

付録：表 2.2 - ヒトにおける突然変異に関する参考文献

この付録にはヒトにおける突然変異に関する表 2.2 に引用された参考文献が含まれている。

表 2.2 に関する参考文献

- Brown, P., et al. (1992) Familial Creutzfeldt-Jakob Disease in Chile is Associated With the Codon 200 Mutation of the PRNP Amyloid Precursor Gene on Chromosome 20, *Journal of Neurological Science*, **112**, 65–7
- Chapman, J., et al. (1994) The Risk of Developing Creutzfeldt-Jakob Disease in Subjects With the PRNP Gene Codon 200 Point Mutation, *Neurology*, **44**, 1683–6
- Collinge, J., et al. (1993) Inherited Prion Disease (PrP lysine 200) in Britain: Two Case Reports, *British Medical Journal*, **306**, 301–2
- Davies, P.T.G., et al. (1993) Creutzfeldt-Jakob Disease in Individual Occupationally Exposed to BSE, *The Lancet*, **342**, 680
- Farlow, M.R., et al. (1989) Gertsmann-Sträussler-Scheinker Disease. I. Extending the Clinical Spectrum, *Neurology*, **39**, 1446–52
- Furukawa, H., et al. (1995) New Variant Prion Protein in a Japanese Family with Gerstmann-Sträussler Syndrome, *Brain Research and Molecular Brain Research*, **30**, 385–8
- Ghetti, B., et al. (1989) Gertsmann-Sträussler-Scheinker Disease. II. Neurofibrillary Tangles and Plaques with PrP-Amyloid Coexist in an Affected Family, *Neurology*, **39**, 1453–61
- Goldfarb, L.G., et al. (1989) Patients with Creutzfeldt-Jakob Disease and Kuru Lack the Mutation in the PRNP Gene Found in Gerstmann-Sträussler Scheinker Syndrome, But they show a Different Double Allele Mutation in the Same Gene, *American Journal of Human Genetics (Suppl.)*, **45**, A189
- Goldfarb, L.G., et al. (1990) Mutation in Codon 200 of Scrapie Amyloid Protein Gene in Two Clusters of Creutzfeldt-Jakob Disease in Slovakia, *The Lancet*, **336**, 514–15
- Goldfarb, L.G., et al. (1990) Mutation in Codon 200 of Scrapie Amyloid Precursor Gene Linked to Creutzfeldt-Jakob Disease in Sephardic Jews of Libyan and Non-Libyan Origin, *The Lancet*, **336**, 637–8
- Goldfarb, L.G., et al. (1991) New Mutation in Scrapie Amyloid Precursor Gene (at codon 178) in Finnish Creutzfeldt-Jakob Kindred, *The Lancet*, **337**, 425
- Goldfarb, L.G., et al. (1991a) Transmissible Familial Creutzfeldt-Jakob Disease Associated with Five, Seven, and Eight Extra Octapeptide Coding Repeats in the PRNP Gene, *Proceedings of the National Academy of Sciences of the USA*, **88**, 10926–30

- Goldfarb, L.G., et al. (1991b) Creutzfeldt-Jakob Disease Associated with the PRNP Codon 200Lys Mutation: An Analysis of 45 Families, *European Journal of Epidemiology*, **7**, 477–86
- Goldfarb, L.G., et al. (1993) A New (two-repeat) Octapeptide Coding Inset Mutation in Creutzfeldt-Jakob Disease, *Neurology*, **43**, 2392–4
- Guiroy, D.C., et al. (1993) Immunolocalization of Scrapie Amyloid in Non-Congophilic, Non-Birefringent Deposits in Golden Syrian Hamsters with Experimental Transmissible Mink Encephalopathy, *Neuroscience Letters*, **155**, 112–15
- Hsiao, K.K., et al. (1989) Linkage of a Prion Protein Missense Variant to Gerstmann-Sträussler Syndrome, *Nature*, **338**, 342–5
- Hsiao, K.K., et al. (1991) Mutation of the Prion Protein in Libyan Jews with Creutzfeldt-Jakob Disease, *New England Journal of Medicine*, **324**, 1091–7
- Hsiao, K.K., et al. (1991b) A Prion Protein Variant in a Family With the Telencephalic Form of Gerstmann-Sträussler-Scheinker Syndrome, *Neurology*, **41**, 681–4
- Hsiao, K.K., et al. (1992) Mutant Prion Proteins in Gerstmann-Sträussler-Scheinker Disease with Neurofibrillary Tangles, *Nature Genetics*, **1**, 68–71
- Kitamoto, T., et al. (1993) An Amber Mutation of Prion Protein in Gerstmann-Sträussler Syndrome with Mutant PrP Plaques, *Biochemical and Biophysical Research Communications*, **30**, 525–31
- Kitamoto, T., et al. (1993a) Novel Missense Variants of Prion Protein in Creutzfeldt-Jakob Disease or Gerstmann-Sträussler Syndrome, *Biochemical and Biophysical Research Communications*, **191**, 709–14
- Korczyn, A.D., et al. (1991) A Mutation in the Prion Protein Gene in Creutzfeldt-Jakob Disease in Jewish Patients of Libyan, Greek and Tunisian Origin, *Annals of the New York Academy of Science*, **640**, 171–6
- Krasemann, S., et al. (1995) Prion Disease Associated with a Novel Nine Octapeptide Repeat Insertion in the PRNP Gene, *Molecular Brain Research*, **34**, 173–6
- Laplanche, J.L., et al. (1995) Deletion in Prion Gene in a Moroccan Family, *Nucleic Acids Research*, **18**, 22

