Alexander Disease Research Bibliography (December 17, 2017)

Recently added:


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


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]


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]


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**


Scola RH, Lorenzoni PJ, Kay CSK, Werneck LC. (2014). **Adult-onset Alexander disease: could facial myokymia be a symptom?** *Arquivos de Neuro-Psiquiatria* 72, 897-898


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**


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


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


2009


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**


[functional studies reported by Bachetti et al.]

[clinical features reported by Balbi et al.]


2007

[no mutation found]


2006


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


[age of onset for this patient would be considered "infantile" according to our classification]


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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**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**


2000


1999


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**1992**


**1991**


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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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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

1898