

## 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
- van Minkelen, R. *et al.* (2013) A clinical and genetic overview of 18 years neurofibromatosis type 1 molecular diagnostics in the Netherlands. *Clin Genet* DOI: 10.1111/cge.12187
- Castrén, E. *et al.* (2012) Treatment of neurodevelopmental disorders in adulthood. *J Neurosci* 32, 14074–14079
- Acosta, M.T. *et al.* (2012) The Learning Disabilities Network (LeaDNet): using neurofibromatosis type 1 (NF1) as a paradigm for translational research. *Am J Med Genet* 158, 2225–2232
- 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
- Krab, L.C. *et al.* (2009) Health-related quality of life in children with neurofibromatosis type 1: contribution of demographic factors, disease-related factors, and behavior. *J Pediatr* 154, 420–5, 425.e1
- D Krab, L.C. *et al.* (2008) Oncogenes on my mind: ERK and MTOR signaling in cognitive diseases. *Trends Genet* 24, 498–510
- enayer, E. *et al.* (2008) Spred1 is required for synaptic plasticity and hippocampus-dependent learning. *J Neurosci* 28, 14443–14449
- Cui, Y. *et al.* (2008) Neurofibromin regulation of ERK signaling modulates GABA release and learning. *Cell* 135, 549–560
- Krab, L.C. *et al.* (2008) Impact of neurofibromatosis type 1 on school performance. *J Child Neurol* 23, 1002–1010
- Krab, L.C. *et al.* (2008) Effect of simvastatin on cognitive functioning in children with neurofibromatosis type 1: a randomized controlled trial. *JAMA* 300, 287–294
- Balgobind, B.V. *et al.* (2008) Leukemia-associated NF1 inactivation in patients with pediatric T-ALL and AML lacking evidence for neurofibromatosis. *Blood* 111, 4322–4328
- 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
- Crossen, M.H. *et al.* (1998) A prospective 10 year follow up study of patients with neurofibromatosis type 1. *Arch. Dis. Child.* 78, 408–412
- Crossen, M.H. *et al.* (1997) Endocrinologic disorders and optic pathway gliomas in children with neurofibromatosis type 1. *Pediatrics* 100, 667–670
- Crossen, M.H. *et al.* (1997) Diagnostic delay in neurofibromatosis type 1. *Eur J Pediatr* 156, 482–487
- Crossen, M.H. *et al.* (1997) Deletions spanning the neurofibromatosis type 1 gene: implications for genotype-phenotype correlations in neurofibromatosis type 1? *Hum Mutat* 9, 458–464