- Laplanche, J.L., et al. (1995) Two Novel Insertions in the Prion Protein Gene in Patients with Late-Onset Dementia, *Human Molecular Genetics*, **4**, 1109–11
- Lugaresi, E., et al. (1986) Fatal Familial Insomnia and Dysautonomia, with Selective Degeneration of Thalamic Nuclei, *New England Journal of Medicine*, **315**, 997–1003
- Mastrianni, J.A., et al. (1995) Identification of a New Mutation of the Prion Protein Gene at Codon 128 in a Patient with Creutzfeldt-Jakob Disease, *Neurology*, **45** (Suppl. 4), A201
- Neufeld, M.Y., et al. (1992) Demyelinating Peripheral Neuropathy in Creutzfeldt-Jakob Disease, *Muscle and Nerve*, **15**, 1234–9
- Nicholl, D., et al. (1995) Inherited Creutzfeldt-Jakob Disease in a British Family Associated with a Novel 144 Base Pair Insertion of the Prion Protein Gene, *Journal of Neurology, Neurosurgery and Psychiatry*, **58**, 65–9
- Nitrini, R., et al. (1997) Familial Spongiform Encephalopathy Associated with a Novel Prion Protein Gene Mutation, *Annals of Neurology*, **42**, 138–46
- Oda, T., et al. (1995) Prion Disease with 144 Base Pair Insertion in a Japanese Family Line, *Acta Neuropathologica (Berlin)*, **90**, 80–6
- Owen, F., et al. (1989) Insertion in Prion Protein Gene in Familial Creutzfeldt-Jakob Disease, *The Lancet*, **1**, 51–2
- Palmer, M.S., et al. (1991) Homozygous Prion Protein Genotype Predisposes to Sporadic Creutzfeldt-Jakob Disease, *Nature*, **352**, 340–2
- Palmer, M.S., et al. (1993) Deletions in the Prion Protein Gene Are Not Associated With CJD, *Human Molecular Genetics*, **2**, 541–4
- Ripoll, L., et al. (1993) A New Point Mutation in the Prion Protein Gene at Codon 210 in Creutzfeldt-Jakob Disease, *Neurology*, **43**, 1934–8
- Tagliavini, F., et al. (1993) A68 is a Component of Paired Helical Filaments of Gertsman-Sträussler-Scheinker Disease, Indiana Kindred, *Brain Research*, **616**, 325–8
- Tateishi, J., et al. (1995) First Experimental Transmission of Fatal Familial Insomnia, *Nature*, **376**, 434–5
- Telling, G.C., et al. (1995) Prion Propagation in Mice Expressing Human and Chimeric PrP Transgenes Implicates the Interaction of Cellular PrP with another Protein, *Cell*, **83**, 79–90

Wu, Y., et al. (1987) A PvuII RFLP Detected in the Human Prion Protein (PrP) Gene, *Nucleic Acids Research*, **15**, 3191

Young, K., et al. (1997) Gerstmann-Sträussler-Scheinker Disease with the PRNP P102L Mutation and Valine at Codon 129, *Brain Research and Molecular Brain Research*, **44**, 147–50

Young, K., et al. (1998) Gerstmann-Sträussler-Scheinker Disease (GSS) with a Mutation at Prion Protein (PrP) residue 212, *Journal of Neuropathology and Experimental Neurology*, **57**, 518