydroxyglutaric aciduria as a rare cause of leukencephalopathy in adults
Weimar C, Schlamann M, Krägeloh-Mann I, Schöls L (2012) Clin Neurol Neurosurg. [Epub ahead of print]

Therapy of Pelizaeus-Merzbacher disease in mice by feeding a cholesterol-enriched diet
Saher G, Rudolphi F, Corthals K, Ruhwedel T, Schmidt KF, Löwel S, Dibaj P, Barrette B, Möbius W, Nave KA (2012) Nat Med, July 2012, doi: 10.1038/nm.2833

Molecular characterization of FOLR1 mutations delineates cerebral folate transport deficiency
Grapp M, Just IA, Linnankivi T, Wolf P, Lücke T, Häusler M, Gärtner J, Steinfeld R (2012) Brain 135:2022-2031.

Genetic disruption of Pten in a novel mouse model of tomaculous neuropathy
Goebbels S, Oltrogge JH, Wolfer S, Wieser GL, Nientiedt T, Pieper A, Ruhwedel T, Groszer M, Sereda MW, Nave KA (2012) EMBO Molecular Medicine 4:486

Glycolytic oligodendrocytes maintain myelin and long-term axonal integrity
Fünfschilling U, Supplie LM, Mahad D, Boretius S, Saab AS, Edgar J, Brinkmann BG, Kassmann CM, Tzvetanova ID, Möbius W, Diaz F, Meijer D, Suter U, Hamprecht B, Sereda MW, Moraes CT, Frahm J, Goebbels S, Nave KA (2012).Nature. 485:517

Imaging evidence of early brain tissue degeneration in patients with vanishing white matter disease: A multimodal MR study
Ding, X. Q., A. Bley, A. Ohlenbusch, A. Kohlschutter, J. Fiehler, W. Zhu and H. Lanfermann (2012). J Magn Reson Imaging 35(4): 926-32.

Long-term diffusion impairment of cerebral white matter in a degenerative disease of the central and peripheral nervous system: reflection of chronic excitotoxicity?
Blum, K. S., C. Hagel, E. Neuen-Jacob, P. Herkenrath, J. Fiehler, A. Kohlschutter, H. Lanfermann and X. Q. Ding J Child Neurol 27(2): 229-33.

Mutations in SLC33A1 cause a lethal autosomal recessive disorder with congenital cataracts and hearing loss associated with low serum copper and ceruloplasmin
Huppke P, Brendel C, Kalscheuer V, Korenke GC, Marquardt I, Freisinger P, Christodoulou J, Hillebrand M, Pitelet G, Wilson C, Gruber-Sedlmayr U, Ullmann R, Haas S, Elpeleg O, Nürnberg G, Nürnberg P, Dad S, Birk Møller L, Kaler SG, Gärtner J (2012) Am J Hum Genet 90:61-68.

Newborn Screening for Lysosomal Storage Disorders in Hungary
Rolfs A, Wittmann J, Karg E, Sandor T, Legnini E, Wittmann G, Giese AK, Lukas J, Gölnitz U, Klingenhäger M, Bodamer O, Mühl A (2012) JIMD Reports Volume 6, pp 117-125.

Determining the Demyelination Load in Metachromatic Leukodystrophy
Clas P, Wilke M, Gröschel S (2012) Acad Radiol 19:26–34

Quantitative and integrative proteome analysis of peripheral nerve myelin identifies novel myelin proteins and candidate neuropathy loci
Patzig J, Jahn O, Tenzer S, Wichert SP, de Monasterio-Schrader P, Rosfa S, Kuharev J, Yan K, Bormuth I, Bremer J, Aguzzi A, Orfaniotou F, Hesse D, Schwab MH, Möbius W, Nave KA, Werner HB (2011) Journal of Neuroscience 31:16369

Acetazolamide-responsive exercise-induced episodic ataxia associated with a novel homozygous DARS2 mutation
Synofzik M, Schicks J, Lindig T, Biskup S, Schmidt T, Hansel J, Lehmann-Horn F, Schols L (2011) Journal of Medical Genetics. 48(10):713-715

Metachromatic leukodystrophy: natural course of cerebral MRI changes in relation to clinical course
Groeschel S, Kehrer C, Engel C, Dali C, Bley A, Steinfeld R, Grodd W, Krägeloh-Mann I (2011) J Inherit Metab Dis 34(5):1095-102.

Misalignment of PLP/DM20 transmembrane domains determines protein misfolding in Pelizaeus-Merzbacher disease
Dhaunchak AS, Colman DR, Nave KA (2011). J Neurosci 31(42): 14961-71.

Telencephalic histopathology and changes in behavioural and neural plasticity in a murine model for metachromatic leukodystrophy
Faldini E, Stroobants S, Lüllmann-Rauch R, Eckhardt M, Gieselmann V, Balschun D, D'Hooge R (2011) Behav Brain Res 222:309-314.

The natural course of gross motor deterioration in metachromatic leukodystrophy
Kehrer C, Blumenstock G, Gieselmann V, Krägeloh-Mann I (2011) Dev Med Child Neurol. 53(9):850-5.

Rare differential diagnosis of primary adrenal insufficiency: case 6/2011
Blaschka F, Synofzik M, Schols L, Rau I, Gal A, Mussig K (2011). Deutsche Medizinische Wochenschrift (1946). 136(24):1316

Development and reliability of a classification system for gross motor function in children with metachromatic leucodystrophy
Kehrer C, Blumenstock G, Raabe C, Krägeloh-Mann I (2011) Dev Med Child Neurol 53(2):156-60.
Kohlschütter, A, Eichler, F (2011). Childhood leukodystrophies: a clinical perspective. Expert Rev Neurother 11(10): 1485-96.

rnaset2 mutant zebrafish model familial cystic leukoencephalopathy and reveal a role for RNase T2 in degrading ribosomal RNA
Haud N, Firat KF, Diekmann S, Henneke M, Willer JR, Hillwig MS, Gregg RG MacIntosh GC, Gärtner J, Alia A, Hurlstone AFL (2011) Proc Natl Acad Sci USA108:1099-1103.