## Tubereuze sclerose Erasmus MC/ ENCORE publicaties:

- Abs, E. *et al.* (2013) TORC1-dependent epilepsy caused by acute biallelic Tsc1 deletion in adult mice. *Ann Neurol* 74, 569–579
- Overwater, I.E. *et al.* (2013) Behandelingen voor genetische neurocognitieve aandoeningen. *Neuropraxis* 5, 132–138
- van Eeghen, A.M. *et al.* (2013) The neuroanatomical phenotype of tuberous sclerosis complex: focus on radial migration lines. *Neuroradiology* 55, 1007–1014
- Melser, S. *et al.* (2013) Rheb regulates mitophagy induced by mitochondrial energetic status. *Cell Metabolism* 17, 719–730
- Castrén, E. *et al.* (2012) Treatment of neurodevelopmental disorders in adulthood. *J Neurosci* 32, 14074–14079
- van Eeghen, A.M. *et al.* (2013) Central TSC2 missense mutations are associated with a reduced risk of infantile spasms. *Epilepsy Res* 103, 83–87
- 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
- van Eeghen, A.M. *et al.* (2012) Genotype and cognitive phenotype of patients with tuberous sclerosis complex. *Eur J Hum Genet* 20, 510–515
- Hoogeveen-Westerveld, M. *et al.* (2012) Functional assessment of TSC1 missense variants identified in individuals with tuberous sclerosis complex. *Hum Mutat* 33, 476–479
- van Eeghen, A.M. *et al.* (2012) Cognitive and adaptive development of patients with tuberous sclerosis complex: A retrospective, longitudinal investigation. *Epilepsy & Behavior* 23, 10–15
- Goorden, S.M.I. *et al.* (2011) Rheb is essential for murine development. *Mol Cell Biol* 31, 1672–1678
- Goorden, S.M.I. and Elgersma, Y. (2011) Rheb: enrichment beyond the brain. *Cell Cycle* 10, 2412–2413
- van den Ouweland, A.M.W. *et al.* (2011) Characterisation of TSC1 promoter deletions in tuberous sclerosis complex patients. *Eur J Hum Genet* 19, 157–163
- van Eeghen, A.M. *et al.* (2011) Characterizing sleep disorders of adults with tuberous sclerosis complex: a questionnaire-based study and review. *Epilepsy Behav* 20, 68–74
- Krab, L.C. *et al.* (2008) Oncogenes on my mind: ERK and MTOR signaling in cognitive diseases. *Trends Genet* 24, 498–510
- Goorden, S.M.I. *et al.* (2007) Cognitive deficits in Tsc1<sup>+/-</sup> mice in the absence of cerebral lesions and seizures. *Ann Neurol* 62, 648–655
- 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
- Nellist, M. *et al.* (2003) Regulation of tuberous sclerosis complex (TSC) function by 14-3-3 proteins. *Biochem Soc Trans* 31, 587–591
- Nellist, M. *et al.* (2001) TSC2 missense mutations inhibit tuberin phosphorylation and prevent formation of the tuberin-hamartin complex. *Hum Mol Genet* 10, 2889–2898
- Goedbloed, M.A. *et al.* (2001) Analysis of TSC2 stop codon variants found in tuberous sclerosis patients. *European Journal of Human Genetics* 9, 823–828
- Nellist, M. *et al.* (1999) Characterization of the cytosolic tuberin-hamartin complex. Tuberin is a cytosolic chaperone for hamartin. *J Biol Chem* 274, 35647–35652
- Verhoef, S. *et al.* (1999) High rate of mosaicism in tuberous sclerosis complex. *Am J Hum Genet* 64, 1632–1637
- van Slegtenhorst, M. *et al.* (1999) Mutational spectrum of the TSC1 gene in a cohort of 225 tuberous sclerosis complex patients: no evidence for genotype-phenotype correlation. *J Med Genet* 36, 285–289
- Verhoef, S. *et al.* (1999) Malignant pancreatic tumour within the spectrum of tuberous sclerosis complex in childhood. *Eur J Pediatr* 158, 284–287
- van Slegtenhorst, M. *et al.* (1998) Interaction between hamartin and tuberin, the TSC1 and TSC2 gene products. *Hum Mol Genet* 7, 1053–1057

- Wang, Q. *et al.* (1998) Identification of a large insertion and two novel point mutations (3671del8 and S1221X) in tuberous sclerosis complex (TSC) patients. Mutations in brief no. 119. Online. *Hum Mutat* 11, 331–332
- 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
- van Sleightenhorst, M. *et al.* (1995) Cosmid contigs from the tuberous sclerosis candidate region on chromosome 9q34. *European Journal of Human Genetics* 3, 78–86
- Janssen, B. *et al.* (1994) Refined localization of TSC1 by combined analysis of 9q34 and 16p13 data in 14 tuberous sclerosis families. *Hum Genet* 94, 437–440

