Alexander Disease Research Bibliography (August 22, 2017)

Recently added:


2017


2017


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-Archiives of Neuropsychiatry. 53, 169-172 [note that the mutation is reported incorrectly, and should be M415I]


2015


**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]


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2012


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]


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


**2009**


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


2006


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


**2005**


**2004**


2003


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


[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]


2001


2000


1999


1998


1997


1996


1995


1994

1993


1992

[see Messing et al., 2011, for genetics]


1991


[Included two Alexander disease patients]


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


1986


1985


1984


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1983


1982


1981


1980


1979


1977

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1976


1974


1973


1972


1970


1968


**1967**


**1966**


**1964**


**1962**


**1959**


**1953**


**1952**

preparing by Albee Messing
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**