



CURRICULUM VITAE

Name Vincent Thomas Ramaekers
Nationality Dutch
Date of birth March 16 1954

Married to : Godelieve Vos (Belgian)
Born on January 7th 1958 in Kalenda - Belgian Congo

Children Lara, born november 3rd 1983
Valérie, born September 8th 1987

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TRAINING

1966-1972 Gymnasium-beta
Henric van Veldeke College
Maastricht
The Netherlands

1972-1975 Medical School
Free University Brussels
Belgium

1975-1979 Medical School
State University of Utrecht
The Netherlands

November 1979 Degree of Doctor in Medicine

1976 Training in Pathology
Hospital St-Annadal, Maastricht
(Prof Dr V Swaen)

1980-September 1981 Registrar in Paediatrics
Wilhelmina Children's Hospital Utrecht
The Netherlands

Sept 1981-March 1982 Registrar in Paediatric Neurology
University Hospital Utrecht
The Netherlands
(Prof Dr J Willemse)

March 1982-Mai 1984 Registrar in Paediatrics
Emma Children's Hospital of the University of Amsterdam

	The Netherlands (Prof Dr PA Voûte)
Mai 1984	Degree of Paediatrician
October 1984-October 1985	Training in Paediatric Neurology Hospital for Sick Children Great Ormond Street, London (Dr EM Brett and Dr J Wilson) Stipendium by the Dutch Organization for Pure Scientific Research (ZWO) Certificate clinical neurophysiology for EEG and evoked potential recordings (Dr S Boyd)
October 1985-February 1988	Fellow in Paediatric Neurology, Neonatal Neurology and Research Fellow at the Paediatric Clinic of the University of Leuven, Belgium (Prof Dr P Casaer, Prof Dr J Jaeken and Prof Dr E Eggermont) Stipendium of the Ter Meulen Fund and Janssen Research and Development
February 1988-October 2001	Paediatric Neurologist and Oberarzt University Hospital Aachen
November 2001-October 2007	Head of the Division of Neuropaediatrics University Hospital Aachen
October 2007	Nomination Professor in Paediatrics and Child Neurology Aachen Faculty of Medicine
Current Position	Professor in Paediatrics and Child Neurology Division of Neuropaediatrics Head of Centre de Référence d'Autisme Liège (CRAL). Centre Hospitalier Universitaire Liège; Belgium

THESIS

29 Mai 1986	University of Amsterdam
Title	Selective Decontamination of the Digestive Tract in Paediatric Oncology
Promotores	Prof Dr PA Voûte Prof Dr J van der Noordaa

HABILITATION

May 2002	University of Aachen, Faculty of Medicine Cerebellar structural Abnormalities in Childhood : Significance of antioxidative defences.
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Promotores

Prof Dr G Heimann
Prof Dr J Noth
Prof Dr PG Barth

PRICES

Nestlé Research first Prize 2004.
Cerebral Folate Deficiency – An autoimmune disorder.
Swiss Pediatric Research Meeting, November 18th, 2004
University Childrens Hospital, Inselspital Bern.

MEMBERSHIPS

International Child Neurology Association
Belgian Society of Paediatric Neurology
European Paediatric Neurology Society
Gesellschaft Neuropädiatrie
Dutch Society of Paediatrics
Belgian Society of Paediatrics
Society for the Study of Inborn Errors of Metabolism (SSIEM)

NOMINATIONS

Top-physician in Paediatrics of the German-speaking countries (Austria, Germany and Switzerland).
See also : <http://www.die-besten-nennen.de>
Nomination by the Aerztekammer Nordrhein on the examining-board for pediatric neurologists
Research funding 2010-2011 by Fonds National de Recherches Scientifiques

RESEARCH TOPICS AND MAIN CLINICAL ACTIVITIES (in chronologic order)

I. Inborn Errors of Metabolism

- I.1. ***Hereditary disorders in the metabolism of Glycoproteins*** and the recently discovered diseases known as Carbohydrate-Deficient-Glycoproteins (CDG) syndromes.
First discovery of the CDG type II syndrome in an Iranian family. Cooperation with Prof Dr J Jaeken, University of Leuven.
- I.2. ***Disturbances in the metabolism of Biotin*** and first discovery of a new biochemical variant of the so-called late-onset Biotinidase Km Variant.
Cooperation with the Enzym Labor of the Children's Hospital Basel and Prof B Wolf of the University of Virginia (USA).
- I.3. ***Disturbances in the metabolism of the biogenic monoamines, pterins and folates.***
New discovery of a synthetic error of serotonin by tryptophane hydroxylase deficiency.
New discovery of folate transport disorder across the blood-brain barrier.
Cooperation with Prof Dr N Blau, Laboratory of Chemistry and Biochemistry, Children's Hospital Zürich, Switzerland.
Cooperation with Dr Baader, Max Delbrück Center, Berlin ; Germany.
Cooperation with Prof E Quadros, Department of Biochemistry, State University New York, Brooklyn; USA.

- I.4. ***Refractory Epilepsy and disturbed metabolism of selenium*** with the study of the effect of additional supplementation with selenium or selenocysteine.
Cooperation with the Department of Pharmaceutical Microbiology, University of Antwerp (Prof Dr D van den Berghe) and Laboratory of Umweltmedizin, University of Aachen (Prof Dr Dott)
- I.5. ***Cerebellar structural abnormalities in Childhood and the Significance of oxidative/nitrosative stress.***
Joint study between Dept Paediatrics, Division of Child Neurology and the Institute of Biochemistry (Dr G Steffens) and the University of Amsterdam, Division of Child Neurology (Prof Dr PG Barth), Laboratory of Genetic Metabolic Diseases (Prof Dr R Wanders, Dr A van Gennip and Dr N Abeling) and the Department of Pharmaceutical Microbiology, University of Antwerp (Prof Dr D van den Berghe).
- I.6. ***Cerebral Folate Deficiency Syndrome in Childhood and Adolescence***
Discovery of new syndromes characterized by low methylfolate values in the brain and characterization of the aetiologies, development of diagnostic procedures and treatment.
Cooperation with the following institutions :
Prof Dr Nenad Blau, Division of Clinical Chemistry and Biochemistry, University Children's Hospital Zürich ; Switzerland.
Prof Dr E Quadros and Dr J Sequeira, State University New York ; USA
- II. **Surface EMG Studies in Childhood**
Cooperation with the Helmholtz Institute
- III. **Doppler studies of cerebral blood flow in preterm and term infants**
- IV. **Genetic Studies of Manganese Superoxide Dismutase in children with lowered MnSOD values.**
Cooperation with Prof VandenBerghe, University Antwerp and Dr R Pfäffle, Paediatric Endocrinology.
Cooperation with Dr S Kuery and Prof S Beziau (dept Genetics University Nantes)

