

Neurofibromatose Erasmus MC/ ENCORE publicaties:

- Van Der Vaart, T. *et al.* (2013) Simvastatin for cognitive deficits and behavioural problems in patients with neurofibromatosis type 1 (NF1-SIMCODA): a randomised, placebo-controlled trial. *Lancet Neurol* 12, 1076–1083
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- van der Vaart, T. *et al.* (2011) Motor deficits in neurofibromatosis type 1 mice: the role of the cerebellum. *Genes Brain Behav* 10, 404–409
- Krab, L.C. *et al.* (2011) Motor learning in children with neurofibromatosis type I. *Cerebellum* 10, 14–21
- Verdijk, R.M. *et al.* (2010) TP53 mutation analysis of malignant peripheral nerve sheath tumors. *J Neuropathol Exp Neurol* 69, 16–26
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- van Engelen, S.J.P.M. *et al.* (2008) Quantitative differentiation between healthy and disordered brain matter in patients with neurofibromatosis type I using diffusion tensor imaging. *AJNR American journal of neuroradiology* 29, 816–822
- Oostenbrink, R. *et al.* (2007) Parental reports of health-related quality of life in young children with neurofibromatosis type 1: influence of condition specific determinants. *J Pediatr* 151, 182–6– 186.e1–2
- Crossen, M.H. *et al.* (1998) Minor disease features in neurofibromatosis type 1 (NF1) and their possible value in diagnosis of NF1 in children. *J Med Genet* 35, 624–627
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- Crossen, M.H. *et al.* (1997) Diagnostic delay in neurofibromatosis type 1. *Eur J Pediatr* 156, 482–487
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Tubereuze sclerose Erasmus MC/ ENCORE publicaties:

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- van Eeghen, A.M. *et al.* (2012) Understanding relationships between autism, intelligence, and epilepsy: a cross-disorder approach. *Developmental Medicine & Child Neurology* 55, 146–153
- Hoogeveen-Westerveld, M. *et al.* (2012) Functional Assessment of TSC2 Variants Identified in Individuals with Tuberous Sclerosis Complex. *Hum Mutat* DOI: 10.1002/humu.22202
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- Sancak, O. *et al.* (2005) Mutational analysis of the TSC1 and TSC2 genes in a diagnostic setting: genotype-phenotype correlations and comparison of diagnostic DNA techniques in Tuberous Sclerosis Complex. *European Journal of Human Genetics* 13, 731–741
- Nellist, M. *et al.* (2005) Large deletion at the TSC1 locus in a family with tuberous sclerosis complex. *Genet. Test.* 9, 226–230
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- van Sleightenhorst, M. *et al.* (1997) Identification of the tuberous sclerosis gene TSC1 on chromosome 9q34. *Science* 277, 805–808
- Rinke de Wit, T.F. *et al.* (1996) Expression of tyrosine kinase gene in mouse thymic stromal cells. *Int. Immunol.* 8, 1787–1795
- Vrtel, R. *et al.* (1996) Identification of a nonsense mutation at the 5' end of the TSC2 gene in a family with a presumptive diagnosis of tuberous sclerosis complex. *J Med Genet* 33, 47–51
- Halley, D.J. (1996) Tuberous sclerosis: between genetic and physical analysis. *Acta Genet Med Gemellol (Roma)* 45, 63–75
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- van Woerden, G.M. *et al.* (2007) Rescue of neurological deficits in a mouse model for Angelman syndrome by reduction of α CaMKII inhibitory phosphorylation. *Nat Neurosci* 10, 280–282
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- Buiting, K. *et al.* (1998) Sporadic imprinting defects in Prader-Willi syndrome and Angelman syndrome: implications for imprint-switch models, genetic counseling, and prenatal diagnosis. *Am J Hum Genet* 63, 170–180
- Horsthemke, B. *et al.* (1996) Familial translocations involving 15q11-q13 can give rise to interstitial deletions causing Prader-Willi or Angelman syndrome. *J Med Genet* 33, 848–851
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- van 't Padje, S. *et al.* (2005) Characterisation of Fmrp in zebrafish: evolutionary dynamics of the fmr1 gene. *Dev. Genes Evol.* 215, 198–206
- Blonden, L. *et al.* (2005) Two members of the Fxr gene family, Fmr1 and Fxr1, are differentially expressed in *Xenopus tropicalis*. *Int. J. Dev. Biol.* 49, 437–441
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- Faust, C.J. *et al.* (1992) Genetic mapping on the mouse X chromosome of human cDNA clones for the fragile X and Hunter syndromes. *Genomics* 12, 814–817
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Costello Syndroom Erasmus MC/ ENCORE publicaties:

Publicaties:

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- Poulton, C. *et al.* (2013) Progressive cerebellar atrophy and polyneuropathy: expanding the spectrum of PNKP mutations. *Neurogenetics* 14, 43–51
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Autisme Erasmus MC publicaties:

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- Marroun, El, H. *et al.* (2013) Prenatal Tobacco Exposure and Brain Morphology: A Prospective Study in Young Children. *Neuropsychopharmacology* DOI: 10.1038/npp.2013.273
- Louwerse, A. *et al.* (2013) Autonomic Responses to Social and Nonsocial Pictures in Adolescents With Autism Spectrum Disorder. *Autism Res* DOI: 10.1002/aur.1327
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