Recently added:


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.]


2018


2017


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


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]


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


prepared by Albee Messing


**2015**


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*2014*


**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]


prepared by Albee Messing


2012


prepared by Albee Messing


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]


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

Alexander's disease: reassessment of a neonatal form. Childs Nervous System 28, 2029-2031

Late-onset Alexander disease with a V87L mutation in glial fibrillary acidic protein (GFAP) and calcifying lesions in the sub-cortex and cortex. Journal of Neurology 259, 457-461

Clinical aspects and pathology of Alexander disease, and morphological and functional alteration of astrocytes induced by GFAP mutation. Neuropathology 32, 440-446

GFAP mutations, age of onset, and clinical sub-types in Alexander disease. Neurology 77, 1287-94. [proposes new classification system, with updated survival statistics]

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]


2009


Hagemann TL, Boelens W, Wawrousek E, Messing A. (2009). **Suppression of GFAP toxicity by αB-crystallin in mouse models of Alexander disease.** *Human Molecular Genetics* 18, 1190-1199 [link to full article]


Yoshida T, Sasayama H, and Nakagawa M. (2009). **The process of inducing GFAP aggregates in astrocytoma-derived cells is different between R239C and R416W mutant GFAP. A time-lapse recording study.** *Neuroscience Letters* 458, 11-14


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


**2007**


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GFAP mutations and polymorphisms in 13 unrelated Italian patients affected by Alexander disease. Clinical Genetics 72, 427-433


2006


prepared by Albee Messing


2005


prepared by Albee Messing


**2004**


prepared by Albee Messing

**2003**


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


**2002**


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prepared by Albee Messing


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**1993**


prepared by Albee Messing


**1992**


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prepared by Albee Messing


[Included two Alexander disease patients]


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1990


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

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1989


1988


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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 (Méx.)* 12, 71-74

1949


[First description of a child with Alexander disease]

1898