CURRENT RESEARCH PROGRAM (Professor Dr V Ramaekers)

1. Research-fellow sponsored by the Medical Faculty Aachen is Dr. Thomas Opladen who worked for a period of two years (2004 to 2006) together with Prof. Dr. N. Blau on a joint project for Cerebral Folate Deficiencies. The protocol for this basic scientific studies has been written by Dr Ramaekers and Prof Dr Blau.
2. Christina Peters (Doctors's Thesis) has been working at the Department of Biochemistry (Dr. Guy Steffens) performing basic research on markers for nitrosative stress (3-nitrotyrosine) in cerebrospinal fluid. Her thesis has been completed this year.
3. Joint Research project on Cerebral Folate Deficiencies in Childhood is now in progress together with departments of child neurology and laboratories Liege, Amsterdam, Leuven, UCL, Zürich, New York, Barcelona, Porto and Paris. These new projects will be able to recruit more patients from Europe and the US for further studies.
These studies have been approved last year by the Aachen Ethics Committee and by the Ethics Committee of the University of Liege.

ORIGINAL CONTRIBUTIONS

1. Ramaekers VT, Lake BD, Harding B, Boyd S, Harden A, Brett EM, Wilson J.
Diagnostic difficulties in infantile neuroaxonal dystrophy. A clinicopathological study of eight cases.
Neuropediatrics. 1987 Aug;18(3):170-5.
2. Ramaekers VT, Casaer P.
Influence of behavioral states on cerebral blood flow velocity patterns in preterm infants.
J Pediatr. 1987 Nov;111(5):795.
3. Daniels H, Devlieger H, Casaer P, Ramaekers V, van den Broeck J, Eggermont E.
Feeding, behavioural state and cardiorespiratory control.
Acta Paediatr Scand. 1988 May;77(3):369-73.
4. Ramaekers VT, Casaer P, Marchal G, Smet M, Goossens W.
The effect of blood transfusion on cerebral blood-flow in preterm infants: a Doppler study.
Dev Med Child Neurol. 1988 Jun;30(3):334-41.
5. Ramaekers VT, Casaer P, Daniels H, Smet M, Marchal G.
The influence of behavioural states on cerebral blood flow velocity patterns in stable preterm infants.
Early Hum Dev. 1989 Dec;20(3-4):229-46.
6. Ramaekers VT, Casaer P.
Defective regulation of cerebral oxygen transport after severe birth asphyxia.
Dev Med Child Neurol. 1990 Jan;32(1):56-62.
7. Ramaekers VT, Casaer P, Daniels H, Marchal G.
Upper limits of brain blood flow autoregulation in stable infants of various conceptional age.
Early Hum Dev. 1990 Dec;24(3):249-58.
8. Ramaekers VT, Stibler H, Kint J, Jaeken J.
A new variant of the carbohydrate deficient glycoproteins syndrome.
J Inher Metab Dis. 1991;14(3):385-8.

9. Ramaekers VT, Suormala TM, Brab M, Duran R, Heimann G, Baumgartner ER. A biotinidase Km variant causing late onset bilateral optic neuropathy. Arch Dis Child. 1992 Jan;67(1):115-9.
10. Brab M, Ramaekers VT, Baumgartner ER, Heimann G, Reim M. Juvenile optic neuropathy caused by Km variants of biotinidase. Klin Monatsbl Augenheilkd. 1992 Mar;200(3):204-9.
11. Ramaekers VT, Daniels H, Casaer P. Brain oxygen transport related to levels of fetal haemoglobin in stable preterm infants. J Dev Physiol. 1992 May;17(5):209-13.
12. Ramaekers VT, Casaer P, Daniels H, Marchal G. The influence of blood transfusion on brain blood flow autoregulation among stable preterm infants. Early Hum Dev. 1992 Oct;30(3):211-20.
13. Otterbach B, Stoffel W, Ramaekers V. A novel mutation in the proteolipid protein gene leading to Pelizaeus-Merzbacher disease. Biol Chem Hoppe Seyler. 1993 Jan;374(1):75-83.
14. Jaeken J, De Cock P, Stibler H, Van Geet C, Kint J, Ramaekers V, Carchon H. Carbohydrate-deficient glycoprotein syndrome type II. J Inher Metab Dis. 1993;16(6):1041.
15. Ramaekers VT, Brab M, Rau G, Heimann G. Recovery from neurological deficits following biotin treatment in a Biotinidase Km variant. Neuropediatrics. 1993 Apr;24(2):98-102.
16. Hageman G, Ramaekers VT, Hilhorst BG, Rozeboom AR. Congenital cervical spinal muscular atrophy: a non-familial, non-progressive condition of the upper limbs. J Neurol Neurosurg Psychiatry. 1993 Apr;56(4):365-8.
17. Ramaekers VT, Disselhorst-Klug C, Schneider J, Silny J, Forst J, Forst R, Kotlarek F, Rau G. Clinical application of a noninvasive multi-electrode array EMG for the recording of single motor unit activity. Neuropediatrics. 1993 Jun;24(3):134-8.
18. Reul J, Ramaekers V, Thron A. Suprasellar space-occupying lesion as initial manifestation of tuberculosis in childhood. Dtsch Med Wochenschr. 1993 Jun 4;118(22):820-4.
19. Ramaekers VT, Casaer P, Daniels H. Cerebral hyperperfusion following episodes of bradycardia in the preterm infant. Early Hum Dev. 1993 Oct;34(3):199-208.

