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


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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]


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**2016**


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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

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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Archetypal and new families with Alexander disease and novel mutations in GFAP. Archives of Neurology 69, 208-214 [includes genetics on patients and families originally described by Seil 1968, Duckett 1992, and Schwankhaus 1995] [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

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


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

**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


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**2005**


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


**2004**


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**2003**


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]


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2001


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[Included two Alexander disease patients]


1990


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1989


1988


1987

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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


31

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1982


1981


1980


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1979


1977


1976


1974


1973


1972


1970


1968


1967


1966


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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


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