Alexander Disease Research Bibliography
(updated July 11, 2019)

Recently added:


2019


2018

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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.]


2017


prepared by Albee Messing


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]
*BMC Neurology* 16, 211

Elmali AD, Çetinçelik Ü, Islak C, Adatepe NU, Savrun FK, Yağıcıkaya 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]

*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]


Ferreira MC, Dorboz I, Rodriguez D, Boespflug-Tanguy O. (2015). *Screening for GFAP rearrangements in a cohort of Alexander disease and undetermined leukoencephalopathy patients.* *European Journal of Medical Genetics* 58, 466-70 [no deletions or duplications found]


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2014

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2013

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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]

Journal of Neuroscience 33, 7439-7450

ASN Neuro 5:art:e00109.doi:10.1042/AN20130003  [link to full article and podcast]

Biancheri, R., Rossi, A., Ceccherini, I., Pezzella, M., Prato, G., Striano, P., and Minetti, C.  
Magnetic Resonance Imaging "Tigroid Pattern" in Alexander Disease.  
Neuropediatrics 44, 174-6

Brain & Development 35, 441-444

Journal of Pediatrics 162, 648

Journal of Human Genetics 58, 183-188

Journal of Proteome Research 12, 719-728

Glia 61, 210-224  [studies involved mouse model expressing R239H mutant]准备的Albee Messing


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
2011


Prust M et al. (2011). **GFAP mutations, age of onset, and clinical sub-types in Alexander disease**. *Neurology* 77, 1287-94 [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


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2010


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2009


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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


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


[T] [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]


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2005


2004

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2003


**2002**


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

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2001


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**2000**


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


**1999**


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*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

*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**


*Biotechnic & Histochemistry* 70, 285-29

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

**1994**

*Clinical Neuropathology* 13, 31-38

**1993**

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American Journal of Pathology 143, 1743-1753

Revue Neurologique 149, 781-787

Journal of Neurology, Neurosurgery, and Psychiatry 56, 977-981

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

*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


1990


1989


1988

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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

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*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

*Clinical Neuropathology* 2, 16-22

*Nippon Hifuka Gakkai Zasshi - Japanese Journal of Dermatology* 93, 1533-1535

*Acta Neuropathologica* 61, 36-42

**1982**

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1981


1980


1979


prepared by Albee Messing
*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

Missouri Medicine  69, 23-25

1970

Journal of Neuropathology and Experimental Neurology  29, 524-551

Archives of Pathology & Laboratory Medicine  89, 321-328

1968

Neurology  18, 543-549

Archives of Neurology  19, 494-502 [see Messing et al., 2012, for genetics]

1967

Shinkei Kenkyu No Shimpo 11, 765-774

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."

prepared by Albee Messing
*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”]

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