Alexander Disease Research Bibliography (updated February 6, 2016)

Recently added:


2015


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2014


**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]


2012


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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


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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


(2007). **GFAP mutations and polymorphisms in 13 unrelated Italian patients affected by Alexander disease.** *Clinical Genetics* 72, 427-433


2006


pathway involving filament aggregation and the association of alphaB-crystallin and HSP27.  
A
mERICAN JOURNAL OF HUMAN GENETICS  79, 197-213 [link to full article]

van der Knaap MS, Ramesh V, Schiffmann R, Blaser S, Kyllerman M, Gholkar A, Elliso
lEXANDER DISEASE: VENTRICULAR GARLANDS AND ABNORMALITIES OF THE MEDULLA AND SPINAL CORD. Neurology  66, 494-8


2005


2004


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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]


2001


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1999


1998


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1997


1996


1995


1994


1993


1992


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**1991**


[Included two Alexander disease patients]


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1990


Wardinsky TD, Weinberger E, Pagon RA, Clarren SK, Thuline HC. (1990). *Partial deletion of the long arm of chromosome 11 [del(11)(q23.3----qter)] with abnormal white matter* [see comments]. American Journal of Medical Genetics 35, 60-63

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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1977

1976


1974


1973


1972


1970


1968


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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


31

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