Alexander Disease Research Bibliography  
(updated February 28, 2019)

Recently added:

[concise review of clinical genetics]


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


prepared by Albee Messing
*Acta Neuropathologica Communications* 6, 112
[putative pathogenic variant in the minor isoform, GFAP-delta]

*Neurologia* 33, 526-533 [review]

2018

*Brain & Development* 41, 195-200

*Nature Methods* 15, 693-+

*European Journal of Neurology* 25, e105-e106

*Cell Stem Cell* 23, 239-251

*European Journal of Medical Genetics* (in press)

*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


Boczek NJ, Sigafos 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


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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-Archives of Neuropsychiatry 53, 169-172 [note that the mutation is reported incorrectly, and should be M415I]


prepared by Albee Messing


2015


prepared by Albee Messing
*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]


preparred 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)*


Wada Y, Yanagihara C, Nishimura Y, Namekawa M. (2013). **Familial adult-onset Alexander disease with a novel mutation (D78N) in the glial fibrillary acidic protein gene with unusual bilateral basal ganglia involvement.** *Journal of the Neurological Sciences* 331, 161-164


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


prepared by Albee Messing


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


prepared by Albee Messing
2010


prepared by Albee Messing

*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

prepared by Albee Messing


prepared by Albee Messing


2007


*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

*Neuropediatrics* 37:v1112 (abstract)

[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]


*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

prepared by Albee Messing


2004


prepared by Albee Messing


**2003**


*prepared by Albee Messing*

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


prepared by Albee Messing
*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

prepared by Albee Messing


2000


prepared by Albee Messing

1999


prepared by Albee Messing
1998


1997


1996


prepared by Albee Messing

1995


1994


1993


31

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
*Clinical Neuropathology* 10, 122-126

*Neuroradiology* 33, 438-440

*American Journal of Pathology* 139, 933-938

*Radiology* 181, 173-181
[Included two Alexander disease patients]

*American Journal of Medical Genetics* 39, 226-227

*Biochemical and Biophysical Research Communications* 179, 1030-1035

*Veterinary Pathology* 28, 536-538

*Immunoelectron microscopy with colloidal gold. American Journal of Pathology* 138, 875-885

*FEBS Lett* 294:133-6

1990


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

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, Clareng 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]
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**

Brain Research 210, 419-425

Developmental Medicine & Child Neurology 23, 660-661

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


1976


1974


1973


1972


1970

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1968


1967


1966


1964


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


1962

1959


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


prepared by Albee Messing