20. Schroder JM, Heide G, Ramaekers V, Mortier W.
Subtotal aplasia of myelinated nerve fibers in the sural nerve.
Neuropediatrics. 1993 Oct;24(5):286-91.
21. Ramaekers VT, Calomme M, Vanden Berghe D, Makropoulos W.
Selenium deficiency triggering intractable seizures.
Neuropediatrics. 1994 Aug;25(4):217-23.
22. Ramaekers VT, Reul J, Siller V, Thron A.
Mesencephalic and third ventricle cysts: diagnosis and management in four cases.
J Neurol Neurosurg Psychiatry. 1994 Oct;57(10):1216-20.
23. Suormala T, Ramaekers VT, Schweitzer S, Fowler B, Laub MC, Schwermer C, Bachmann J, Baumgartner ER.
Biotinidase Km-variants: detection and detailed biochemical investigations.
J Inherit Metab Dis. 1995;18(6):689-700.
24. Lissens W, De Meirleir L, Seneca S, Benelli C, Marsac C, Poll-The BT, Briones P, Ruitenbeek W, van Diggelen O, Chaigne D, Ramaekers V, Liebaers I.
Mutation analysis of the pyruvate dehydrogenase E1 alpha gene in eight Patients with a pyruvate dehydrogenase complex deficiency.
Hum Mutat. 1996;7(1):46-51.
25. Weis J, Reul J, Mayfrank L, Ramaekers V, Thron A.
Duplication of a vertebral artery associated with epidermoid cyst of the posterior fossa.
Eur Radiol. 1997;7(3):412-4.
26. Ramaekers VT, Reul J, Kusenbach G, Thron A, Heimann G.
Central pontine myelinolysis associated with acquired folate depletion.
Neuropediatrics. 1997 Apr;28(2):126-30.
27. Ramaekers VT, Heimann G, Reul J, Thron A, Jaeken J.
Genetic disorders and cerebellar structural abnormalities in childhood.
Brain. 1997 Oct;120 (Pt 10):1739-51.
28. Ramaekers VT, Bosman B, Jansen GA, Wanders RJ.
Increased plasma malondialdehyde associated with cerebellar structural defects.
Arch Dis Child. 1997 Sep;77(3):231-4.
29. Wolf B, Pomponio RJ, Norrgard KJ, Lott IT, Baumgartner ER, Suormala T, Ramaekers VT, Coskun T, Tokatli A, Ozalp I, Hymes J.
Delayed-onset profound biotinidase deficiency.
J Pediatr. 1998 Feb;132(2):362-5.
30. Senderek J, Bergmann C, Quasthoff S, Ramaekers VT, Schroder JM.
X-linked dominant Charcot-Marie-Tooth disease: nerve biopsies allow morphological evaluation and detection of connexin32 mutations (Arg15Trp,Arg22Gln).
Acta Neuropathol (Berl). 1998 May;95(5):443-9.
31. Hausler MG, Ramaekers VT, Reul J, Meilicke R, Heimann G.

- Early and late onset manifestations of cerebral vasculitis related to Varicella zoster.
Neuropediatrics. 1998 Aug;29(4):202-7.
32. Rohde V, Mayfrank L, Ramaekers VT, Gilsbach JM.
Four-year experience with the routine use of the programmable Hakim valve in the management of children with hydrocephalus.
Acta Neurochir (Wien) 1998; 140 (11): 1127-34.
 33. Kuker W, Ramaekers V.
Persistent hyperplastic primary vitreous: MRI.
Neuroradiology. 1999 Jul;41(7):520-2.
 34. Senderek J, Hermanns B, Bergmann C, Boroojerdi B, Bajbouj M, Hungs M, Ramaekers VT, Quasthoff S, Karch D, Schroder JM.
X-linked dominant Charcot-Marie-Tooth neuropathy: clinical, electrophysiological, and morphological phenotype in four families with different connexin32 mutations(1).
J Neurol Sci. 1999 Aug 15;167(2):90-101.
 35. Abicht A, Stucka R, Karcagi V, Herczegfalvi A, Horvath R, Mortier W, Schara U, Ramaekers V, Jost W, Brunner J, Janssen G, Seidel U, Schlotter B, Muller-Felber W, Pongratz D, Rudel R, Lochmuller H.
A common mutation (epsilon1267delG) in congenital myasthenic patients of Gypsy ethnic origin.
Neurology. 1999 Oct 22;53(7):1564-9.
 36. Disselhorst-Klug C, Bahm J, Ramaekers V, Trachtena A, Rau G.
Non-invasive approach of motor unit recording during muscle contractions in humans.
Eur J Appl Physiol. 2000 Oct;83(2-3):144-50. Review.
 37. Kolker S, Ramaekers VT, Zschocke J, Hoffmann GF.
Acute encephalopathy despite early therapy in a patient with homozygosity for E365K in the glutaryl-coenzyme A dehydrogenase gene.
J Pediatr. 2001 Feb;138(2):277-9.
 38. Ramaekers VT, Senderek J, Hausler M, Haring M, Abeling N, Zerres K, Bergmann C, Heimann G, Blau N.
A novel neurodevelopmental syndrome responsive to 5-hydroxytryptophan and carbidopa.
Mol Genet Metab. 2001 Jun;73(2):179-87.
 39. Muller HD, Mugler M, Ramaekers VT, Schroder JM.
Hereditary motor and sensory neuropathy with absence of large myelinated fibers due to absence of large neurons in dorsal root ganglia and anterior horns, clinically associated with deafness, mental retardation, and epilepsy (HMSN-ADM).
J Peripher Nerv Syst. 2000 Sep;5(3):147-57.
 40. Hausler M, Jaeken J, Monch E, Ramaekers VT.
Phenotypic heterogeneity and adverse effects of serine treatment in 3-phosphoglycerate dehydrogenase deficiency: report of two siblings.
Neuropediatrics 2001 Aug; 32 (4): 191-5.

