Curriculum vitae

ACADEMIC POSITION

Executive senior consultant and lecturer in Neurology at Dept. of Neurology at University Hospital of Essen, Germany

ACADEMIC EDUCATION

2017 -	Executive senior consultant and lecturer Dept. of Neurology at University Hospital of Essen, Germany
2015 - 2017	Senior consultant and lecturer Dept. of Neurology at University Hospital of Freiburg, Germany; Scientific Affiliation with Dept. of Neurology at University Hospital of Würzburg, Germany
2011-2015	Senior consultant and lecturer Dept. of Neurology at University Hospital of Würzburg, Germany
2010-2011	Board certified Neurologist, Departement of Neurology, Salpetriere Hospital, Paris, France
2009-2010	Board certified Neurologist and lecturer in Neurology at Dept. of Neurology at University Hospital of Schleswig – Holstein, Kiel
2008-2009	Residency in Psychiatry at "Zentrum für integrative Psychiatrie" (ZIP) Christian Albrecht University Kiel
2006-2008	Residency and Research Scientist at the Dept. of Neurology at University Hospital of Schleswig – Holstein, Kiel
2004-2006	Scholarship of the "German Academic Exchange Service" (DAAD) in Paris/France (Hôpital de la Salpêtrière/INSERM U289): "Identification of new chromosomal Loci and mutations of autosomal recessice forms of hereditary spastic paraplegia"
2001-2004	Residency and Research Scientist at the Dept. of Neurology of Christian Albrecht University Kiel
2000-2001	Medical Internship at the Dept. of Neurology of Christian Albrecht University Kiel
1993-2000	Studies of Medicine at Georg – August University Göttingen and Christian Albrecht University Kiel

CERTIFICIATION

2017-	Professorship in Neurology of the University of Duisburg-Essen, Essen, Germany
2015 -2017	Professorship in Neurology of the of the Julius-Maximilians-Universität (JMU) Würzburg, Germany
07/2009	Board certified Neurologist
02/2009	Habilitation in „Molecular Neurology“ at Christian-Albrechts-University, Kiel: “Chromosomal linkage, candidate gene analysis and direct sequencing in hereditary spastic paraplegia and spinocerebelar ataxia type 14 (SCA14)”
2008	EMG and EP Certification of the German Society of Clinical Neurophysiology **(**DGKN**)**
2007	EEG Certification of the German Society of Clinical Neurophysiology **(**DGKN**)**
2006	Certification « Botulinum toxin » therapy of the German society of Botulinum (AK Botulinumtoxin)
2001	Medical Thesis: Theme: „Detection of transcripts for Parathormone-related-protein (PTHrP) in peripheral stem cell specimen of patients with high risk breast cancer” (Magna cum laude). Supervisor: Prof. B. Wörmann, Dept. of Internal Medicine, University of Göttingen.

AREAS OF CLINICAL EXPERTISE

Neurogenetic Disorders, esp. hereditary spastic paraplegia (HSP),

spinocerebellar ataxias (SCA), Huntington

Parkinson Disease

Botulinum toxin treatment of movement disorders

FIELDS OF RESEARCH

Genetic analysis of neurological disorders with monogenic and/or complex transmission

Clinical trials of movement disorders, esp. essential tremor and hereditary spastic paraplegia

MEMBERSHIPS

German Society of Neurology (DGN)

Arbeitskreis Botulinumtoxin

REGULAR REVIEWS FOR

Brain, Neurology, Movement Disorder, Journal of Neurology, Neurology und Neuropsychiatric, Human Genetics, Clinical Genetics

ORIGINAL ARTICLES

Klebe S, Timmann D. [Genetics of movement disorders-rare but important]. Nervenarzt. 2019 Feb.

Weber J, Frings L, Rijntjes M, Urbach H, Fischer J, Weiller C, Meyer PT, **Klebe S**. Chorea-Acanthocytosis Presenting as Autosomal Recessive Epilepsy in a Family With a Novel *VPS13A* Mutation. Front Neurol. 2019 Jan 9

Matlach J, Wagner M, Malzahn U, Schmidtmann I, Steigerwald F, Musacchio T, Volkmann J, Grehn F, Göbel W, **Klebe S**. Retinal changes in Parkinson's disease and glaucoma. Parkinsonism Relat Disord. 2018:41-46

Whittaker K, Schroeter N, Rijntjes M, Guggenberger K, Lambeck J, Fritsch B, Weiller C, Meyer PT, Doostkam S, Klebe S. Severe camptocormia due to myositis of paraspinal muscles as an early manifestation of Parkinson's disease. Parkinsonism Relat Disord. 2018: 95-97.

