Alexander Disease Research Bibliography
(updated June 19, 2019)

Recently added:


[concise review of clinical genetics]


2018


prepared by Albee Messing
Sofroniew MV. (2018). Stem-Cell-Derived Astrocytes Divulge Secrets of Mutant GFAP. *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
Astrocytes Impair Oligodendrocyte Progenitor Proliferation and Myelination in an hiPSC Model of Alexander Disease.

Cell Stem Cell  23, 239-251


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

Pediatric Neonatology  59, 624-627

Neurologia  33, 526-533  (review)

2017

Neurology: Clinical Practice  7, 425-429

Neurology: Clinical Practice  7, 523-526

Neuropediatrics  49, 118-122

Journal of Clinical Neurology  13, 426-428  [full text]

Neurologist 22, 247-248

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]


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, Yağcinkaya 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


prepared by Albee Messing
*Tidsskr Nor Laegeforen* 135, 1753-1755  [full text]

*Practical Neurology* 15, 393-395 [one of the patients with onset at 79 years]

*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]


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

prepared by Albee Messing
Brain Research 1582, 211-219

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]


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, Savoardo 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


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.


2010


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


prepared by Albee Messing


2009


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


prepared by Albee Messing
[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**


prepared by Albee Messing
*Journal of Neurology* 254, 1390-1394

*Acta Neuropathologica* 114, 543-545

*Experimental Cell Research* 313, 2766-2779

*Lancet Neurology* 6, 562-570

*Journal of Neurology* 254, 1278-1280

*Experimental Cell Research* 313, 2077-2087 [review] [full text]

*Glia* 55, 617-31

*Journal of Human Genetics* 52, 362-9

*Neurology* 68, 1322-3

**2006**


prepared by Albee Messing
**Neuropediatrics** 37:v1112 (abstract)

*Journal of Neuroscience* 26, 11162-11173  [link to full article]

*Journal of Child Neurology* 21, 1075-80  
[Provides additional clinical information on E207K patient initially reported in Van der Knaap, et al, 2005]

*Journal of Biological Chemistry* 281, 38634-38643  [link to full article]

*American Journal of Neuroradiology* 27, 2088-2092

*Brain & Development* 28, 663-667  
[age of onset for this patient would be considered "infantile" according to our classification]

*Journal of Korean Medical Science* 21, 954-957

*Neuroscience Letters* 407, 127-130

*Pediatric Neurology* 35, 293-296

prepared by Albee Messing


2005


prepared by Albee Messing


2004


prepared by Albee Messing


2003


prepared by Albee Messing
[German]
Monatsschrift fur Kinderheilkunde 151, 311-314

Brain & Development 25, 116-121

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

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


2001


prepared by Albee Messing
Neuroscience Letters 312, 71-74


2000


prepared by Albee Messing


**1999**


**1998**


**1997**


**1996**


1995


1994


1993


prepared by Albee Messing
*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

prepared by Albee Messing


(Included two Alexander disease patients)


1990


prepared by Albee Messing
Journal of Histochemistry and Cytochemistry 38, 103-109


1989


1988


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


1983


1982


1981


1980


1979


1977


prepared by Albee Messing
1976


1974


1973


1972


1970


1968


1967


1966


1964


1962


1959


prepared by Albee Messing
1953


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


[First description of a child with Alexander disease]

1898