Alexander Disease Research Bibliography (updated August 7, 2018)

Recently added:


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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]


2016

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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, 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]


partialis continua. *Journal of Child Neurology* 31, 869-72


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]


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**Lithium decreases glial fibrillary acidic protein in a mouse model of Alexander disease.** PLoS One 10, e0138132


Ferreira MC, Dorboz I, Rodriguez D, Boespflug-Tanguy O. (2015). **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**


**2013**


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:art:e00109.doi:10.1042/AN20130003 *[link to full article and podcast]*

prepared by Albee Messing


2012


prepared by Albee Messing
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


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**


prepared by Albee Messing


2007


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


2006


[Provides additional clinical information on E207K patient initially reported in Van der Knaap, et al, 2005]


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2005


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2004


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**2003**


[same patients for whom clinical/genetic data reported in Meins et al., 2002]


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[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

Brenner M, Johnson AB, Boespflug-Tanguy O, Rodriguez D, Goldman JE, Messing A. (2001). **Mutations in GFAP, encoding glial fibrillary acidic protein, are associated with Alexander disease.** *Nature Genetics* 27, 117-120 [first description of genetics] [link to full article]


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[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

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1962


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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