Weber J, Piroth T, Rijntjes M, Jung B, Reinacher PC, Weiller C, Coenen VA, **Klebe S** Atypical Presentation of Rapid-onset Dystonia-parkinsonism (DYT12) Unresponsive to Deep Brain Stimulation of the Subthalamic Nucleus. Mov Disord Clin Pract. 2018:427-429

Jost M, Rijntjes M, Urbach H, Egger K, Meyer PT, Frings L, Weiller C, **Klebe S**. Ataxia and autonomic dysfunction as presenting symptoms in late-onset Alexander. disease. Neurol Clin Pract. 2017

Piroth T, Boelmans K, Amtage F, Rijntjes M, Wierciochin A, Musacchio T, Weiller C, Volkmann J, **Klebe S**. Adult-onset Niemann-Pick disease type C: rapid treatment initiation advised, but early diagnosis remains difficult. Frontiers in Neurology, 2017, accepted.

Rauschendorf MA, Jost M, Stock F, Zimmer A, Rösler B, Rijntjes M, Piroth T, Coenen VA, Reinacher PC, Meyer PT, Frings L, Weiller C, Fischer J, **Klebe S**. Novel compound heterozygous synaptojanin-1 mutation causes l-dopa-responsive dystonia-parkinsonism syndrome. *Mov Disord.* 2016 Nov 21. [Epub ahead of print]

Musacchio T, Zaum AK, Üçeyler N, Sommer C, Reiners K, Kunstmann E, Volkmann J, Rost S, **Klebe S**. ALS and MMN mimics in patients with BSCL2 mutations: the expanding clinical spectrum of SPG17 hereditary spastic paraplegia. *J Neurol.* 2016 Oct 13. [Epub ahead of print]

Musacchio T, Purrer V, Papagianni A, Fleischer A, Mackenrodt D, Malsch C, Gelbrich G, Steigerwald F, Volkmann J, **Klebe S**. Non-Motor Symptoms of Essential Tremor Are Independent of Tremor Severity and Have an Impact on Quality of Life. *Tremor Other Hyperkinet Mov*. **2016** .

Klebe S, Depienne C, Stevanin G. Clinical and genetic heterogeneity in hereditary spastic paraplegias: From SPG1 to SPG72 and still counting. *Revue Neurologique*, **2014,** 171; 505 – 30

Mencacci N, Isaias IU, Reich M, Ganos C, Plagnol V, Polke JM, Bras J, Hersheson J, Stamelou M, Pittman A, Alastair NJ, Mok KJ, Opladen T, Kunstmann E, Hodecker S, Münchau A, Volkmann J, Samnick S, Sidle K, Nanji T, Sweeney MG, Houlden H, Batla A, Zecchinelli AL, Pezzoli G, Marotta G, Lees A, Alegria P, Krack P, Cormier F, Lesage S, Brice A, Heutink P, Gasser T, Lubbe SJ, Morris HR, Taba P, Koks S, Majounie E, Gibbs R, Singleton A, Hardy J, **Klebe S*,** Bhatia KP*, Wood NW*, for the International Parkinson's Disease Genomics Consortium and UCL-exomes consortium. Parkinson disease in GTP cyclohydrolase-1 mutation carriers. *Brain*, 137; **2014,** 2480-92

Klebe S, Golmard JL, Nalls MA, Saad M, Singleton AB, Bras JM, Hardy J, Simon-Sanchez J, Heutink P, Kuhlenbäumer G, Charfi R, Klein C, Hagenah J, Gasser T, Wurster I, Lesage S, Lorenz D, Deuschl G, Durif F, Pollak P, Damier P, Tison F, Durr A, Amouyel P, Lambert JC, Tzourio C, Maubaret C, Charbonnier-Beaupel F, Tahiri K, Vidailhet M, Martinez M, Brice A, Corvol JC; French Parkinson's Disease Genetics Study Group and the International Parkinson's Disease Genomics Consortium (IPDGC). The Val158Met COMT polymorphism is a modifier of the age at onset in Parkinson's disease with a sexual dimorphism. ***J Neurol Neurosurg Psychiatry,*** **2013***,* 666 - 672

Klebe S, Depienne C, Gerber S, Challe G, Anheim M, Charles P, Fedirko E, Lejeune E, Cottineau J, Brusco A, Dollfus H, Chinnery PF, Mancini C, Ferrer X, Sole G, Destée A, Mayer JM, Fontaine B, Seze Jd, Clanet M, Ollagnon E, Busson P, Cazeneuve C, Stevanin G, Kaplan J, Rozet JM, Brice A, Durr A. Spastic paraplegia gene 7 in patients with spasticity and/or optic neuropathy. ***Brain,*** **2012,** 2980-93