### **Angelman Syndrome Erasmus MC/ ENCORE publicaties:**

- Steinkellner, T. *et al.* (2012) Ca(2+)/calmodulin-dependent protein kinase II $\alpha$  ( $\alpha$ CaMKII) controls the activity of the dopamine transporter: implications for Angelman syndrome. *J Biol Chem* 287, 29627–29635
- van Woerden, G.M. *et al.* (2007) Rescue of neurological deficits in a mouse model for Angelman syndrome by reduction of  $\alpha$ CaMKII inhibitory phosphorylation. *Nat Neurosci* 10, 280–282
- Elgersma, Y. (2007) Genetic engineering cures mice of neurological deficits: prospects for treating Angelman syndrome. *Pharmacogenomics* 8, 539–541
- van den Ouweland, A.M. *et al.* (1999) Angelman syndrome: AS phenotype correlated with specific EEG pattern may result in a high detection rate of mutations in the UBE3A gene. *J Med Genet* 36, 723–724
- Fang, P. *et al.* (1999) The spectrum of mutations in UBE3A causing Angelman syndrome. *Hum Mol Genet* 8, 129–135
- 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
- van den Ouweland, A.M. *et al.* (1995) DNA diagnosis of Prader-Willi and Angelman syndromes with the probe PW71 (D15S63). *Hum Genet* 95, 562–567

### **Fragile X Syndrome Erasmus MC/ ENCORE publicaties:**

- Pop, A.S. *et al.* (2012) Rescue of dendritic spine phenotype in Fmr1 KO mice with the mGluR5 antagonist AFQ056/Mavoglurant. *Psychopharmacology (Berl.)* DOI: 10.1007/s00213-012-2947-y
- Pop, A.S. *et al.* (2013) Fragile X syndrome: a preclinical review on metabotropic glutamate receptor 5 (mGluR5) antagonists and drug development. *Psychopharmacology (Berl.)* DOI: 10.1007/s00213-013-3330-3
- Vinueza Veloz, M.F. *et al.* (2012) The effect of an mGluR5 inhibitor on procedural memory and avoidance discrimination impairments in Fmr1 KO mice. *Genes Brain Behav* 11, 325–331
- de Esch, C.E.F. *et al.* (2013) Translational endpoints in fragile X syndrome. *Neurosci Biobehav Rev* DOI: 10.1016/j.neubiorev.2013.10.012
- Levenga, J. *et al.* (2011) AFQ056, a new mGluR5 antagonist for treatment of fragile X syndrome. *Neurobiol Dis* 42, 311–317
- Levenga, J. *et al.* (2010) Potential therapeutic interventions for fragile X syndrome. *Trends*