41. Hausler M, Schaade L, Kemeny S, Schweizer K, Schoenmackers C, Ramaekers VT. Encephalitis related to primary varicella-zoster virus infection in immunocompetent children. *J Neurol Sci.* 2002 Mar 30; 195 (2): 111-6.
42. Senderek J, Ramaekers VT, Zerres K, Rudnik-Schoneborn S, Schroder JM, Bergmann C. Phenotypic variation of a novel nonsense mutation in the P0 intracellular domain. *J Neurol Sci.* 2001 Nov 15; 192 (1-2): 49-51.
43. Schefels J, Wenzl TG, Merz U, Ramaekers VT, Holzki J, Rudnik-schoneborn S, Hermanns B, Hornchen H. Functional upper airway obstruction in a child with Freeman-Sheldon syndrome. *ORL J Otorhinolaryngol Relat Spec* 2002 Jan-Feb; 64 (1): 53-6.
44. Hausler M, Ramaekers VT, Doenges M, Schweizer K, Ritter K, Schaade L. Neurological complications of acute and persistent Epstein-Barr virus infection in paediatric patients. *J Med Virol* 2002; 68: 253-63.
45. Senderek J, Bergmann C, Ramaekers VT, Nelis E, Bernert G, Makowski A, Zuchner S, De Jonghe P, Rudnik-Schöneborn S, Zerres K, Schroder JM. Mutations in the ganglioside-induced differentiation-associated protein-1 (GDAP1) gene in intermediate type autosomal recessive Charcot-Marie-Tooth neuropathy. *Brain* 2003; 126 (pt 3): 642-9.
46. Ramaekers, Häusler M, Opladen T, Heimann G, Blau N. Psychomotor Retardation, Spastic Paraplegia, Cerebellar Ataxia and Dyskinesia Associated with low 5-Methyltetrahydrofolate in Cerebrospinal Fluid: A Novel Neurometabolic Condition Responding to Folinic Acid Substitution. *Neuropediatrics* 2002; 33 (6): 301-8.
47. Häusler M, Sellhaus B, Schweizer K, Ramaekers VT, Opladen T, Kleines M. Flow cytometric cerebrospinal fluid analysis in children. *Pathol Res Pract.* 2003;199(10):667-75.
48. Häusler M, Schaade L, Ramaekers VT, Doenges M, Heimann G and Selhaus B. Inflammatory pseudotumors of the Central Nervous System: Report of 3 Cases and a Literature Review. *Hum Pathol* 2003; 34: 253-262.
49. Blau N, Bonafé L, Krägeloh-Mann I, Thöny B, Kierat L, Häusler M, Ramaekers V. Cerebrospinal fluid Pterins and Folates in Aicardi-Goutières Syndrome: A new Phenotype. *Neurology* 2003; 61:642-647.
50. Ramaekers VT, Hansen SI, Holm J, Opladen Th, Senderek J, Häusler M, Heimann G, Fowler B, Maiwald R, Blau N. Reduced Folate transport to the CNS in female Rett Patients. *Neurology* 2003; 61:506-514.
51. Bergmann C, Zerres K, Senderek J, Rudnik-Schöneborn S, Eggermann Th, Häusler M, Mull M, Ramaekers VT. Oligophrenin 1 (OPHN1) gene mutation causes syndromic X-linked mental retardation with myoclonic epilepsy, ataxia, rostral ventricular enlargement and cerebellar hypoplasia. *Brain* 2003; 126:1537-44.

52. Korinth MC, Ramaekers V T, Rhode V.
Cervical cord exostosis compressing the axis in a boy with multiple exostoses. Case illustration.
J Neurosurg Spine 2004; 100: 223.
53. Hausler M, Merz U, Van Tuil C, Ramaekers VT.
Long-term outcome after neonatal parenchymatous brain lesions.
Klin Padiatr. 2004 Jul-Aug;216(4):244-51.
54. Ramaekers VT, Blau N.
Cerebral Folate Deficiency. Annotation.
Dev Med Child Neurol 2004; 46:843-51.
55. Hausler M, Anhuf D, Schuler H, Ramaekers VT, Thron A, Zerres K, Moller-Hartmann W.
White-matter disease in 18q deletion (18q-) syndrome: magnetic resonance spectroscopy indicates demyelination or increased myelin turnover rather than dysmyelination. Neuroradiology 2005;47(1):83-6.
56. Ramaekers VT, Rothenberg SP, Sequeira JM, Opladen T, Blau N, Quadros EV, Selhub J.
Autoantibodies against Folate Receptors Are Associated with the Infantile-onset Cerebral Folate Deficiency Syndrome. New England Journal of Medicine 2005; 352:1985-91.
57. Opladen T, Ramaekers VT, Heimann G, Blau N
Analysis of 5-methyltetrahydrofolate in serum of healthy children.
Mol Genet Metab 2006; 87(1):61-5.
58. Ramaekers VT, Artuch R, Temudo T, Campistol J, Pineda M, Roelens F, Laccone F, Blau N, Sequeira JM and Quadros E.
Folate Receptor Autoantibodies Can Block 5-Methyltetrahydrofolate Transport into CSF of Rett Patients. Neuropediatrics 2007; 38(4):179-83.
59. Ramaekers VT, Blau N, Sequeira J, Quadros EV. Mitochondrial Complex I Encephalomyopathy and Cerebral 5-methyltetrahydrofolate Deficiency. Neuropediatrics 2007;38(4):184-7.
60. Ramaekers VT, Blau N, Sequeira JM, Nassogne MC, Quadros EV. Folate receptor autoimmunity and cerebral folate deficiency in low-functioning autism with neurological deficits. Neuropediatrics. 2007;38(6):276-81.
61. Garcia-Cazorla A, Quadros EV, Nascimento A, Garcia-Silva MT, Briones P, Montoya J, Ormazabal A, Artuch R, Sequeira JM, Blau N, Arenas J, Pineda M, Ramaekers VT. Mitochondrial diseases associated with cerebral folate deficiency. Neurology. 2008;70(16):1360-2.
62. Ramaekers VT, Sequeira JM, Blau N, Quadros EV. A milk-free diet downregulates folate

receptor autoimmunity in cerebral folate deficiency syndrome. *Dev Med Child Neurol.* 2008;50(5):346-52.

