Alexander Disease Research Bibliography
(updated December 17, 2019)

Recently added:


prepared by Albee Messing


**2019**


**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.]


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

*Brain & Development* 40, 330-333

*Annals of Neurology* 83, 27-39
[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]


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]

prepared by Albee Messing
*BMC Neurology* 16, 211

*Noropsikiyatri Arsivi-Archives of Neuropsychiatry* 53, 169-172

[note that the mutation is reported incorrectly, and should be M415I]

*Acta Neuropathologica Communications* 4, 69

*Journal of Proteome Research* 55, 2265-2282

*Journal of Neurology* 263, 821-822

*Journal of Neuroscience* 36, 1445-1455

*Neurological Sciences* 37, 973-977

*Journal of Child Neurology* 31, 869-72

*Neurological Sciences* 37, 143-145 [corresponds to S398F change in the protein sequence]

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


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)


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


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]


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


**2010**


prepared by Albee Messing


2009


its MRI features and a GFAP allele carrying both the p.Arg79His mutation and the 
p.Glu223Gln coding variant.
Journal of Neurology 256, 679-682

Sueda Y, Takahashi T, Ochi K, Ohtsuki T, Namekawa M, Kohriyama T, Takiyama Y, and 
Matsumoto M. (2009). Adult onset Alexander disease with a novel variant (S398F) in the 
glial fibrillary acidic protein gene [Article in Japanese].
Rinsho Shinkeigaku 49, 358-363

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
Disease: MR Imaging Prenatal Diagnosis.
American Journal of Neuroradiology 29, 1973-5

a series of eleven unrelated cases with review of the literature.
Brain 131, 2321-31 [same patients described by Farina 2008] [neuropsychology testing 
for some also described in Draghi 2019]

GFAP aggregates in the cochlear nerve increase the noise vulnerability of sensory cells in 
the organ of Corti in the murine model of Alexander disease.
Neuroscience Research 62, 15-24

Farina L, Pareyson D, Minati L, Ceccherini I, Chiapparini I, Romano S, Gambaro P, Fancellu R, 
American Journal of Neuroradiology 29, 1190-6
[radiology features of same patients described by Pareyson 2008]

disease causing hereditary late-onset ataxia with only minimal white matter changes: A 
report of two sibs.
Movement Disorders 23, 1613-5

prepared by Albee Messing


2007


**2006**


prepared by Albee Messing


prepared by Albee Messing

2005


2004

prepared by Albee Messing


2003


2002


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


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


2001


2000


1999


prepared by Albee Messing


1998


1997


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


Biotechnic & Histochemistry 70, 285-29

Neurology 45, 2266-2271 [see Messing et al., 2011, for genetics]

1994

Clinical Neuropathology 13, 31-38

1993

prepared by Albee Messing


1992


prepared by Albee Messing


**1991**


[Included two Alexander disease patients]


American Journal of Pathology 138, 875-885

FEBS Lett 294:133-6

1990

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, 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

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

prepared by Albee Messing


1984


1983


1982

prepared by Albee Messing

**1981**


**1980**


**1979**

Acta Neuropathologica  45, 133-140

Acta Neuropathologica  47, 81-84

1977

Archives of Pathology & Laboratory Medicine  101, 655-657

1976

Journal of Neurology, Neurosurgery & Psychiatry  39, 803-809

Neurology  26, 607-614

1974

Neurology India  22, 57-64

Archives of Pathology & Laboratory Medicine  98, 379-385

1973

Neuropatologia Polska  11, 127-141

1972

1970


1968


1967


1966


1964

*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

*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”]