- Levenga, J. *et al.* (2011) Subregion-specific dendritic spine abnormalities in the hippocampus of Fmr1 KO mice. *Neurobiol Learn Mem* 95, 467–472
- Brouwer, J.R. *et al.* (2009) The FMR1 gene and fragile X-associated tremor/ataxia syndrome. *American journal of medical genetics Part B, Neuropsychiatric genetics : the official publication of the International Society of Psychiatric Genetics* 150, 782–798
- Levenga, J. *et al.* (2009) Ultrastructural analysis of the functional domains in FMRP using primary hippocampal mouse neurons. *Neurobiol Dis* 35, 241–250
- Van't Padje, S. *et al.* (2009) Reduction in fragile X related 1 protein causes cardiomyopathy and muscular dystrophy in zebrafish. *J. Exp. Biol.* 212, 2564–2570
- Brouwer, J.R. *et al.* (2008) CGG-repeat length and neuropathological and molecular correlates in a mouse model for fragile X-associated tremor/ataxia syndrome. *J Neurochem* 107, 1671–1682
- Smit, A.E. *et al.* (2008) Savings and extinction of conditioned eyeblink responses in fragile X syndrome. *Genes Brain Behav* 7, 770–777
- de Vrij, F.M.S. *et al.* (2008) Rescue of behavioral phenotype and neuronal protrusion morphology in Fmr1 KO mice. *Neurobiol Dis* 31, 127–132
- Brouwer, J.R. *et al.* (2008) Altered hypothalamus-pituitary-adrenal gland axis regulation in the expanded CGG-repeat mouse model for fragile X-associated tremor/ataxia syndrome. *Psychoneuroendocrinology* 33, 863–873
- Govaerts, L.C.P. *et al.* (2007) Exceptional good cognitive and phenotypic profile in a male carrying a mosaic mutation in the FMR1 gene. *Clin Genet* 72, 138–144
- Brouwer, J.R. *et al.* (2007) Elevated Fmr1 mRNA levels and reduced protein expression in a mouse model with an unmethylated Fragile X full mutation. *Exp Cell Res* 313, 244–253
- Mientjes, E.J. *et al.* (2006) The generation of a conditional Fmr1 knock out mouse model to study Fmrp function in vivo. *Neurobiol Dis* 21, 549–555
- Koekkoek, S.K.E. *et al.* (2005) Deletion of FMR1 in Purkinje cells enhances parallel fiber LTD, enlarges spines, and attenuates cerebellar eyelid conditioning in Fragile X syndrome. *Neuron* 47, 339–352
- 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
- Stoyanova, V. *et al.* (2004) Loss of FMR1 hypermethylation in somatic cell heterokaryons. *FASEB J.* 18, 1964–1966
- Schrier, M. *et al.* (2004) Transport kinetics of FMRP containing the I304N mutation of severe fragile X syndrome in neurites of living rat PC12 cells. *Exp Neurol* 189, 343–353
- Mientjes, E.J. *et al.* (2004) Fxr1 knockout mice show a striated muscle phenotype: implications for Fxr1p function in vivo. *Hum Mol Genet* 13, 1291–1302
- De Diego Otero, Y. *et al.* (2002) Transport of fragile X mental retardation protein via granules in neurites of PC12 cells. *Mol Cell Biol* 22, 8332–8341
- Willemsen, R. *et al.* (2002) Timing of the absence of FMR1 expression in full mutation chorionic villi. *Hum Genet* 110, 601–605
- Bontekoe, C.J.M. *et al.* (2002) Knockout mouse model for Fxr2: a model for mental retardation. *Hum Mol Genet* 11, 487–498
- Bontekoe, C.J. *et al.* (2001) Instability of a (CGG)<sub>98</sub> repeat in the Fmr1 promoter. *Hum Mol Genet* 10, 1693–1699
- Willemsen, R. *et al.* (2000) Twin sisters, monozygotic with the fragile X mutation, but with a different phenotype. *J Med Genet* 37, 603–604
- Bakker, C.E. *et al.* (2000) Immunocytochemical and biochemical characterization of FMRP, FXR1P, and FXR2P in the mouse. *Exp Cell Res* 258, 162–170
- Tamanini, F. *et al.* (2000) The fragile X-related proteins FXR1P and FXR2P contain a functional nucleolar-targeting signal equivalent to the HIV-1 regulatory proteins. *Hum Mol Genet* 9, 1487–1493
- Willemsen, R. *et al.* (1999) Noninvasive test for fragile X syndrome, using hair root analysis. *Am J Hum Genet* 65, 98–103
- de Vries, B.B. *et al.* (1999) Screening for the fragile X syndrome among the mentally retarded: a clinical study. The Collaborative Fragile X Study Group. *J Med Genet* 36, 467–470

- Tamanini, F. *et al.* (1999) Different targets for the fragile X-related proteins revealed by their distinct nuclear localizations. *Hum Mol Genet* 8, 863–869
- de Vries, B.B. *et al.* (1999) Dilemmas in counselling females with the fragile X syndrome. *J Med Genet* 36, 167–170
- Wildhagen, M.F. *et al.* (1999) Efficacy of cascade testing for fragile X syndrome. *J Med Screen* 6, 70–76
- de Vries, B.B. *et al.* (1998) Screening with the FMR1 protein test among mentally retarded males. *Hum Genet* 103, 520–522
- Verheij, C. *et al.* (1993) Characterization and localization of the FMR-1 gene product associated with fragile X syndrome. *Nature* 363, 722–724
- Reyniers, E. *et al.* (1993) The full mutation in the FMR-1 gene of male fragile X patients is absent in their sperm. *Nat Genet* 4, 143–146
- Verkerk, A.J. *et al.* (1993) Alternative splicing in the fragile X gene FMR1. *Hum Mol Genet* 2, 399–404
- De Boule, K. *et al.* (1993) A point mutation in the FMR-1 gene associated with fragile X mental retardation. *Nat Genet* 3, 31–35
- de Vries, B.B. *et al.* (1993) Mental status and fragile X expression in relation to FMR-1 gene mutation. *European Journal of Human Genetics* 1, 72–79
- Willems, P.J. *et al.* (1992) Segregation of the fragile X mutation from an affected male to his normal daughter. *Hum Mol Genet* 1, 511–515
- Verkerk, A.J. *et al.* (1992) Intragenic probe used for diagnostics in fragile X families. *Am J Med Genet* 43, 192–196
- Oostra, B.A. and Verkerk, A.J. (1992) The fragile X syndrome: isolation of the FMR-1 gene and characterization of the fragile X mutation. *Chromosoma* 101, 381–387
- 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
- Verkerk, A.J. *et al.* (1991) Identification of a gene (FMR-1) containing a CGG repeat coincident with a breakpoint cluster region exhibiting length variation in fragile X syndrome. *Cell* 65, 905–914

