Alexander Disease Research Bibliography
(updated September 16, 2019)

Recently added:


2019


prepared by Albee Messing
Case Rep Med. 2019, 2986538

Neurocase. DOI: 10.1080/13554794.13552019.11580749

Indian Journal of Pediatrics (in press)

2018

Neuropediatrics 49, 256-261

Cell Stem Cell 23, 630-631. 
[editorial discussing the two new iPS cell papers, Li et al. and Jones et al.]

Cell Stem Cell 23, 239-251

Cell Reports 25, 947-958

Acta Neuropathologica Communications 6, 112 
[putative pathogenic variant in the minor isoform, GFAP-delta]

prepared by Albee Messing


prepared by Albee Messing


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]


prepared by Albee Messing


2016

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


intrafamilial phenotypic variability in infantile onset Alexander disease.
*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

*Neurology: Clinical Practice* 5:259-262

*Tidsskr Nor Laegeforen* 135, 1753-1755 [full text]

*Practical Neurology* 15, 393-395 [one of the patients with onset at 79 years]

*European Journal of Neurology* 22, 552-552 [meeting abstract - onset at 71 years, novel mutation]

*Nature Communications* 6, 6:8966 | DOI: 10.1038/ncomms9966 [full text]


prepared by Albee Messing
Journal of the Neurological Sciences  357, 319-321

*eNeuro*  DOI: 10.1523/ENEURO.0080-15.2015  [full text]

*PLoS One*  10, e0138132

*Acta Neuropathologica*  130, 469-486

*European Journal of Medical Genetics*  58, 466-70  [no deletions or duplications found]

*Clinical Neuropathology*  34, 298-302

*Clinical Neuropathology*  34, 207-214

*Glia*  63, 2285-97

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

prepared by Albee Messing


2014


prepared by Albee Messing


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)


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]


**2012**


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

*A Rosenthal Fiber Encephalomyelopathy Resembling Alexander's Disease in 3 Sheep.*  
Veterinary Pathology  49, 248-254  
[no GFAP mutations were detected]

*Alexander disease.*  
Journal of Neuroscience  32, 5017-5023  
[review]  
[link to full article]

Adult-onset Alexander disease mimicking neuromyelitis optica.  
*Multiple Sclerosis Journal*  18, 546-546  
[but may be mis-diagnosis, since D295N is a known polymorphism, and not proven as disease-causing]

*An unusual presentation of juvenile Alexander disease.*  
Journal of Child Neurology  27, 507-510

*Archetypal and new families with Alexander disease and novel mutations in GFAP.*  
Archives of Neurology  69, 208-214  
[includes genetics on patients and families originally described by Seil 1968, Duckett 1992, and Schwankhaus 1995]  
[link to full article]

*A novel adult case of juvenile-onset Alexander disease: complete remission of neurological symptoms for over 12 years, despite insidiously progressive cervicomedullary atrophy.*  
Neurological Sciences  33, 1389-1392

*Cerebellar ataxia as the first manifestation of Alexander’s disease.*  
Arquivos de Neuro-Psiquiatria  70, 309-310

prepared by Albee Messing


Yoshida, T., and Nakagawa, M. (2012). *Clinical aspects and pathology of Alexander disease, and morphological and functional alteration of astrocytes induced by GFAP mutation.* *Neuropathology* 32, 440-446

**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


[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

prepared by Albee Messing


prepared by Albee Messing


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


prepared by Albee Messing


prepared by Albee Messing

2007


Journal of Neurology  254, 1278-1280

*Experimental Cell Research*  313, 2077-2087  [review]  [full text]

The murine model of Alexander disease: analysis of GFAP aggregate formation and its pathological significance.  
*Glia*  55, 617-31

The functional alteration of mutant GFAP depends on the location of the domain: morphological and functional studies using astrocytoma-derived cells.  
*Journal of Human Genetics*  52, 362-9

Alexander disease with hypothermia, microcoria, and psychiatric and endocrine disturbances.  
*Neurology*  68, 1322-3

2006

Atypical variant of Alexander's disease with involvement of the spinal cord.  
*Neuropediatrics*  37:v1112 (abstract)

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

Unusual diagnosis in a child suffering from juvenile Alexander disease: clinical and imaging report.  
*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]

[age of onset for this patient would be considered "infantile" according to our classification]


_Brain & Development_ 28, 131-3  [not really the first, see Brenner et al, 2001 patient # 2]

_Human Genetics_ 119, 137-44

_American Journal of Pathology_ 168, 888-97

_Brain & Development_ 28, 60-2

2005

_Annals of Neurology_ 58, 813-4

_Neuropediatrics_ 36, 319-23

_Human Molecular Genetics_ 14, 2443-2458

_Journal of Cell Science_ 118, 2057-2065  [link to full article]

_No to Hattatsu [Brain & Development]_ 37, 55-59  [R239C patient]
Li R et al. (2005). *GFAP mutations in infantile, juvenile, and adult forms of Alexander disease.*
*Annals of Neurology* 57, 310-326  [See accompanying editorial]

*Annals of Neurology* 57, 327-338  [See accompanying editorial]

*Annals of Neurology* 57, 307-308  [Editorial]

**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**

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*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

*Neuropediatrics* 33, 194-198

*Neurology* 58, 1541-1543
[see Sawaishi et al., 1999, for more clinical detail on this patient]

*Journal of Child Neurology* 17, 227-230

*Neurology* 58, 1494 - 1500

prepared by Albee Messing


2001

Brenner M, Johnson AB, Boespflug-Tanguy O, Rodriguez D, Goldman JE, Messing A. (2001). **Mutations in GFAP, encoding glial fibrillary acidic protein, are associated with Alexander disease.** Nature Genetics 27, 117-120  [first description of genetics] [link to full article]


Journal of Neurochemistry 76, 730-736

Journal of Neuropathology and Experimental Neurology 60, 553

Journal of Neuropathology and Experimental Neurology 60 (abstract)

American Journal of Neuroradiology 22, 541-552 [link to full article]

2000

Epilepsia 41, 628-630

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]

Neurology India 47, 333-335.

1998

Brain Research 787, 15-18

American Journal of Pathology 152, 391-398

Pediatric Neurology 18, 67-70

1997

prepared by Albee Messing


1996


1995


prepared by Albee Messing

1994


1993


1992


prepared by Albee Messing
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

Immunoelectron microscopy with colloidal gold.
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].


Sorjonen DC, Cox NR, Kwapien RP. (1987). Myeloencephalopathy with eosinophilic refractile bodies (Rosenthal fibers) in a Scottish terrier.

1986


1985


1984


1983


1982


1981


1980


1979


1977


1976


1974


1973


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

prepared by Albee Messing
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

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

prepared by Albee Messing
Alexander WS. (1949). *Progressive fibrinoid degeneration of fibrillary astrocytes associated with mental retardation in a hydrocephalic infant.*

*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”]