Alexander Disease Research Bibliography (updated January 24, 2018)

Recently added:


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]


prepared by Albee Messing


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, Yağlıncaya 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


prepared by Albee Messing


Ferreira MC, Dorboz I, Rodriguez D, Boespflug-Tanguy O. Screening for GFAP rearrangements in a cohort of Alexander disease and undetermined leukoencephalopathy patients. European Journal of Medical Genetics 58, 466-70 [no deletions or duplications found]


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]


Wada Y, Yanagihara C, Nishimura Y, Namekawa M. (2013). **Familial adult-onset Alexander disease with a novel mutation (D78N) in the glial fibrillary acidic protein gene with unusual bilateral basal ganglia involvement.** *Journal of the Neurological Sciences.* 331, 161-164


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]


prepared by Albee Messing


2012


prepared by Albee Messing


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


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


2005


2004


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2003


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**2002**

[Genetic studies reported as patient #4 in Shiroma et al., 2003]


[see Sawaishi et al., 1999, for more clinical detail on this patient]


[Appears to be same as patient #10 in Gorospe, et al., 2002 - there is considerable misinformation in the literature review]


**2001**


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**2000**


**1999**


1998


1997


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1992


1991


American Journal of Pathology 139, 933-938

[Included two Alexander disease patients]


1990


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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


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1964


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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