63. Bonkowsky JL, Ramaekers VT, Quadros EV, Lloyd M. Progressive encephalopathy in a child with cerebral folate deficiency syndrome. *J Child Neurol.* 2008;23(12):1460-3.
64. Scholl UI, Choi M, Liu T, Ramaekers VT, Häusler MG, Grimmer J, Tobe SW, Farhi A, Nelson-Williams C, Lifton RP. Seizures, sensorineural deafness, ataxia, mental retardation, and electrolyte imbalance (SeSAME syndrome) caused by mutations in *KCNJ10*. *Proc Natl Acad Sci U S A.* 2009 7;106(14):5842-7.
65. Hasselmann O, Blau N, Ramaekers VT, Quadros EV, Sequeira JM, Weissert M. Cerebral folate deficiency and CNS inflammatory markers in Alpers disease. *Mol Genet Metab.* 2010 Jan;99(1):58-61.
66. Opladen T, Blau N, Ramaekers VT. Effect of antiepileptic drugs and reactive oxygen species on folate receptor 1 (FOLR1)-dependent 5-methyltetrahydrofolate transport. *Mol Genet Metab.* 2010 Sep;101(1):48-54.
67. Adamsen D, Meili D, Blau N, Thöny B, Ramaekers V. Autism associated with low 5-hydroxyindolacetic acid in CSF and the heterozygous *SLC6A4* gene Gly56Ala plus 5-HTTLPR L/L promoter variants. *Mol Genet Metab.* 2011 Mar;102(3):368-73.
68. Küry S, Ramaekers V, Bézieau S, Wolf B. Clinical utility gene card for: Biotinidase deficiency. *Eur J Hum Genet.* 2012 Feb 29.
69. Ramaekers V, Sequeira JM, Quadros EV. Role of Folate Receptor Autoantibodies in infantile Autism. *Molecular Psychiatry* 2012. In Press
70. Ramaekers VT, Quadros EV, Sequeira JM. Role of folate receptor autoantibodies in infantile autism. *Mol Psychiatry.* 2013 Mar;18(3):270-1.
71. Ramaekers V, Sequeira JM, Quadros EV. Clinical recognition and aspects of the cerebral folate deficiency syndromes. *Clin Chem Lab Med.* 2013;51(3):497-511.
72. Sequeira JM, Ramaekers VT, Quadros EV. The diagnostic utility of folate receptor autoantibodies in blood. *Clin Chem Lab Med.* 2013; 51(3):545-54.
73. Adamsen D, Ramaekers V, Ho HTB, Britschgi C, Rüfenacht V, Meili D, Bobrowski E, Philippe P, Nava C, Van Maldergem L, Bruggmann R, Walitza S, Wang J, Grünblatt E, Thöny B. Autism spectrum disorder associated with low serotonin in CSF and mutations in the *SLC29A4* plasma membrane monoamine transporter (PMAT) gene. *Molecular Autism* 2014 (open access Journal).
74. Ramaekers VT, Thöny B, Sequeira JM, Anseau M, Philippe P, Boemer F, Bours V, Quadros EV. Folinic acid treatment for schizophrenia associated with folate receptor autoantibodies. *Mol Genet Metab.* 2014;113(4):307-14.
75. Farhat N, Desprechins B, Otto B, Ramaekers V, Seghaye MC. Paraspinal Arterio-Venous Fistula in Children: Two More Cases of an Exceptional

Malformation. Clin Pract. 2015 Apr 24;5(2):707.

76. Küry S, Ramaekers V, Bézieau S, Wolf B. Clinical utility gene card for: Biotinidase deficiency-update 2015. Eur J Hum Genet. 2015 Nov 18
77. Boycott KM, Beaulieu CL, Kernohan KD, Gebril OH, Mhanni A, Chudley AE, Redl D, Qin W, Hampson S, Küry S, Tetreault M, Puffenberger EG, Scott JN, Bezieau S, Reis A, Uebe S, Schumacher J, Hegele RA, McLeod DR, Gálvez-Peralta M, Majewski J, Ramaekers VT; Care4Rare Canada Consortium, Nebert DW, Innes AM, Parboosingh JS, Abou Jamra R. Autosomal-Recessive Intellectual Disability with Cerebellar Atrophy Syndrome Caused by Mutation of the Manganese and Zinc Transporter Gene SLC39A8. Am J Hum Genet. 2015;3;97(6):886-93.

BOOK CONTRIBUTIONS

1. V Th Ramaekers, MC Ansink-Schipper, CDM Dobbelaar, J de Kraker, J van der Noordaa, WJ Terpstra, PA Voûte.
Selective decontamination of the Digestive Tract during granulocytopenia as an infection preventive method in paediatric oncology.
In: Third International Symposium on Infections in the Immunocompromised Host, York University Toronto Canada 1984; 124.
2. V Th Ramaekers, CDM Dobbelaar, G van Doornum, P van Keulen, J de Kraker, J van der Noordaa, PA Voûte, P Wertheim.
Viral Infections and cytostatic treatment in paediatric oncology.
In: Third International Symposium on Infections in the Immunocompromised Host, York University Toronto Canada 1984; 125.
3. F Kotlarek, U Rubenstrunk, V Th Ramaekers, G Gross-Selbeck, K Kellermann, K Rheingans, W Mortier, R Pothman, H von Bernuth, U Schauseil-Zipf.
A study of valproate in infants with West syndrome.
Fourth International Symposium on sodium valproate and epilepsy. David Chadwick (Editor), 1989; 87-90.
4. V Th Ramaekers, H Daniels, P Casaer, H Devlieger, E Eggermont, M Smet, G Marchal.
Fluctuerende patronen van de hersendoorbloeding bij de pasgeborene.
Achtste Congres Nederlandse Vereniging voor Kindergeneeskunde. 5-7 November 1986.
5. V Th Ramaekers, BD Lake, B Harding, S Boyd, J Wilson, EM Brett.
Problemen in de diagnostiek van Infantiele Neuroaxonale Dystrophie.
Achtste Congres Nederlandse Vereniging voor Kindergeneeskunde. 5-7 November 1986.
6. V Th Ramaekers, P Casaer, G Marchal.
Reductie van de cerebrale perfusie na bloedtransfusie bij de pasgeborene.
Negende Congres Nederlandse Vereniging voor Kindergeneeskunde. 4-6 November 1987.
7. V Th Ramaekers, P Casaer, H Daniels, G Marchal.
De bovenste limiet der autoregulatie bij pasgeborenen van verschillende postmenstruele leeftijd.
Negende Congres Nederlandse Vereniging voor Kindergeneeskunde. 4-6 November 1987.

