Alexander Disease Research Bibliography (December 3, 2017)

Recently added:


2017


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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, Yağcıkaya 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


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]


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]


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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]


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2012


cord paralysis during sleep. *Journal of Neurology* 259, 2234-2236 [imaging findings reported by Ito et al. 2009]


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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features and a GFAP allele carrying both the p.Arg79His mutation and the p.Glu223Gln coding variant. *Journal of Neurology* 256, 679-682


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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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**


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prepared by Albee Messing
Journal of Cell Science  118, 2057-2065  [link to full article]


2004


2003


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


2002


**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]


2000


1999


prepared by Albee Messing
*Positron emission tomography in juvenile Alexander disease.* *Journal of the Neurological Sciences* 165, 116-120

[likely mis-diagnosis]


1998


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1987

Sorjonen DC, Cox NR, Kwapien RP. (1987). Myeloencephalopathy with eosinophlic refractile bodies (Rosenthal fibers) in a Scottish terrier. Journal of the American Veterinary Medical Association 190, 1004-1006


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1986

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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