Alexander Disease Research Bibliography
(updated August 20, 2019)

Recently added:


2019


prepared by Albee Messing
2018


Nature Methods 15, 693-+


prepared by Albee Messing
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


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

*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]

prepared by Albee Messing

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]


2014

prepared by Albee Messing


2013

prepared by Albee Messing

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]


*Journal of Child Neurology* 28, 396-398

*Journal of Inherited Metabolic Disorders Reports* 9, 67-71
[one patient, initial study reported in Sechi 2010]

2012

*Experimental Cell Research* 318, 1844-54

*American Journal of Medical Genetics Part A* 158A, 2835-2842
[includes an estimate of prevalence/incidence in part of US]

*Journal of Neuroscience* 32, 10507-10515  [link to full article]

*Journal of Neurology* 259, 2234-2236  [imaging findings reported by Ito et al. 2009]


*Human Mutation* 11:1141-1148
[first example of an intronic mutation that affects splicing]


prepared by Albee Messing
2011


[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


[first Drosophila model of the disease] [link to full article]

[no genetic confirmation of diagnosis, however]


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]


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


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


2007


prepared by Albee Messing


2006


prepared by Albee Messing

2005


2004

prepared by Albee Messing


2003


*Archives of Neurology* 60, 1307-1312

*Journal of Neurology, Neurosurgery & Psychiatry* 74, 807-810

*Journal of Neurology* 250, 300-306

*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]


2001


**2000**


**1999**


prepared by Albee Messing


1998


1997


prepared by Albee Messing

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1995


1994


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prepared by Albee Messing


1992


[see Messing et al., 2011, for genetics]


1991


prepared by Albee Messing


1990


1989


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
*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**

*Morphologai 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**

**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


*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”]