8. V Th Ramaekers, P Casaer, H Daniels, G mmarchal.
Verstoorde regulatie van cerebraal zuurstoftransport bij ernstig post-asphyxiale encephalopathie.
Tiende Congres Nederlandse Vereniging voor Kindergeneeskunde. 2-4 november 1988.
9. V Th Ramaekers, TM Suormala, M Brab, ER Baumgartner.
A biotinidase Km variant causing acute onset optic neuropathy followed by spastic paraparesis and motor neuropathy.
Presented at the Vth International Congress of Inborn Errors of Metabolism 1990, Pacific Grove, June 1-5; California USA.
10. V Th Ramaekers, H Stibler, J Kint and J Jaeken.
A new variant of the Carbohydrate Deficient Glycoproteins Syndrome.
Proceedings of the Society for the Study of Inborn Errors of Metabolism; Birmingham 1990.
11. V Th Ramaekers, F Kotlarek, J Schneider, J Silny, G Rau.
Nichtinvasive Elektromyographie motorischer Einheiten. Erste klinische Erfahrungen im Kindesalter.
35 Jahrestagung der Deutsche EEG-Gesellschaft, 18-21 Oktober 1990, Bonn.
12. V Th Ramaekers, J Schneider, C Disselhorst-Klug, J Silny, G Rau, R Forst, F Kotlarek.
Nichtinvasive Elektromyographie motorischer Einheiten. Vorstellung einer neuen Methodik und erste klinische Erfahrungen im Kindesalter.
Neuropaediatric Kongress Basel 1990; published in " Aktuelle Neuropädiatrie 1990 " (edited by Springer Verlag).
13. V Th Ramaekers, B Bosman, J Jaeken.
Genetic diseases associated with cerebellar abnormalities on CT scan.
Proceedings of the Society for the Study of Inborn Errors of Metabolism; Leuven 1992.
Leuven 30th Annual Symposium, 8-11th September 1992.
14. Th Peschgens, R Mertens, V Th Ramaekers, G Heimann.
Infantiles Myoklonie-Opsoklonus-Syndrom (Kinsbourne-Syndrome) als fakultatives Symptom des Neuroblastoms- eine Fallvorstellung. In: Aktuelle Neuropädiatrie 1993 (Eds.: H Todt, D Heinicke).
15. V Th Ramaekers, J Jaeken, G Heimann.
Carbohydrate-Deficient-Glycoproteins Syndrom Typ II.
In: Aktuelle Neuropädiatrie 1994 (Eds.: D Rating).
16. V Th Ramaekers, W Makropoulos.
Seleniummangel und therapieresistente Epilepsie.
In: Aktuelle Neuropädiatrie 1994 (Eds.: D Rating).
17. M Dreuw, G Kusenbach, V Th Ramaekers, G Heimann.
Ernährungsbedingtes Vitamin B12-Mangel-Syndrom mit schweren neurologischen Symptomen.
In: Aktuelle Neuropädiatrie 1994 (Eds.: D Rating).
18. T Peschgens, V Th Ramaekers, U Merz, O Blankenstein, W Dott, H Hörnchen.
Selen-Status und Selen-Substitution bei Frühgeborenen < 1500 g.
21. Symposium des Deutsch Osterreichischen Gesellschaft für Neonatologie und Pädiatrische Intensivmedizin.
(26-28.10.1995 in Mannheim) Kongressband, Alete Wissenschaftlicher Dienst, ISBN: 3-924057-87-

7, S. 273-274.

19. V Th Ramaekers, J Reul, M Häusler, G Heimann.
Varicella-Zoster assoziierte cerebrale Vaskulitis.
In: Aktuelle Neuropädiatrie 1996 (Eds.: E Boltshauser).
20. MR Calomme, V Th Ramaekers, W Makropoulos, DA VandenBerghe.
Selenium deficiency triggering intractable seizures: A case study. In: Therapeutic Uses of Trace Elements (Eds. Nève J et al.) 1996; chapter 62: 359-364.
21. Ramaekers VTh. Cerebellar malformations. In: Handbook of Ataxia Disorders. Editor: Th Klockgether. Marcel Dekker Publishers - New York 2000.
22. Ramaekers V Th. Assessment of Food Quality in North and South with respect to the Health and Development of Childhood. Book of the IAAS World Congress Symposium held on July 31st and August 1st 2003 at the University of Leuven – Begium.
24. Ramaekers VT. Folate Deficiency States. In: New Encyclopaedia of Neuroscience (2008).
25. Ramaekers VT and Quadros EV. Folate receptor autoimmunity in Cerebral Folate Deficiency. Chapter 19 in: Inflammatory and autoimmune disorders of the Central nervous system in children. Editors: Russell C Dale and Angela Vincent (2010; Mac Keith Press).

PUBLISHED PRESENTATIONS

1. F Kotlarek, U Rubenstrunk, V Th Ramaekers *, G Gross-Selbeck, K Kellermann, K Rheingans, W Mortier, R Pothmann, H von Bernuth, U Schauseil-Zipf.
* presenting
Controlled study of Valproate in infants with West's syndrome.
The fourth International Symposium on Sodium Valproate and Epilepsy- 26-28 April 1989.