#### **Costello Syndroom Erasmus MC/ ENCORE publicaties:**

##### Publicaties:

- Giannoulatou, E. *et al.* (2013) Contributions of intrinsic mutation rate and selfish selection to levels of de novo HRAS mutations in the paternal germline. *Proceedings of the National Academy of Sciences* DOI: 10.1073/pnas.1311381110
- Beukers, W. *et al.* (2013) HRAS mutations in bladder cancer at an early age and the possible association with the Costello Syndrome. *Eur J Hum Genet* DOI: 10.1038/ejhg.2013.251
- Krab, L.C. *et al.* (2008) Oncogenes on my mind: ERK and MTOR signaling in cognitive diseases. *Trends Genet* 24, 498–510
- Kushner, S.A. *et al.* (2005) Modulation of presynaptic plasticity and learning by the H-ras/extracellular signal-regulated kinase/synapsin I signaling pathway. *J Neurosci* 25, 9721–9734

#### **Aanleg en Migratie stoornissen Erasmus MC/ ENCORE publicaties:**

- Oegema, R. *et al.* (2013) Novel no-stop FLNA mutation causes multi-organ involvement in males. *Am J Med Genet* 161, 2376–2384
- Oegema, R. *et al.* (2013) A single strand that links multiple neuropathologies in human disease. *Brain* DOI: 10.1093/brain/awt197
- Meuwissen, M.E.C. *et al.* (2013) ACTA2 mutation with childhood cardiovascular, autonomic and brain anomalies and severe outcome. *Am J Med Genet* 161, 1376–1380
- Poulton, C. *et al.* (2013) Progressive cerebellar atrophy and polyneuropathy: expanding the spectrum of PNKP mutations. *Neurogenetics* 14, 43–51
- Verhagen, J.M.A. *et al.* (2012) Phenotypic variability of atypical 22q11.2 deletions not including TBX1. *Am J Med Genet* 158, 2412–2420

