Alexander Disease Research Bibliography  
(updated May 15, 2019)

**Recently added:**

Neuropsychological features of adult form of Alexander disease.  
*Journal of the Neurological Sciences* 401, 87-89

Cabrera-Galván JJ, Martínez-Martin MS, Déniz-García D, Araujo-Ruano E, Travieso-Aja MDM.  
(2019).  
Adult-onset Alexander disease with a heterozygous D128N GFAP mutation: a pathological study.  
*Histology and Histopathology* (in press)

[concise review of clinical genetics]

*Case Rep Med.* 2019, 2986538

*Neurocase.* DOI: 10.1080/13554794.13552019.11580749

*Indian Journal of Pediatrics* (in press)

2018

*Neuropediatrics* 49, 256-261

*Cell Stem Cell* 23, 630-631.  
[editorial discussing the two new iPS cell papers, Li et al. and Jones et al.]

prepared by Albee Messing


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


2017


prepared by Albee Messing
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]


*Clinical Neurology and Neurosurgery* 157, 31-33


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


*Clinical Neuropathology* 34, 207-214

*Glia* 63, 2285-97

*Journal of the Neurological Sciences* 354, 131-132

*European Journal of Human Genetics* 23:72-78 [the first nonsense mutation reported, predicting expression of a severely shortened protein]


2014

*Journal of Clinical & Diagnostic Research* 8, Pd03-04 [full text]

*Neurology* 83, e197-198

*Arquivos de Neuro-Psiiquiatria* 72, 897-898

*Brain Research* 1582, 211-219

prepared by Albee Messing
*Journal of Neuroscience* 34, 6448-6558 (* joint first authors)*


*Neurology* 82, 49-56

*APMIS* 122, 76-80

*Parkinsonism & Related Disorders* 20, 241-2

2013

*Iranian Journal of Pediatrics* 23, 481-484 [link to full article]

*Journal of Neuroscience* 33, 18698-18706  
[describes an entirely new phenotype not previously known to be part of the disease]

*Journal of Human Genetics* 58, 635-638

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]


prepared by Albee Messing


**2012**


LaPash Daniels CM, Austin EV, Rockney DE, Jacka EM, Hagemann TL, Johnson DA, Johnson JA, Messing A. (2012) **Beneficial effects of Nrf2 overexpression in a mouse model of Alexander disease.** *Journal of Neuroscience* 32, 10507-10515  [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]

Messing A, Brenner M, Feany MB, Nedergaard M, Goldman JE. (2012). **Alexander disease.** *Journal of Neuroscience* 32, 5017-5023  [review]  [link to full article]

Nam TS, Choi KH, Lee SH, Park MS, Kim JT, Choi SM, Kim BC, Kim MK, Cho KH. (2012). **Adult-onset Alexander disease mimicking neuromyelitis optica.** *Multiple Sclerosis Journal* 18, 546-546  [but may be mis-diagnosis, since D295N is a known polymorphism, and not proven as disease-causing]


2011


[proposes new classification system, with updated survival statistics]

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

*Journal of Neurology* 258, 1998-2008 [first population-based estimate of prevalence]

*Journal of Child Neurology* 26, 356-60

*Journal of Neuroscience* 31, 2868-2877 [first Drosophila model of the disease] [link to full article]

*Journal of Neuroophthalmology* 31, 155-9 [no genetic confirmation of diagnosis, however]

*Veterinary Pathology* 49, 248-254

*Journal of Neurology* 258, 938-40

*Journal of Neurology* 258, 935-7

*Brain & Development* 33, 604-7

*Acta Neurologica Scandinavia* 124, 104-108

*The Clinical Neuropsychologist* 25, 1266-1277
2010


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*Glia* 58, 1228-1234

*Experimental Cell Research* 316, 2152-65

2009

*Rinsho Shinkeigaku* 49, 358-363

*Clinical Nuclear Medicine* 34, 931-933 
[mutation and other clinical findings reported by Hida et al. 2012]

*Pediatric Blood & Cancer* 34, 931-933

*Neuroradiology* 10, 669-675

*Human Molecular Genetics* 18, 1190-1199 [link to full article]

*Experimental Cell Research* 315, 1260-1272


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


prepared by Albee Messing


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]


*Neurology* 66, 494-8


*Neurology* 66, 468-9 [editorial]


*Brain & Development* 28, 131-3 [not really the first, see Brenner et al, 2001 patient # 2]


*Human Genetics* 119, 137-44


*American Journal of Pathology* 168, 888-97


*Brain & Development* 28, 60-2

2005


*Annals of Neurology* 58, 813-4


*Neuropediatrics* 36, 319-23

preparing by Albee Messing

*Journal of Cell Science* 118, 2057-2065 [link to full article]


2004


**2003**


[same patients for whom clinical/genetic data reported in Meins et al., 2002]


2002


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**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


*Neurology* 58, 1494 - 1500


*International Journal of Developmental Neuroscience* 20, 259-268


*International Journal of Developmental Neuroscience* 20, 391-394


*Journal of the Neurological Sciences* 195, 71-76

**2001**


*Nature Genetics* 27, 117-120 [first description of genetics] [link to full article]


*Nature Genetics* 27, 10-11


*Neuroscience Letters* 312, 71-74
*Journal of Neuropathology and Experimental Neurology* 60, 563-573 [review]

*American Journal of Human Genetics* 69, 1134-1140. [see also 69, 1413]

*Journal of Human Genetics* 46, 579-582

*Journal of Neurochemistry* 76, 730-736

*Journal of Neuropathology and Experimental Neurology* 60, 553

*Journal of Neuropathology and Experimental Neurology* 60 (abstract)

*American Journal of Neuroradiology* 22, 541-552 [link to full article]

2000

*Epilepsia* 41, 628-630

Head MW, Goldman JE. (2000). Small heat shock proteins, the cytoskeleton, and inclusion body formation. 
*Neuropathology & Applied Neurobiology* 26, 304-312

prepared by Albee Messing
*Neuropediatrics* 31, 86-92

1999


prepared by Albee Messing
1998


1997


1996


1995


1994


1993


American Journal of Pathology 143, 487-495

*Journal of Child Neurology* 8, 134-144

*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

prepared by Albee Messing


[Included two Alexander disease patients]


1990


**Journal of Child Neurology** 5, 253-258

*Journal of Child Neurology* 5, 259-260

*Journal of Child Neurology* 5, 248-252

Wardinsky TD, Weinberger E, Pagon RA, Clarren SK, Thuline HC. (1990). Partial deletion of the long arm of chromosome 11 [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]

well as Rosenthal fibres in cerebellar astrocytomas, cytoplasmic bodies in muscle, and mallory bodies in alcoholic liver disease. 
Journal of Pathology  155, 9-15

Neurology  38, 152-154

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

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


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**1983**


**1982**


**1981**


**1980**

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**1979**


**1977**


**1976**


prepared by Albee Messing

**1974**


**1973**


**1972**


**1970**


**1968**

Neurology 18, 543-549


1967


1966


1964


[Sixth case, first use of the name "Alexander's disease."


1962


1959


1953


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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 (México)* 12, 71-74

1949

*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”]