PRESENTATIONS AND POSTERS CITED AS ABSTRACT

1. V Th Ramaekers, P Casaer, G Marchal, H Daniels.
Variability in cerebral blood flow and the upper limits of cerebral blood flow auto-regulation in preterm infants.
Meeting of the European Federation of Child Neurology Societies. Hyvinkaa, Finland, June 15-18, 1987.
Neuropediatrics 1987; 18:121.
2. V Th Ramaekers, P Casaer, G Marchal, M Smet and W Goossens.
The effect of adult red blood cell transfusion on cerebral blood flow velocity in preterm infants: a Doppler study.
Internationales Symposium in Münster – Neue Aspekte des Blutkreislaufes und Sauerstofftransportes bei Frühgeborenen. 11-12 June 1988.
Klinische Pädiatrie 1988; 200: 149.

3. V Th Ramaekers, J Jaeken, P Casaer.
Zerebrale Durchblutungsstörungen bei vorübergehender neonataler Hyperammoniemie (VNHS).
Monatschrift für Kinderheilkunde 1988; 136: 552.
4. V Th Ramaekers, M Brab, T Suormala, ER Baumgartner, G Heimann.
A biotinidase Km variant causing acute onset optic neuropathy associated with neurological abnormalities.
Presented at the 26th Workshop for Pediatric Research
(held at the Kinderklinik of the Georg-August-University Göttingen, February 22-23, 1990).
European Journal of Pediatrics 1990; 149:370.
5. V Th Ramaekers, J Schneider, C Disselhorst-Klug, J Silny, G Rau, R Forst, F Kotlarek.
Nichtinvasive Elektromyographie motorischer Einheiten. Vorstellung einer neuen Methodik und erste klinische Erfahrungen im Kindesalter.
Proceedings of the Neuropaediatric Congress Basel 1990; published in " Aktuelle Neuropädiatrie 1990" (edited by Springer Verlag).
6. J Reul, V Ramaekers, U Spetzger, A Thron.
Mesenzephalen Ventrikelysten als Ursache eines persistierenden Hydrozephalus im Kindesalter.
Diagnostische und therapeutische Probleme dargestellt an vier Fällen.
Der Radiologe (1995) Band 35 Heft 4; page 130.
7. FM Müller, S Noth, F Speckkamp, G Alzen, M Mayer, R Mertens, V Ramaekers, G Heimann.
Differentialdiagnostische Schwierigkeiten bei der Frühdiagnostik einer Dermatomyositis.
Monatschrift der Kinderheilkunde (1995).
8. T Peschgens, V Th Ramaekers, U Merz, O Blankenstein, W Dott, H Hörnchen.
Selen-Status und Selen-Substitution bei Frühgeborenen < 1500 g..
Zeitschr Geburtsh Neonatol (1995) 199: 231.
9. J Reul, U Weber, F Kotlarek, V Ramaekers, A Thron.
The "Otitic Hydrocephalus": pseudotumor cerebri syndrome (benign intracranial hypertension) and dural sinus thrombosis in childhood.
Neuroradiology (1996) 38: S 37.
10. V Th Ramaekers, J Reul, M Häusler, G Heimann.
Varizella-Zoster assoziierte cerebrale Vaskulitis.
Monatsschr Kinderheilkd (1996) 144: 981.
11. V Th Ramaekers, P Deutz, R Pfäffle, P Cos, M Calomme, D Vanden Berghe, A van Gennip, R Wanders.
Pontocerebellar hypoplasia type 2 associated with defective Mn-superoxide dismutase gene transcription or lowered extracellular Cu/Zn-superoxide dismutase activity.
8th International Child Neurology Congress Sept 13-17, 1998 Ljubljana, Slovenia
Brain and Development 20 (6) (1998): 358 (abstracted).
12. V Th Ramaekers, B Steinau, AC Sewell, H-J Boehles, G Heimann.
Co-enzyme Q10 deficiency associated with acrocallosal syndrome and an autosomal recessive

spastic atxix syndrome.

8th International Child Neurology Congress Sept 13-17, 1998 Ljubljana, Slovenia
Brain and Development 20 (6) (1998): 430 (abstracted)

13. V Th Ramaekers, J Jaeken.

Genetic disorders and cerebellar structural abnormalities in childhood.

8th International Child Neurology Congress Sept 13-17, 1998 Ljubljana, Slovenia
Brain and Development 20 (6) (1998): 430 (abstracted)

14. V Th Ramaekers, M Calomme, P Cos, D Vanden Berghe

Selenium deficiency: an important trigger of intractable epilepsy amenable to supplementation.

8th International Child Neurology Congress Sept 13-17, 1998 Ljubljana, Slovenia
Brain and Development 20 (6) (1998): 483 (abstracted)

15. V Th Ramaekers, M Häusler, N Blau.

Tryptophan Hydroxylase Deficiency

VIII International Congress of Inborn Errors of Metabolism, Cambridge (2000) 13-17 September. J
Inherit Metab Dis 23 (2000) Suppl. 1: 207.

16. V Th Ramaekers, M Häusler, N Blau.

A novel syndrome due to impaired folate transport to the nervous system.

4th Congress of the European Paediatric Neurology Society (2001) Baden-Baden 12-16
September.

17. V Th Ramaekers, J Senderek, M Häusler, K Zerres, N Blau.

A novel neurodevelopmental syndrome affecting de novo serotonin synthesis.

4th Congress of the European Paediatric Neurology Society (2001) Baden-Baden 12-16
September.

18. V Ramaekers, M Calomme, P Cos, R Wanders, M Häusler, N Abeling, A van Gennip, D
Vanden berghe, N Blau.

Ataxia and Retardation associated with disturbed Manganese Uptake.

J Inherit Metab Dis 25 (2002) Suppl. 1: 166 (Abstracted).

19. V Th Ramaekers, N Blau.

Reduced folate transport to the brain in Rett syndrome.

Thirty-First Annual Meeting of the Child Neurology Society.

October 9-12 October 2002, Washington DC.

20. V Th Ramaekers

Folate Transportstörungen über die Blut-Hirnschranke im Kindesalter.

Fortbildung Kinderkliniken Heidelberg und Mannheim

14-5-2003.

