Alexander Disease Research Bibliography (December 12, 2017)

Recently added:


2017


2016


Elmali AD, Çetinçelik Ü, Islak C, Adatepe NU, Savrun FK, Yaçinkaya C. (2016). Familial adult-onset Alexander disease: clinical and neuroradiological findings of three cases. *Noropsikiyatri Arşivi-Archives of Neuropsychiatry.* 53, 169-172 [note that the mutation is reported incorrectly, and should be M415I]


**Alexander disease in a Labrador Retriever.** *European Journal of Human Genetics* 24, 852-856

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]


prepared by Albee Messing


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]


prepared by Albee Messing

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]


2012


Hagemann TL, Jobe EM, Messing A. (2012) Genetic ablation of Nrf2/antioxidant response pathway in Alexander disease mice reduces hippocampal gliosis but does not impact survival. PLoS ONE 7, e37304  [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**


prepared by Albee Messing


2009


prepared by Albee Messing
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


Prepared by Albee Messing


**2007**


2006

prepared by Albee Messing


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**


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


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


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


**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**


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

*likely mis-diagnosis*


**1998**


**1997**


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prepared by Albee Messing
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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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