Alexander Disease Research Bibliography  
(updated December 4, 2019)

Recently added:


2019


2018


prepared by Albee Messing


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Brain & Development 40, 330-333

Annals of Neurology 83, 27-39 
[first real prospect for treatment] [full text]

Pediatric Neonatology 59, 624-627


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]

Neurologist 22, 247-248

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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


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]

European Journal of Human Genetics 24, 852-856


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]


*Clinical Neuropathology* 34, 298-302

*Clinical Neuropathology* 34, 207-214

*Glia* 63, 2285-97

*Journal of the Neurological Sciences* 354, 131-132

*European Journal of Human Genetics* 23:72-78 *[the first nonsense mutation reported, predicting expression of a severely shortened protein]*

*European Journal of Human Genetics* 23:1-2 *[editorial discussing Nam et al.]*

2014

*Journal of Clinical & Diagnostic Research* 8, Pd03-04 [*full text*]

Lee KJ, Moon J, Lee ST. (2014). **Teaching NeuImages: Late-onset Alexander disease.**
*Neurology* 83, e197-198


2013


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[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

(Sincludes comparison of mutant keratin and GFAP)


[Sraises 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]


2012


2011


prepared by Albee Messing
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


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]


prepared by Albee Messing
[contains review of GFAP in blood or CSF as a potential biomarker for various diseases]  
[link to full article]

2008

American Journal of Neuroradiology 29, 1973-5

Brain 131, 2321-31  [same patients described by Farina 2008] [neuropsychology testing for some also described in Draghi 2019]

Neuroscience Research 62, 15-24

American Journal of Neuroradiology 29, 1190-6  [radiology features of same patients described by Pareyson 2008]

Movement Disorders 23, 1613-5

Human Molecular Genetics 17, 1540-1555  [link to full article]

Clinical Neuropathology 27, 64-71

Pediatric Neurology 38, 50-2

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2007


2006


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Neuropediatrics 37:v1112 (abstract)

Journal of Neuroscience 26, 11162-11173 [link to full article]

Journal of Child Neurology 21, 1075-80
[Provides additional clinical information on E207K patient initially reported in Van der Knaap, et al, 2005]

Journal of Biological Chemistry 281, 38634-38643 [link to full article]

American Journal of Neuroradiology 27, 2088-2092

Brain & Development 28, 663-667
[age of onset for this patient would be considered "infantile" according to our classification]

Journal of Korean Medical Science 21, 954-957

Neuroscience Letters 407, 127-130

Pediatric Neurology 35, 293-296

23

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2005


2004


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2003


Journal of Neurology 250, 300-306
[same patients for whom clinical/genetic data reported in Meins et al., 2002]

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


**2001**


prepared by Albee Messing
Neuroscience Letters  312, 71-74


2000


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

*Neuropediatrics* 31, 86-92

1999

*Alzheimer Disease & Associated Disorders* 13, 232-235

*Acta Neurologica Scandinavica* 99, 158-165

*Journal of Child Neurology* 14, 325-329  [see Li et al. 2005 for genetics]

Herndon RM. (1999). *Is Alexander's disease a nosologic entity or a common pathologic pattern of diverse etiology?* 
*Journal of Child Neurology* 14, 275-276

*Movement Disorders* 14, 689-693

*Archives of Ophthalmology* 117, 265-267

*Journal of the Neurological Sciences* 165, 116-120

*Nature Genetics* 21, 260-261  [likely mis-diagnosis]

prepared by Albee Messing
*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

prepared by Albee Messing


1995


1994


1993


prepared by Albee Messing


1992


1991

prepared by Albee Messing


1990

Journal of Histochemistry and Cytochemistry 38, 103-109


1989


1988


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


1984


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


1981

*Neuropediatrics*  12, 382-391

*Brain Research*  210, 419-425

prepared by Albee Messing

**1980**


**1979**


**1977**


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prepared by Albee Messing
**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**

Colmant HJ. (1973). *[Alexander's dystrophy with coexisting sudanophilic leukodystrophy and subacute necrotizing encephalopath (Leigh). Light and electron microscopy studies]*. 
[German]. 
*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."]

*Acta Neuropathologica* 4, 212-217

1962

*Acta Neuropathologica* 2, 126-143

1959

*Journal of Neuropathology and Experimental Neurology* 18, 359-383

prepared by Albee Messing
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