21. V Th Ramaekers

Reduced folate transport to the brain in Rett syndrome.

Invited Lecture at the Rett TEP Conference of the Rett Association Illinois

Chicago November 1st 2003.

Folate and it's impact upon motor and cognitive function.

Invited lecture for teachers and therapists at the Rett TEP conference

Chicago November 2nd 2003.

22. V Th Ramaekers
Invited Lectures at Baylor College of Medicine, Institute of Molecular and Human Genetics and Texas Children's Hospital. Houston November 3-6th 2003.
23. V Th Ramaekers
Folate Transporter Deficiency.
Invited Lecture on January 15th 2004.
Focus on Neuropaediatrics. Brain Metabolism revisited – concepts and treatment.
24. V Th Ramaekers, M Hausler, C Bergmann, J Senderek, T Eggermann, M Mull, K Zerres.
Deletion of Oligophrenin-1 Gene causes a Syndrome of X-linked Mental Retardation.
5th Congress of the European Pediatric Neurology Society EPNS. Taormina, Italy.
22-25 October 2003.
25. V Ramaekers, M Hausler, N Blau.
Ataxia and Retardation associated with Manganese Deficiency.
5th Congress of the European Pediatric Neurology Society EPNS. Taormina, Italy.
22-25 October 2003.
26. V Ramaekers
Cerebral folate deficiency in childhood: differential diagnosis and treatment.
Invited lecture on december 4th 2003.
Symposium of the Belgian Society of Pediatric Neurology, Gent.
27. V Ramaekers
Cerebral Folate Deficiency. What's in a name. Invited Lecture May 12th 2006.
Symposium of the Belgian Society of Pediatric Neurology, Antwerp.

ORGANIZATION OF SYMPOSIUM ON PROCEEDINGS OF NEW NEUROMETABOLIC DISEASES.

1. Neue Neurometabole Erkrankungen im Kindesalter

University Clinic Aachen held on 22. Januar 2003

Organiser: PD Dr V Th Ramaekers

Programm: Prof Dr J Jaeken.

G protein defects: A newly recognized cause of metabolic Encephalopathy.

Prof Dr G Hoffmann.

Primäre und sekundäre Störung der Neurotransmission

PD Dr V Th Ramaekers.

Störungen der Blut-Hirn-Schranke:

Perspektiven der Diagnostik und Therapie.

2. Invitation to present the Grand Rounds at Harvard Medical School, Boston, USA

“ Cerebral Folate Deficiency in Childhood ”.

“ CFD: Diagnosis and Clinical management ”.

Lecture at the Longwood Neurology Grand Rounds (Brigham Women's Hospital, Beth Israel and Boston Children's Hospital) on December 15th 2004.

Lecture at the Massachusetts General Hospital (Ether Dome) on December 16th.
Lecture at the USDA Department, Tufts University, Boston on December 17th.

Comment by Professor Verne Caviness (Head of Child Neurology, Mass General Hospital):

‘ The presentation at the Dome was a highlight of the year and for us a strong finish for the year. This was a window into an entirely new landscape of clinical biology and will alter the way we think about this range of disorders.’

3. Workshop on “Cerebral Folate Deficiency”, 38th EMG meeting in Istanbul; 2006 from may 24-28.

EMG publications on Proceedings of the European Metabolic Group

NON-CITED PRESENTATIONS

1. V Th Ramaekers, BD Lake, B Harding, S Boyd, A Harden, EM Brett and J Wilson.
Diagnostic difficulties in Infantile Neuroaxonal Dystrophy. A clinicopathological study of eight cases.
Meeting of the British Paediatric Neurology Association; Cardiff, 3rd-5th January 1986.
2. V Th Ramaekers
Selective Decontamination of the Digestive Tract in Paediatric Oncology.
Antoni van Leeuwenhoek Hospital
Amsterdam, March 3rd, 1986.
3. V Th Ramaekers, H Daniels, P Casaer, G Marchal, E Eggermont, H Devlieger.
Fluctuating patterns of cerebral blood flow in infants and prematures.
Second Annual Meeting of the Federation of the Belgian Societies for Ultrasound Diagnosis.
May 10th, 1986.
4. FJ Kirkham, S Levin, V Th Ramaekers, BGR Neville, Padayachee, RG Gosling.
Clinical Use of Transcranial Pulsed Doppler Ultrasound to measure Changes in Middle Cerebral Artery Blood Flow.
In: British Paediatric Association, York 1985; 57th Annual Meeting.
5. V Th Ramaekers, P Casaer, G Marchal, H Daniels.
Variability in cerebral blood flow and the upper limits of cerebral blood flow autoregulation in preterm infants.
Seminarium Kindergeneeskunde – Katholieke Universiteit Leuven
19 December 1986.
6. V Th Ramaekers, P Casaer, G Marchal.
Cerebrale circulatie van de boorling en zijn relatie tot gedragstoestand, bloeddruk en arteriële zuurstofspanning.
Belgische Vereniging voor Kinderneurologie
Brussel, 25 April 1987.
7. M Smet, V Th Ramaekers, P Casaer, G Marchal, H Daniëls, AL Baert.
Cerebrale flowmetingen bij de neonatus
8. V Th Ramaekers
Die Rolle von Selen bei Epilepsien im Kindesalter.

Symposium der Kinderklinik RWTH 24-6-1995.

9. V Th Ramaekers. Cerebellar degeneration in infancy and childhood. Teaching course Diagnosis of childhood onset neurogenetic diseases in adults. Eleventh Meeting European Neurological Society April 21-25, 2001; Paris; France.

10. Symposium Neue Neurometabolische Erkrankungen im Kindesalter Januar 22th, 2003.
Organisation: PD Dr V Th Ramaekers
Prof Dr J Jaeken: G Protein defects: a newly recognized cause of metabolic encephalopathy.
Prof Dr G Hoffmann: Primäre und sekundäre Störung der Neurotransmission.
PD Dr V Th Ramaekers: Störungen der Blut-Hirn Schranke: Perspektive der Diagnostik und Therapie.