

## Publicatielijst CADASIL LUMC

1. Rutten JW, Haan J, Terwindt GM, van Duinen SG, Boon EM, Lesnik Oberstein SAJ. Interpretation of NOTCH3 mutations in the diagnosis of CADASIL. Expert Rev Mol Diagn. 2014 Jun;14(5):593-603.
2. Opherk C, Gonik M, Duering M, Malik R, Jouvent E, Hervé D, Adib-Samii P, Bevan S, Pianese L, Silvestri S, Dotti MT, De Stefano N, Liem M, Boon EM, Pescini F, Pachai C, Bracoud L, Müller-Myhsok B, Meitinger T, Rost N, Pantoni L, Oberstein SL, Federico A, Ragno M, Markus HS, Tournier-Lasserre E, Rosand J, Chabriat H, Dichgans M. *Genome-wide genotyping demonstrates a polygenic risk score associated with white matter hyperintensity volume in CADASIL.* **Stroke.** 2014 Apr;45(4):968-72
3. Rutten JW, Boon EM, Liem MK, Dauwelse JG, Pont MJ, Vollebregt E, Maat-Kievit AJ, Ginjaar HB, Lakeman P, van Duinen SG, Terwindt GM, Lesnik Oberstein SA. *Hypomorphic NOTCH3 alleles do not cause CADASIL in humans.* **Hum Mutat.** 2013 Nov;34(11):1486-9.
4. Liem MK, Lesnik Oberstein SA, Versluis MJ, Maat-Schieman ML, Haan J, Webb AG, Ferrari MD, van Buchem MA, van der Grond J. *7 T MRI reveals diffuse iron deposition in putamen and caudate nucleus in CADASIL.* **JNNP** 2012 Dec;83(12):1180-5.
5. Liem MK, Lesnik Oberstein SA, van der Grond J, Ferrari MD, Haan J. *CADASIL and migraine: A narrative review.* **Cephalgia.** 2010; 30(11):1284-1289
6. Liem MK, van der Grond J, Versluis MJ, Haan J, Webb AG, Ferrari, MD, van Buchem MA, Lesnik Oberstein SA. *Lenticulostriate arterial lumina are normal in CADASIL – a high field *in vivo* magnetic resonance imaging study.* **Stroke.** 2010; 41(12):2812-2816
7. Liem MK, Lesnik Oberstein SA, Haan J et al. *MRI correlates of cognitive decline in CADASIL: a 7-year follow-up study.* **Neurology.** 2009; 72:143-148
8. Liem MK, Lesnik Oberstein SA, Haan J et al. *Cerebrovascular reactivity is a main determinant of white matter hyperintensity progression in CADASIL.* **AJNR.** 2009; 30:1244-1247
9. Lesnik Oberstein SA, Maat-Schieman ML, Boon EM et al. *No vessel wall abnormalities in a human foetus with a NOTCH3 mutation.* **Acta Neuropathol.** 2008; 115:369-370
10. Liem MK, Lesnik Oberstein SA, Vollebregt MJ et al. *Homozygosity for a NOTCH3 mutation in a 65-year-old CADASIL patient with mild symptom a family report.* **J Neurol.** 2008; 255:1978-1980
11. Liem MK, Lesnik Oberstein SA, Haan J et al. *Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy: progression of MR abnormalities in prospective 7-year follow-up study.* **Radiology.** 2008; 249:964-971
12. Liem MK, van der Grond J, Haan J, van den Boom R, Ferrari MD, Knaap YM, Breuning MH, van Buchem MA, Middelkoop HAM, Lesnik Oberstein SA. *Lacunar infarcts are the main correlate with cognitive dysfunction in CADASIL.* **Stroke.** 2007; 38:923-928

13. Haan J, Lesnik Oberstein SA, Ferrari MD. *Epilepsy in cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy*. **Cerebrovasc Dis.** 2007; 24:316-317
14. van den Boom R, Oberstein SAJL, van den Berg-Huysmans A, Ferrari M, van Buchem M, Haan J. *Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy: Structural MR imaging changes and apolipoprotein E genotype*. **AJNR.** 2006; 27(2):359-362
15. Lesnik Oberstein SA and Haan J. *Cerebral autosomal dominant Arteriopathy with Subcortical Infarcts and Leukoencephalopathy (CADASIL)*. **Panminerva Medica.** 2004; 46.4:265-276
16. Oberstein SAJL and Haan J. *Diagnostic strategies in CADASIL*. **Neurology.** 2003; 60.12:2020
17. Oberstein SAJL, van den Boom R, Middelkoop HA et al. *Incipient CADASIL*. **Arch Neurol.** 2003; 60:707-712
18. Oberstein SAJL, van Duinen SG, van den Boom R et al. *Evaluation of diagnostic NOTCH3 immunostaining in CADASIL*. **Acta Neuropathol.** 2003; 106:107-111
19. van den Boom R, Oberstein SAL, Spilt A et al. *Cerebral hemodynamics and white matter hyperintensities in CADASIL*. **J Cereb Blood Flow Metab.** 2003; 23:599-604
20. Oberstein SAJL, Jukema JW, van Duinen SG et al. *Myocardial infarction in cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL)*. **Medicine (Baltimore).** 2003; 82:251-256
21. van den Boom R, Oberstein SAJL, Ferrari MD et al. *Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy: MR imaging findings at different ages--3rd-6th decades*. **Radiology.** 2003; 229:683-690
22. van den Boom R, Lesnik SAJ, van Duinen SG et al. *Subcortical lacunar lesion an MR imaging finding in patients with cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy*. **Radiology.** 2002; 224:791-796
23. Oberstein SAJL, van den Boom R, Van Buchem MA et al. *Cerebral microbleeds in CADASIL*. **Neurology** 2001; 57:1066-1070
24. Lesnik Oberstein SA, Bakker E, Ferrari MD et al. [From gene to disease; from Notch3 to cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy]. **Ned Tijdschr Geneeskde.** 2001; 145:359-360
25. Oberstein SAJL, Ferrari MD, Bakker E et al. *Diagnostic Notch3 sequence analysis in CADASIL: three new mutations in Dutch patients*. **Neurology.** 1999; 52:1913-1915