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Phenylketonuria: towards mechanism-based treatment

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List of publications

List of publications

- van Vliet D, Anjema K, Jahja R, **de Groot MJ**, Liemburg GB, Heiner-Fokkema MR, van der Zee EA, Derks TG, Kema IP, van Spronsen FJ. BH4 treatment in BH4-responsive PKU patients: preliminary data on blood prolactin concentrations suggest increased cerebral dopamine concentrations. *Mol. Genet. Metab.* 114 (2015) 29-33.
- **de Groot MJ**, Sijens PE, Reijngoud DJ, Paans AM, van Spronsen FJ. Phenylketonuria: brain phenylalanine concentrations relate inversely to cerebral protein synthesis. *J. Cereb. Blood Flow Metab.* 35 (2015) 200-205.
- van Vliet D, Derks TG, van Rijn M, **de Groot MJ**, MacDonald A, Heiner-Fokkema MR, van Spronsen FJ. Single amino acid supplementation in aminoacidopathies: a systematic review. *Orphanet J. Rare Dis.* 9 (2014) 7.
- **de Groot MJ***, Hoeksma M*, Reijngoud DJ, de Valk HW, Paans AM, Sauer PJ, van Spronsen FJ. Phenylketonuria: reduced tyrosine brain influx relates to reduced cerebral protein synthesis. *Orphanet J. Rare Dis.* 8 (2013) 133-141. * Equally contributing authors.
- **de Groot MJ**, Hoeksma M, van Rijn M, Slart RH, van Spronsen FJ. Relationships between lumbar bone mineral density and biochemical parameters in phenylketonuria patients. *Mol. Genet. Metab.* 105 (2012) 566-570.
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- **de Groot MJ**, Cuppen M, Eling M, Verheijen FW, Rings EH, Reijngoud DJ, de Vries MM, van Spronsen FJ. Metabolic investigations prevent liver transplantation in two young children with citrullinemia type I. *J. Inherit. Metab. Dis.* 33 Suppl. 3 (2010) S413-S416.
- **de Groot MJ**, Hoeksma M, Blau N, Reijngoud DJ, van Spronsen FJ. Pathogenesis of cognitive dysfunction in phenylketonuria: review of hypotheses. *Mol. Genet. Metab.* 99 Suppl.1 (2010) S86-S89.
- **de Groot MJ**, Bruinenberg VM, van Vliet D, Douwenga W, Kema IP, Reijngoud DJ, van Spronsen FJ, van der Zee EA. The absence of learning and memory

deficits in C57Bl/6 Pah-enu2 PKU mice is associated with normalization of reduced pCREB to CREB ratios. Under revision.

- Anjema K, **de Groot MJ**, Bruinenberg VM, van Vliet D, van der Goot E, Heiner-Fokkema MR, Kema IP, van Faassen M, Scherer T, Thöny B, van der Zee EA, van Spronsen FJ. Effects of tetrahydrobiopterin on brain neurotransmitter concentrations in C57Bl/6 PKU mice. Submitted.
- **de Groot MJ**, Bruinenberg VM, Mazzola PN, Kema IP, Reijngoud DJ, van Spronsen FJ, van der Zee EA. C57Bl/6 Pah-enu2 PKU mice show motor deficits, altered exploration behavior, and increased depression-like behavior associated with reduced brain concentrations of monoaminergic neurotransmitters. In preparation.

