

SCA2 – kirjallisuusviitteet

Burk K et al., Autosomal dominant cerebellar ataxia type I. Clinical features and MRI in families with SCA1, SCA2 and SCA3. *Brain* 119: 1497 – 1505, 1996

Imbert G et al., Cloning of the gene for spinocerebellar ataxia 2 reveals a locus with high sensitivity to expanded CAG/glutamine repeats. *Nat Genet* 14: 285 – 291, 1996

Pulst SM et al., Moderate expansion of a normally biallelic trinucleotide repeat in spinocerebellar ataxia type 2. *Nat Genet* 14: 269 – 276, 1996

Sanpei K et al: Identification of the spinocerebellar ataxia type 2 gene using a direct identification of repeat expansion and cloning technique, DIRECT. *Nat Genet* 14: 277 – 284, 1996

Adams C et al., Clinical and molecular analysis of a pedigree of southern Italian ancestry with spinocerebellar ataxia type 2. *Neurology* 49: 1163 – 1166, 1997

Cancel G et al., Molecular and clinical correlations in spinocerebellar ataxia 2: a study of 32 families. *Hum Mol Genet* 6: 709 – 715, 1997

Geschwind DH et al., The prevalence and wide clinical spectrum of the spinocerebellar ataxia type 2 trinucleotide repeat in patients with autosomal dominant cerebellar ataxia. *Am J Hum Genet* 60: 842 – 850, 1997

Kaakkola S ja Rinne R: Ataksiat ja niiden erotusdiagnostiikka (katsaus). *Duodecim* 113: 1773 – 1782, 1997

Lorenzetti D et al., The expansion of the CAG repeat in ataxin-2 is a frequent cause of autosomal dominant spinocerebellar ataxia. *Neurology* 49: 1009 – 1013, 1997

Buttner N et al., Oculomotor phenotypes in autosomal dominant ataxias. *Arch Neurol* 55: 1353 – 1357, 1998

Gambardella A et al., CAG repeat length and clinical features in three Italian families with spinocerebellar ataxia type 2 (SCA2): early impairment of Wisconsin Card Sorting Test and saccade velocity. *J Neurol* 245: 647 – 652, 1998

Grewal RP et al.: Clinical and genetic analysis of a distinct autosomal dominant spinocerebellar ataxia. *Neurology* 51: 1423 – 1426, 1998

Klockgether T and Evert B: Genes involved in hereditary ataxias. *Trends Neurosci* 21: 413 – 418, 1998

Klockgether T et al., The natural history of degenerative ataxia: a retrospective study in 466 patients. *Brain* 121: 589 – 600, 1998

Moseley ML et al., Incidence of dominant spinocerebellar and Friedreich triplet repeats among 361 ataxia families. *Neurology* 51: 1666 – 1671, 1998

Bürk K et al., Cognitive deficits in spinocerebellar ataxia 2. *Brain* 122: 769 – 777, 1999

Huynh DP et al., Expression of Ataxin-2 in brain from normal individuals and patients with Alzheimer`s Disease and Spinocerebellar Ataxia 2. *Ann Neurol* 45: 232 –241, 1999

Storey E et al., Spinocerebellar Ataxia Type 2. Clinical features of a pedigree displaying prominent frontal-executive dysfunction. *Arch Neurol* 56: 43 – 50, 1999

Bürk K and Dichgans J: Spinocerebellar Ataxia Type 2. Kirjassa: Handbook of Ataxia Disorders, 363 - 384. Ed. Klockgether T. Marcel Dekker Inc., New York-Basel, 2000

Costanzi-Porrini S et al., An interrupted 34-CAG repeat SCA-2 allele in patients with sporadic spinocerebellar ataxia. *Neurology* 54: 491 – 493, 2000

Fernandez M et al., Late-onset SCA2: 33 CAG repeats are sufficient to cause disease. *Neurology* 55: 569 – 572, 2000

Gwinn-Hardy K et al., Spinocerebellar ataxia type 2 with parkinsonism in ethnic Chinese. *Neurology* 55: 800 – 805, 2000

Hayes S et al., CAG repeat length in RAI1 is associated with age at onset variability in spinocerebellar ataxia type 2 (SCA2). *Hum Mol Genet* 9: 1753 – 1758, 2000

Huynh DP et al., Nuclear localization or inclusion body formation of ataxin-2 are not necessary for SCA2 pathogenesis in mouse or human. *Nature Genetics* 26: 44 – 50, 2000

Saleem O et al., Molecular analysis of autosomal dominant hereditary ataxias in the Indian population: high frequency of SCA2 and evidence for common founder mutation. *Hum Genet* 106: 179 – 187, 2000

