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Autosomal dominant adult neuronal ceroid lipofuscinosis

Nijssen, P.C.G.

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List of abbreviations

ABR: Auditory Brainstem Responses
AD-ANCL : autosomal dominant adult neuronal ceroid lipofuscinosis
ANCL: adult NCL
CLN 1 to 8: NCL genes
CNS: central nervous system
CONCL: congenital ovine NCL
CTSD: cathepsin D
EEG : electroencephalogram
EM: electron microscopy
EPMR: progressive epilepsy with mental retardation
FPs: fingerprint profiles
GRODs : granular osmiophilic deposits
IBZM: iodobenzamide
INCL: infantile NCL
JNCL: juvenile NCL
LAG1: longevity assurance gene 1.
LINCL : late-infantile NCL
MFSD8: major facilitator superfamily domain containing 8 gene
MIM: Mendelian inheritance of man
MRI: magnetic resonance imaging
NCL: neuronal ceroid lipofuscinosis
OCT: optical coherence tomography
PAS: periodic acid Schiff
PPT : palmitoyl protein thioesterase
SAPs : saposins
SCMAS : subunit c of mitochondrial ATPase
STR: short tandem repeat
TPP1: tripeptidyl peptidase 1
UBQ: ubiquitin

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Curriculum vitae

Peter CG Nijssen

born 25-11-1961 in Geleen, The Netherlands

In 1980 he participated in the Bessie F Lawrence summer program of the Weizmann Institute of Science, Rehovot, Israel. He studied chemistry in Utrecht, but switched to medicine.

In 1984-85 an exchange program consisted of a neuroscience program and neuropharmacology at the University of Florida, Gainesville, studying opiate receptors and second messengers with dr. S.R. Childers. In 1986 neuropharmacological studies focussing on beta-endorphin and behaviour were performed at the Rudolf Magnus Institute, Utrecht (prof. dr. J. van Ree).

From 1989 – 1996 he was a neurology resident in Tilburg (dr. A.A.W. op de Coul and dr. C.C. Tijssen). He studied nonlinear analysis of electroencephalography in coma with prof. F.H. Lopes da Silva and prof. C.J. Stam.

Since 1996 he works as a neurologist specialised in Parkinsons disease, movement disorders and functional stereotaxy, in both St. Elisabeth hospital and Tweestedenziekenhuis, Tilburg, The Netherlands. He is a member of the medical advisory board of the Dutch Parkinson patient organisation, a boardmember of the Dutch movement disorders workinggroup of the Nederlandse Vereniging voor Neurologie and chairman of the Dutch Parkinson Genes and Environment Studygroup (PAGES).

He married Ruth Fleischeuer on may 29, 2010.

