Alexander Disease Research Bibliography
(updated January 13, 2019)

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


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]


prepared by Albee Messing


2016


Alfke H, Schimrigk S. (2016). *Tumor-mimicking brainstem lesion in an adult with Alexander disease.* Rofo-Fortschritte Auf Dem Gebiet Der Rontgenstrahlen Und Der Bildgebenden Verfahren. 188, 869-870 [no genetic diagnosis given, although it says there was one]


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]


prepared by Albee Messing


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]


cognitive decline and behavioural disturbance in late-onset Alexander disease. J Neurol Sci. 357, 319-321


2014


2013

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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:e00109.doi:10.1042/AN20130003 [link to full article and podcast]


2012

prepared by Albee Messing


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


2010

prepared by Albee Messing


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


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2007


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2006


[Provides additional clinical information on E207K patient initially reported in Van der Knaap, et al, 2005]


[age of onset for this patient would be considered "infantile" according to our classification]


prepared by Albee Messing


**2005**


prepared by Albee Messing
reveals a stress response followed by glial activation and neuronal dysfunction. *Human Molecular Genetics* 14, 2443-2458


**2004**


prepared by Albee Messing


**2003**


prepared by Albee Messing
Japanese patients with Alexander disease: a novel mutation, R79L. Brain & Development 25, 116-121

[Appears to be same as patient #10 in Gorospe, et al., 2002 - there is considerable misinformation in the literature review]


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


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2000


1999


prepared by Albee Messing


**1998**


**1997**


prepared by Albee Messing
**1996**


**1995**


**1994**


**1993**


1992


1991

prepared by Albee Messing


1990


prepared by Albee Messing


**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


1986


1985


1984


1983


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1982


1981


1980


prepared by Albee Messing


1979


1977


1976


1974


prepared by Albee Messing

1973


1972


1970


1968


1967


1966


prepared by Albee Messing
1964

[Sixth case, first use of the name "Alexander's disease."]


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