- Kheradmand Kia, S. *et al.* (2012) RTTN mutations link primary cilia function to organization of the human cerebral cortex. *Am J Hum Genet* 91, 533–540
- Verbeek, E. *et al.* (2012) COL4A2 mutation associated with familial porencephaly and small-vessel disease. *Eur J Hum Genet* 20, 844–851
- Oegema, R. *et al.* (2012) Asymmetric polymicrogyria and periventricular nodular heterotopia due to mutation in ARX. *Am J Med Genet* 158, 1472–1476
- Meuwissen, M.E.C. and Mancini, G.M.S. (2012) Neurological findings in incontinentia pigmenti; a review. *Eur J Med Genet* 55, 323–331
- Poulton, C.J. *et al.* (2011) Microcephaly with simplified gyration, epilepsy, and infantile diabetes linked to inappropriate apoptosis of neural progenitors. *Am J Hum Genet* 89, 265–276
- de Wit, M.C.Y. *et al.* (2011) Lung disease in FLNA mutation: confirmatory report. *Eur J Med Genet* 54, 299–300
- de Wit, M.-C.Y. *et al.* (2011) Long-term follow-up of type 1 lissencephaly: survival is related to neuroimaging abnormalities. *Developmental Medicine & Child Neurology* 53, 417–421
- Meuwissen, M.E.C. *et al.* (2011) Sporadic COL4A1 mutations with extensive prenatal porencephaly resembling hydranencephaly. *Neurology* 76, 844–846
- de Wit, M.-C.Y. *et al.* (2011) Combined cardiological and neurological abnormalities due to filamin A gene mutation. *Clin Res Cardiol* 100, 45–50
- de Wit, M.C.Y. *et al.* (2010) Absence epilepsy and periventricular nodular heterotopia. *Seizure: the journal of the British Epilepsy Association* 19, 450–452
- Oegema, R. *et al.* (2010) KBG syndrome associated with periventricular nodular heterotopia. *Clin. Dysmorphol.* 19, 164–165
- Verkerk, A.J.M.H. *et al.* (2010) Unbalanced der(5)t(5;20) translocation associated with megalencephaly, perisylvian polymicrogyria, polydactyly and hydrocephalus. *Am J Med Genet* 152, 1488–1497
- de Wit, M.-C.Y. *et al.* (2010) Periventricular nodular heterotopia and distal limb deficiency: a recurrent association. *Am J Med Genet* 152, 954–959
- de Wit, M.C.Y. *et al.* (2009) Movement disorder and neuronal migration disorder due to ARFGEF2 mutation. *Neurogenetics* 10, 333–336
- Verkerk, A.J.M.H. *et al.* (2009) Mutation in the AP4M1 gene provides a model for neuroaxonal injury in cerebral palsy. *Am J Hum Genet* 85, 40–52
- de Wit, M.C.Y. *et al.* (2009) Filamin A mutation, a common cause for periventricular heterotopia, aneurysms and cardiac defects. *J Neurol Neurosurg Psychiatr* 80, 426–428
- Maingay-de Groof, F. *et al.* (2008) Extensive cerebral infarction in the newborn due to incontinentia pigmenti. *Eur J Paediatr Neurol* 12, 284–289
- de Wit, M.C.Y. *et al.* (2008) Cortical brain malformations: effect of clinical, neuroradiological, and modern genetic classification. *Arch Neurol* 65, 358–366
- van Ramshorst, G.H. *et al.* (2006) A case of split notochord syndrome: a child with a neuroenteric fistula presenting with meningitis. *J. Pediatr. Surg.* 41, e19–23
- de Wit, M.C.Y. *et al.* (2006) Microcephaly and simplified gyral pattern of the brain associated with early onset insulin-dependent diabetes mellitus. *Neurogenetics* 7, 259–263
- Breedveld, G. *et al.* (2006) Novel mutations in three families confirm a major role of COL4A1 in hereditary porencephaly. *J Med Genet* 43, 490–495
- de Wit, M.C.Y. *et al.* (2006) Brain abnormalities in a case of malonyl-CoA decarboxylase deficiency. *Mol. Genet. Metab.* 87, 102–106
- Brooks, A.S. *et al.* (2005) Homozygous nonsense mutations in KIAA1279 are associated with malformations of the central and enteric nervous systems. *Am J Hum Genet* 77, 120–126
- Wessels, M.W. *et al.* (2005) Autosomal dominant inheritance of left ventricular outflow tract obstruction. *Am. J. Med. Genet. A* 134, 171–179
- Mancini, G.M.S. *et al.* (2005) Two novel mutations in SLC6A8 cause creatine transporter defect and distinctive X-linked mental retardation in two unrelated Dutch families. *Am. J. Med. Genet. A* 132, 288–295
- Wessels, M.W. *et al.* (2004) Three new families with arterial tortuosity syndrome. *Am. J. Med. Genet. A* 131, 134–143
- Kleijer, W.J. *et al.* (2004) [From gene to disease; Krabbe disease and galactosylceramidase deficiency]. *Ned Tijdschr Geneesk* 148, 826–828
- Mancini, G.M.S. *et al.* (2004) Hereditary porencephaly: clinical and MRI findings in two Dutch families. *Eur J Paediatr Neurol* 8, 45–54

