Alexander Disease Research Bibliography (updated September 16, 2018)

Recently added:


2018


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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


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]


2015


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**2014**


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**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)


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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]


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**2012**


LaPash Daniels CM, Austin EV, Rockney DE, Jacka EM, Hagemann TL, Johnson DA, Johnson JA, Messing A. (2012) **Beneficial effects of Nrf2 overexpression in a mouse model of Alexander disease.** *Journal of Neuroscience* 32, 10507-10515  [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**


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**2009**


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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


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**2006**


[TProvides 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]


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2005


2004


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**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]


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**2001**


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2000


1999

prepared by Albee Messing


Herndon RM. (1999). **Is Alexander's disease a nosologic entity or a common pathologic pattern of diverse etiology?** *Journal of Child Neurology* 14, 275-276


**1998**


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**1991**


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1990


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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


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**1976**


**1974**


**1973**


**1972**


**1970**


**1968**


1967


1966


1964


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**

Alexander WS. (1949). *Progressive fibrinoid degeneration of fibrillary astrocytes associated with mental retardation in a hydrocephalic infant*. *Brain* 72, 373-381
[First description of a child with Alexander disease]

**1898**