Alexander Disease Research Bibliography  
(updated October 16, 2019)

Recently added:


2019


prepared by Albee Messing


2018


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Atypical Alexander disease with dystonia, retinopathy, and a brain mass mimicking astrocytoma.

*Neurology: Genetics* 4, e248  [full text]


*Brain & Development* 40, 587-591


*Neurology* 91, e396-e397


*Journal of Voice* (in press) (variant not specified, nor confirmed as pathogenic)


*Annals of the Academy of Medicine, Singapore* 47, 191-193  [full text]


*Nature Communications* 9, 1899  [full text]


*Brain Pathology* 28, 388-398  [review]


*Brain & Development* 40, 330-333


Antisense suppression of glial fibrillary acidic protein as a treatment for Alexander disease.

*Annals of Neurology* 83, 27-39  [first real prospect for treatment]  [full text]


*Pediatric Neonatology* 59, 624-627

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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]
*Neurology India* 65, 887-889

*Human Genome Variation* 4, 17028 [full text]

*Zhonghua Er Ke Za Zhi* 55, 504-508

*Frontiers in Neurology* 8:255

*Journal of Child Neurology* 32:184-187

*European Neurology* 77:296-302

*BMJ Case Reports* doi: 10.1136/bcr-2016-218484 [genetic results not provided in text, but personal communication from author as N386S]


*Acta Neuropathologica Communications* 5, 27

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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, Yalçinkaya C. (2016). Familial adult-onset Alexander disease: clinical and neuroradiological findings of three cases. *Noropsikiyatri Arsivi-Arsvi- Archives of Neuropsychiatry* 53, 169-172 [note that the mutation is reported incorrectly, and should be M415I]


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**2015**


onset leukodystrophy - a possible Alexander's disease.

*European Journal of Neurology* 22, 552-552

[meeting abstract - onset at 71 years, novel mutation]


*Nature Communications* 6, 6:8966 | DOI: 10.1038/ncomms9966 [full text]


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


*eNeuro* DOI: 10.1523/ENEURO.0080-15.2015 [full text]


*PLoS One* 10, e0138132


*Acta Neuropathologica* 130, 469-486


*European Journal of Medical Genetics* 58, 466-70 [no deletions or duplications found]


*Clinical Neuropathology* 34, 298-302


*Clinical Neuropathology* 34, 207-214

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]


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


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


**2009**


[mutation and other clinical findings reported by Hida et al. 2012]


prepared by Albee Messing


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


**2006**


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


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2005


2004


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2003


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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


prepared by Albee Messing
*Acta Neurologica Scandinavica* 99, 158-165

*Journal of Child Neurology* 14, 325-329  
[see Li et al. 2005 for genetics]

Herndon RM. (1999). *Is Alexander's disease a nosologic entity or a common pathologic pattern of diverse etiology?*  
*Journal of Child Neurology* 14, 275-276

*Movement Disorders* 14, 689-693

*Archives of Ophthalmology* 117, 265-267

*Journal of the Neurological Sciences* 165, 116-120

*Nature Genetics* 21, 260-261  
[likely mis-diagnosis]

*Neurology India* 47, 333-335.

**1998**

*Brain Research* 787, 15-18

*American Journal of Pathology* 152, 391-398

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


prepared by Albee Messing
1992


[see Messing et al., 2011, for genetics]


1991


**1990**


prepared by Albee Messing
Wardinsky TD, Weinberger E, Pagon RA, Clareen SK, Thuline HC. (1990). Partial deletion of the long arm of chromosome 11 \([\text{del}(11)(q23.3----qter)]\) with abnormal white matter [see comments].
*American Journal of Medical Genetics* 35, 60-63

1989

*Cell* 57, 71-78

*American Journal of Anatomy* 185, 335-341

1988


*American Journal of Pathology* 130, 569-578

*Pediatric Radiology* 18, 227-228

*Annals of Neurology* 24, 302 [really no evidence]

*Journal of Pathology* 155, 9-15

*Neurology* 38, 152-154

1987

prepared by Albee Messing
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

*Rinsho Shinkeigaku - Clinical Neurology* 27, 1141-1144

1986

*Acta Neuropathologica* 71, 163-166

1985

*Brain* 108, 367-385

*Acta Neuropathologica* 67, 163-166

1984

*Annals of Neurology* 15, 605-607

*Revue Neurologique* 140, 179-189

*No to Hattatsu [Brain & Development]* 16, 76-78

*Journal of Neurology, Neurosurgery, and Psychiatry* 47, 399-403

1983

*Morphologiai Es Igazsagugyi Orvosi Szemle* 23, 169-175

prepared by Albee Messing


1982


1981


1980


1979


1977


1976


1974


Archives of Pathology & Laboratory Medicine 98, 379-385

1973


1972


1970


1968


1967


prepared by Albee Messing
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

1953

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

preparing by Albee Messing

[First description of a child with Alexander disease]

1898


[first description of what later came to be known as “Rosenthal fibers”]