### **Autisme Erasmus MC publicaties:**

- Eussen, M.L. *et al.* (2013) The association of quality of social relations, symptom severity and intelligence with anxiety in children with autism spectrum disorders. *Autism* 17, 723–735
- 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
- So, P. *et al.* (2013) Using the Child Behavior Checklist and the Teacher's Report Form for identification of children with autism spectrum disorders. *Autism* 17, 595–607
- Román, G.C. *et al.* (2013) Association of gestational maternal hypothyroxinemia and increased autism risk. *Ann Neurol* DOI: 10.1002/ana.23976
- Greaves-Lord, K. *et al.* (2013) Empirically based phenotypic profiles of children with pervasive developmental disorders: interpretation in the light of the DSM-5. *Journal of autism and developmental disorders* 43, 1784–1797
- White, T. *et al.* (2013) Pediatric population-based neuroimaging and the Generation R Study: the intersection of developmental neuroscience and epidemiology. *Eur. J. Epidemiol.* 28, 99–111
- Jaspers, M. *et al.* (2013) Early childhood assessments of community pediatric professionals predict autism spectrum and attention deficit hyperactivity problems. *J Abnorm Child Psychol* 41, 71–80
- Baudouin, S.J. *et al.* (2012) Shared synaptic pathophysiology in syndromic and nonsyndromic rodent models of autism. *Science* 338, 128–132
- van der Vlugt, J.J.B. *et al.* (2012) Cognitive and behavioral functioning in 82 patients with trigonocephaly. *Plast. Reconstr. Surg.* 130, 885–893
- Hermans, H. *et al.* (2012) Feasibility, reliability and validity of the Dutch translation of the Anxiety, Depression And Mood Scale in older adults with intellectual disabilities. *Res Dev Disabil* 33, 315–323
- Vuijk, R. *et al.* (2012) [Personality traits in adults with autism spectrum disorders measured by means of the Temperament and Character Inventory]. *Tijdschr Psychiatr* 54, 699–707
- Nijmeijer, J.S. *et al.* (2010) Perinatal risk factors interacting with catechol O-methyltransferase and the serotonin transporter gene predict ASD symptoms in children with ADHD. *J Child Psychol Psychiatry* 51, 1242–1250
- Tiemeier, H. *et al.* (2010) Cerebellum development during childhood and adolescence: a longitudinal morphometric MRI study. *Neuroimage* 49, 63–70
- de Bruin, E.I. *et al.* (2009) Autistic features in girls from a psychiatric sample are strongly associated with a low 2D:4D ratio. *Autism* 13, 511–521
- Herba, C.M. *et al.* (2008) Face and emotion recognition in MCDD versus PDD-NOS. *Journal of autism and developmental disorders* 38, 706–718
- de Bruin, E.I. *et al.* (2007) Multiple complex developmental disorder delineated from PDD-NOS. *Journal of autism and developmental disorders* 37, 1181–1191
- de Bruin, E.I. *et al.* (2007) High rates of psychiatric co-morbidity in PDD-NOS. *Journal of autism and developmental disorders* 37, 877–886
- de Bruin, E.I. *et al.* (2006) Differences in finger length ratio between males with autism, pervasive developmental disorder-not otherwise specified, ADHD, and anxiety disorders. *Developmental Medicine & Child Neurology* 48, 962–965
- de Bruin, E.I. *et al.* (2006) WISC-R subtest but no overall VIQ-PIQ difference in Dutch children with PDD-NOS. *J Abnorm Child Psychol* 34, 263–271
- Dekker, M.C. *et al.* (2002) Assessing emotional and behavioral problems in children with intellectual disability: revisiting the factor structure of the developmental behavior checklist. *Journal of autism and developmental disorders* 32, 601–610
- van der Geest, J.N. *et al.* (2002) Gaze behavior of children with pervasive developmental disorder toward human faces: a fixation time study. *J Child Psychol Psychiatry* 43, 669–678
- van der Geest, J.N. *et al.* (2002) Looking at images with human figures: comparison between autistic and normal children. *Journal of autism and developmental disorders* 32, 69–75