Alexander Disease Research Bibliography
(updated April 29, 2019)

Recently added:


prepared by Albee Messing
*Cell Reports* 25, 947-958

*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)
**Atypical Alexander disease with dystonia, retinopathy, and a brain mass mimicking astrocytoma.**  
*Neurology: Genetics* 4, e248  [*full text*]

*Brain & Development* 40, 587-591

Nam TS, Kang KW, Choi SY, Kim MK. (2018). **Teaching NeuroImages: Alexander disease with features of both frontal and bulbospinal involvement.**  
*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*]

prepared by Albee Messing


2017


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]


prepared by Albee Messing
*Acta Neuropathologica Communications* 5, 27

*Journal of Biological Chemistry* 292, 5814-5824

*Annual Review of Pathology* 12, 131-152

2016

*Movement Disorders: Clinical Practice* 3, 300-302

*Scientific Reports* 6, 35936

*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

*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]

European Journal of Human Genetics 24, 852-856

2015


prepared by Albee Messing
Neurology: Clinical Practice 5:259-262


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


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

prepared by Albee Messing
[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, 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
*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

*Alexander disease with mild dorsal brainstem atrophy and infantile spasms.*  
*Brain & Development* 35, 441-444

*Alexander Disease.*  
*Journal of Pediatrics* 162, 648

*Follow-up study of 22 Chinese children with Alexander disease and analysis of parental origin of de novo GFAP mutations.*  
*Journal of Human Genetics* 58, 183-188

*Protein changes in immunodepleted cerebrospinal fluid from transgenic mouse models of Alexander disease detected using mass spectrometry.*  
*Journal of Proteome Research* 12, 719-728

*Increased astrocytic ATP release results in enhanced excitability of the hippocampus.*  
*Glia* 61, 210-224  [studies involved mouse model expressing R239H mutant]

*Infantile-onset Alexander disease: a genetically proven case with mild clinical course in a 6-year-old Indian boy.*  
*Journal of Child Neurology* 28, 396-398

*Ceftriaxone for Alexander's Disease: A Four-Year Follow-Up.*  
*Journal of Inherited Metabolic Disorders Reports* 9, 67-71  [one patient, initial study reported in Sechi 2010]
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
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*. Experimental Cell Research 317, 2252-66


2010


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


2009


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


prepared by Albee Messing


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


2006


[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]


2005

prepared by Albee Messing


2004


Movement Disorders 19, 1244-1248

Journal of the Neurological Sciences 225, 125-127

Neurology 63, 1302-1304

Brain & Development 26, 206-208

Cellular & Molecular Life Sciences 61, 369-385

2003

Neuroscience Letters 350, 169-172

Neurology 61, 1014-1015

European Neurology 50, 100-105

Archives of Neurology 60, 1307-1312


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


[Appears to be same as patient #10 in Gorospe, et al., 2002 - there is considerable misinformation in the literature review]


**2002**


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


prepared by Albee Messing


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


2001


prepared by Albee Messing


2000

prepared by Albee Messing


**1999**


prepared by Albee Messing


1998


1997


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

American Journal of Pathology 143, 1743-1753

Revue Neurologique 149, 781-787


**1992**


preparation by Albee Messing

1991


[Included two Alexander disease patients]


prepared by Albee Messing
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


American Journal of Pathology 130, 569-578


prepared by Albee Messing
*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


1983


1982


1981

prepared by Albee Messing
*Neuropediatrics* 12, 382-391

*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

*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

Kepes JJ, Ziegler DK. (1972). *Alexander's disease in an adult (leukodystrophy with Rosenthal fibers).*


1968


1967


1966


1964


[Sixth case, first use of the name "Alexander's disease." ]
*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)* 1 2, 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”]