Alexander Disease Research Bibliography
(updated November 14, 2018)

Recently added:

Sofroniew MV. (2018). Stem-Cell-Derived Astrocytes Divulge Secrets of Mutant GFAP. Cell Stem Cell. 23, 630-631. [editorial discussing the two new iPS cell papers, Li et al. and Jones et al.]


2018


prepared by Albee Messing


prepared by Albee Messing

2017


Boczek NJ, Sigafoos AN, Zimmermann MT, Maus RL, Cousin MA, Blackburn PR, Urrutia R, Clark KJ, Patterson MC, Wick MJ, Klee EW. (2016). Functional characterization of a GFAP variant of uncertain significance in an Alexander disease case within the setting of an individualized medicine clinic. Clinical Case Reports. 4, 885-895 [there is much misinformation here, but it is an interesting case report of an uncommon variant]


2016

prepared by Albee Messing


Elmali AD, Çetinçelik Ü, Islak C, Adatepe NU, Savrun FK, Yalçinkaya C. (2016). Familial adult-onset Alexander disease: clinical and neuroradiological findings of three cases. *Noropsikiyatri Arsivi-Archives of Neuropsychiatry*. 53, 169-172 [note that the mutation is reported incorrectly, and should be M415I]


partialis continua. Journal of Child Neurology 31, 869-72


2015


Ahmad O, Rowe DB. (2015). Adult-onset Alexander's disease mimicking degenerative disease. Practical Neurology 15, 393-395 [one of the patients with onset at 79 years]


Ferreira MC, Dorboz I, Rodriguez D, Boesplug-Tanguy O. (2015). **Screening for GFAP rearrangements in a cohort of Alexander disease and undetermined leukoencephalopathy patients.** *European Journal of Medical Genetics* 58, 466-70 [no deletions or duplications found]


**2014**


prepared by Albee Messing


2013


Hagemann TL, Paylor R, Messing A. (2013). Deficits in adult neurogenesis, contextual fear conditioning and spatial learning in a Gfap mutant mouse model of Alexander disease. Journal of Neuroscience. 33, 18698-18706 [describes an entirely new phenotype not previously known to be part of the disease]


Snider NT, Park H, Omary MB. (2013). A conserved rod domain phosphotyrosine that is targeted by the phosphatase PTP1B promotes keratin 8 insolubility and filament organization. Journal of Biological Chemistry. 288, 31329-37 (includes comparison of mutant keratin and GFAP)


Melchionda L, Fang M, Wang H, Fugnanesi V, Morbin M, Liu X, Li W, Ceccherini I, Farina L, Savoiardo M, P DA, Zhang J, Costa A, Ravaglia S, Ghezzi D, Zeviani M. (2013). Adult-onset Alexander disease, associated with a mutation in an alternative GFAP transcript, may be phenotypically modulated by a non-neutral HDAC6 variant. Orphanet Journal of Rare Diseases. 8, 66 [raises the possibility of a pathogenic mutation in the GFAP-delta form of GFAP (a minor variant) and also germline mosaicism in the mother – however, the evidence for both of these claims is weak]


Jany, P.L., Hagemann, T.L., and Messing, A. GFAP expression as an indicator of disease severity in mouse models of Alexander disease. ASN Neuro 5:art:e00109.doi:10.1042/AN20130003 [link to full article and podcast]


**2012**


prepared by Albee Messing


Hagemann TL, Jobe EM, Messing A. (2012) Genetic ablation of Nrf2/antioxidant response pathway in Alexander disease mice reduces hippocampal gliosis but does not impact survival. PLoS ONE 7, e37304 [link to full article]


prepared by Albee Messing
neurological symptoms for over 12 years, despite insidiously progressive cervicomedullary atrophy. *Neurological Sciences* 33, 1389-1392


2011


Chen YS, Lim SC, Chen MH, Quinlan RA, Perng MD. (2011). *Alexander disease causing mutations in the C-terminal domain of GFAP are deleterious both to assembly and network formation with the potential to both activate caspase 3 and decrease cell viability*. *Experimental Cell Research* 317, 2252-66


prepared by Albee Messing


2010


prepared by Albee Messing
**2009**


prepared by Albee Messing


Liem RKH and Messing A. (2009). Dysfunctions of neuronal and glial intermediate filaments in disease. *Journal of Clinical Investigation* 119, 1814-1824 [contains review of GFAP in blood or CSF as a potential biomarker for various diseases] [link to full article]

2008


2007


GFAP mutations and polymorphisms in 13 unrelated Italian patients affected by Alexander disease. *Clinical Genetics* 72, 427-433


2006


prepared by Albee Messing


**2005**


20

prepared by Albee Messing


2004


prepared by Albee Messing

**2003**


[same patients for whom clinical/genetic data reported in Meins et al., 2002]


**2002**


[Genetic studies reported as patient #4 in Shiroma et al., 2003]


[see Sawaishi et al., 1999, for more clinical detail on this patient]


2001


prepared by Albee Messing


2000


1999


prepared by Albee Messing


1998


1997


1996


prepared by Albee Messing


1995


1994


1993


prepared by Albee Messing


1992


1991


preparation by Albee Messing


1990


1989


1988


1987

Sorjonen DC, Cox NR, Kwapien RP. (1987). Myeloencephalopathy with eosinophilic refractile bodies (Rosenthal fibers) in a Scottish terrier. Journal of the American Veterinary Medical Association 190, 1004-1006


prepared by Albee Messing


1982


1981


1980


prepared by Albee Messing
1979


1977


1976


1974


1973

1972


1970


1968


1967


1966


1964


prepared by Albee Messing
1962


1959


1953


1952

Stevenson LD, Vogel FS. (1952). A case of macrocephaly associated with feeble-mindedness and encephalopathy with peculiar deposits throughout the brain and spinal cord. *Ciencia* (Méx.) 12, 71-74

1949

[First description of a child with Alexander disease]

1898