Alexander Disease Research Bibliography (updated March 13, 2016)

Recently added:


2015


prepared by Albee Messing
Knaap MS, Messing A. CSF and blood levels of GFAP in Alexander disease. eNeuro DOI: 10.1523/ENEURO.0080-15.2015 [full text]


2014


2013

prepared by Albee Messing

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**


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

Messing A, Daniels CM, Hagemann TL. (2010). Strategies for treatment in Alexander disease. Neurotherapeutics 7, 507-515 [review] [link to full article]


prepared by Albee Messing
In vitro treatments with ceftriaxone promote elimination of mutant glial fibrillary acidic protein and transcription down-regulation. *Experimental Cell Research* 316, 2152-65

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


[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
pathway involving filament aggregation and the association of alphaB-crystallin and HSP27. *American Journal of Human Genetics* 79, 197-213  [link to full article]


2005


2004


2003


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[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**


2000


prepared by Albee Messing
1999


1998


prepared by Albee Messing

1997


1996


1995


1994


1993


1992


1991


**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


1966


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

1949

[First description of a child with Alexander disease]
1898