Storey E et al., Frequency of spinocerebellar ataxia types 1, 2, 3, 6, and 7 in Australian patients with spinocerebellar ataxia. *Am J Med Genet* 95: 351 – 357, 2000

Tang B et al., Frequency of SCA1, SCA2, SCA3/MJD, SCA6, SCA7, and DRPLA CAG trinucleotide repeat expansions in patients with hereditary spinocerebellar ataxia from Chinese kindreds. *Arch Neurol* 57: 540 – 544, 2000

Abele M et al., Restless legs syndrome in spinocerebellar ataxia types 1, 2, and 3. *J Neurol* 248: 311 – 4, 2001

Boesch SM et al., Proton magnetic resonance spectroscopic imaging reveals differences in spinocerebellar ataxia types 2 and 6. *J Magn Reson Imaging* 13: 553 – 559, 2001

Furtado S et al., SCA-2 presenting as parkinsonism in an Alberta family. Clinical, genetic, and PET findings. *Neurology* 59: 1625 – 1627, 2002

Huynh DP et al., Expansion of the polyQ repeat in ataxin-2 alters its Golgi localization, disrupts the Golgi complex and causes cell death. *Hum Mol Genet* 12: 1485 – 1496, 2003

Boesch SM et al., Abnormalities of dopaminergic neurotransmission in SCA2: a combined 123I-betaCIT and 123I-IBZM SPECT study.

Guerrini L et al., Brainstem neurodegeneration correlates with clinical dysfunction in SCA1 but not in SCA2. A quantitative volumetric, diffusion and proton spectroscopy MR study. *Brain* 127: 1785 – 1795, 2004

Infante J et al., Spinocerebellar ataxia type 2 with Levodopa-responsive parkinsonism culminating in motor neuron disease. *Mov Disord* 19: 848 – 952, 2004

Lu C-C et al., The parkinsonian phenotype of spinocerebellar ataxia type 2. *Arch Neurol* 61: 35 – 38, 2004

Morretti P et al., Spinocerebellar ataxia type 2 (SCA2) presenting with ophthalmoplegia and developmental delay in infancy. *Am J Med Genet* 124: 392 – 396, 2004

Rüb U et al., Damage to the reticulotegmental nucleus of pons in spinocerebellar ataxia type 1, 2, and 3. *Neurology* 63: 1258 – 1263, 2004

Sinha KK et al., Autosomal dominant cerebellar ataxia: SCA2 is the most frequent mutation in Eastern India. *J Neurol Neurosurg Psychiatry* 75: 448 – 452, 2004

van de Warrenburg BPC et al., Peripheral nerve involvement in spinocerebellar ataxias. *Arch Neurol* 61: 257 – 261, 2004

Gierga K et al., Involvement of the cranial nerves and their nuclei in spinocerebellar ataxia type 2 (SCA2). *Acta Neuropathol* 109: 617 – 131, 2005

Inagaki A et al., Positron emission tomography and magnetic resonance imaging in spinocerebellar ataxia type 2: a study of symptomatic and asymptomatic individuals. *Eur J Neurol* 12: 725 – 728, 2005

Koeppen AH: The pathogenesis of spinocerebellar ataxia. *Cerebellum* 4: 62 – 73, 2005

Mascke M et al., Clinical feature profile of spinocerebellar ataxia type 1 – 8 predicts genetically defined subtypes. *Mov Disord* 20: 1405 – 1412, 2005

Pulst SM et al., Spinocerebellar ataxia type 2: polyQ repeat variation in the CACBA1A calcium channel modifies age at onset. *Brain* 128: 2297 – 2303, 2005

Rüb U et al., Spinocerebellar ataxias type 2 and 3: degeneration of the precerebellar nuclei isolates the three phylogenetically defined regions of the cerebellum. *J Neural Transm* 112: 1523 – 1545, 2005

Scifried C et al., Saccade velocity as a surrogate disease marker in spinocerebellar ataxia type 2. *Ann N Y Acad Sci* 1039: 524 – 527, 2005

van de Warrenburg BPC et al., Age at onset variance analysis in spinocerebellar ataxias: a study in a Dutch-French cohort. *Ann Neurol* 57: 505 – 512, 2005

Wüllner U et al., Dopamine transporter positron emission tomography in spinocerebellar ataxias type 1, 2, 3 and 6. *Arch Neurol* 62: 1280 – 1285, 2005

Crum BA and Josephs KA: Varied electrophysiologic patterns in spinocerebellar ataxia type 2. *Eur J Neurol* 13: 104 – 197, 2006

Gonatas NK et al., Fragmentation of the Golgi apparatus in neurodegenerative diseases and cell death. *J Neurol Sci*

de Rosa A et al., Suppression myoklonus in SCA2 by piracetam. *Mov Disord* 21: 116 – 118, 2006