Alexander Disease Research Bibliography  
(updated January 13, 2019)

Recently added:


[editorial discussing the two new iPSC cell papers, Li et al. and Jones et al.]


[putative pathogenic variant in the minor isoform, GFAP-delta]


prepared by Albee Messing


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


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, 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]


prepared by Albee Messing


**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
*Journal of the Neurological Sciences* 357, 319-321


prepared by Albee Messing


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]


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)

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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


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]


Kessell, A.E., Finnie, J.W., Manavis, J., Cheetham, G.D., and Blumbergs, P.C. (2012). _A Rosenthal Fiber Encephalomyelopathy Resembling Alexander's Disease in 3 Sheep._ _Veterinary Pathology_ 49, 248-254  [no GFAP mutations were detected]


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


prepared by Albee Messing
[link to full article]

[no genetic confirmation of diagnosis, however]


prepared by Albee Messing
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


prepared by Albee Messing


2007


2006


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


prepared by Albee Messing


2005


No to Hattatsu [Brain & Development] 37, 55-59 [R239C patient]


2004


2003


prepared by Albee Messing
*Pediatric Radiology* 33, 47-49

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

*Lancet Neurology* 2, 75

*Annals of Neurology* 53, 118-120

**2002**

*Brain & Development* 24, 723-726

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

*Annals of Neurology* 52, 779-785

*Journal of Neurogenetics* 16, 175-179

*Neuropediatrics* 33, 194-198

*Neurology* 58, 1541-1543

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

*Journal of Child Neurology* 17, 227-230

prepared by Albee Messing


2001


2000


1999


1998


prepared by Albee Messing
  *Pediatric Neurology* 18, 67-70

1997

  *Neuroscience Letters* 231, 79-82

  *Neurology* 48, 552

  *Neurology* 48, 552

  *Bone Marrow Transplantation* 20: 247-249

1996


  *Pediatric Pathology & Laboratory Medicine* 16, 327-343

  *Acta Neuropathologica* 91, 200-204

  *Clinical Neuropathology* 15, 13-16

1995


1994


1993


*Developmental Medicine & Child Neurology* 35, 732-736

**1992**

*Pediatric Neurosurgery* 18, 134-138
[see Messing et al., 2011, for genetics]

*Neurology* 42, 1733-1735

*Journal of Child Neurology* 7, 168-171

Iwaki A, Iwaki T, Goldman JE, Ogomori K, Tateishi J, Sakaki Y. (1992). Accumulation of alpha B-crystallin in brains of patients with Alexander's disease is not due to an abnormality of the 5'-flanking and coding sequence of the genomic DNA. 
*Neuroscience Letters* 140, 89-92

*Patologia Polska* 43, 193-195

*Acta Neuropathologica* 84, 322-327

**1991**

*Clinical Neuropathology* 10, 122-126

*Neuroradiology* 33, 438-440

prepared by Albee Messing


[Included two Alexander disease patients]


1990


prepared by Albee Messing


1989


1988


prepared by Albee Messing

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


1985


1984


**1983**


**1982**


**1981**


prepared by Albee Messing
1980

*Acta Neurologica* 2, 1-9

*Italian Journal of Neurological Sciences* 1, 131-138

*Neuroradiology* 20, 103-106

*Rivista di Neurobiologia* 26, 357-364

*Acta Neuropathologica* 50, 237-240

1979

*Journal of Neurology, Neurosurgery & Psychiatry* 42, 619-624

*Acta Neuropathologica* 45, 133-140

*Acta Neuropathologica* 47, 81-84

1977

*Archives of Pathology & Laboratory Medicine* 101, 655-657
1976


1974


1973


1972


1970


prepared by Albee Messing
1968

*Neurology* 18, 543-549

*Archives of Neurology* 19, 494-502 [see Messing et al., 2012, for genetics]

1967

*Shinkei Kenkyu No Shimpo* 11, 765-774

1966

*Shinkei Kenkyu No Shimpo - Advances in Neurological Sciences* 10, 716-720

1964

*Archives of Neurology* 11, 414-422  
[Sixth case, first use of the name "Alexander's disease."]

*Acta Neuropathologica* 4, 212-217

1962

*Acta Neuropathologica* 2, 126-143

1959

*Journal of Neuropathology and Experimental Neurology* 18, 359-383

prepared by Albee Messing
1953

Crome L. (1953). **Megalencephaly associated with hyaline pan-neuropathy.**  
*Brain* 76, 215-228

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 (México)* 12, 71-74

1949

Alexander WS. (1949). **Progressive fibrinoid degeneration of fibrillary astrocytes associated with mental retardation in a hydrocephalic infant.**  
*Brain* 72, 373-381  
[First description of a child with Alexander disease]

1898

*Bietr.Pathol.Anat.* 23, 111-143 [first description of what later came to be known as “Rosenthal fibers”]