Thier S, Lorenz D, Nothnagel M, Poremba C, Papengut F, Appenzeller S, Paschen S, Hofschulte F, Hussl AC, Hering S, Poewe W, Asmus F, Gasser T, Schöls L, Christensen K, Nebel A, Schreiber S, **Klebe S*,** Deuschl G, Kuhlenbäumer G. Polymorphisms in the glial glutamate transporter SLC1A2 are associated with essential tremor. ***Neurology,*** **2012***,* 243-248

Klebe S, Lossos A, Azzedine H, Mundwiller E, Sheffer R, Gaussen M, Marelli C, Nawara M, Carpentier W, Meyer V, Rastetter A, Martin E, Bouteiller D, Orlando L, Gyapay G, El Hachimi KH, Zimmerman B, Gamliel M, Misk A, Lerer I, Brice A, Durr A, Stevanin G. KIF1A missense mutations in SPG30, an autosomal recessive spastic paraplegia: distinct phenotypes according to the nature of the mutations. ***Eur J Hum Genet,*** **2012***,* 645-49.

Thier S. Kuhlenbäumer G, Lorenz D, Nothnagel M, Nebel A, Christensen K, Schreiber S, Deuschl G, **Klebe, S.** $GABA_A$ Receptor- and GABA Transporter Polymorphisms and Risk for Essential Tremor. ***Eur J Neurol*****, 2011,** 1098-100

Lang N, Optenhoefel T, Deuschl G, **Klebe S.** Axonal integrity of corticospinal projections to the upper limbs in patients with pure hereditary spastic paraplegia. **Clinical Neurophysiology, 2011,** 1417-20

Klebe S, Their S, Lorenz D, Nothnagel M, Schreiber S, Klein C, Hagenah J, Kasten M, Gasser T, Deuschl G, Kuhlenbäumer G. LINGO1 is not associated with Parkinson's disease in German patients. ***AJMG Part B.***, **2010**, 1173-8

Thier S, Lorenz D, Nothnagel, M, Stevanin G,Dürr A, Nebel A, Schreiber S, Kuhlenbäumer G, Deuschl G, **Klebe S.** LINGO1 polymorphisms are associated with essential tremor in Europeans. ***Mov Disord.*****, 2010,** 717-23

Klebe S, Lorenz D, Stevanin G, Thier S, Nebel A, Feingold J, Frederiksen H, Denis E, Christensen K, Schreiber S, Brice A, Deuschl G, Dürr A. Dopamine receptor D3 gene and essential tremor in large series of German, Danish and French patients. ***Eur J Hum Genet.*** **2009**,17(6):766-73

Klebe S, Durr A, Bouslam N, Grid D, Paternotte C, Depienne C, Hanein S, Bouhouche A, Elleuch N, Azzedine H, Poea-Guyon S, Forlani S, Denis E, Charon C, Hazan J, Brice A, Stevanin G. Spastic paraplegia 5: locus refinement, candidate gene analysis and clinical description. ***Am J Med Genet B Neuropsychiatr Genet.*** **2007**, 144B(7):854-61

Klebe S, Faivre L, Forlani S, Dussert C, Tourbah A, Brice A, Stevanin G, Durr A. Another mutation in the cysteine 131 residue in Protein Kinase C causes Spinocerebellar Ataxia Type 14 (SCA14). ***Arch Neurol***. **2007**, 64(6):913-4.

Klebe S, Lacour A, Durr A, Stojkovic T, Depienne C, Forlani S, Poea-Guyon S, Vuillaume I, Sablonniere B, Vermersch P, Brice A, Stevanin G. NIPA1 (SPG6) mutations are a rare cause of autosomal dominant spastic paraplegia in Europe. ***Neurogenetics***. **2007**, (2):155-7.

Klebe S, Deuschl G, Stolze H. Methylphenidate fails to improve gait and muscle tone in patients with sporadic and hereditary spastic paraplegia. ***Mov Disord.*** **2006**, 21: 1468-1471

Klebe S, Azzedine H, Durr A, Bastien P, Bouslam N, Elleuch N, Forlani S, Charon C, Koenig M, Melki J, Brice A, Stevanin G. Autosomal recessive spastic paraplegia (SPG30) with mild ataxia and sensory neuropathy maps to chromosome 2q37.3. ***Brain.*** **2006**, 129: 1456-1462

Klebe S, Durr A, Rentschler A, Hahn-Barma V, Abele M, Bouslam N, Schöls L, Jedynak P, Forlani S, Denis E, Dussert C, Agid Y, Bauer P, Globas C, Wüllner U, Brice A, Riess O, Stevanin G. New mutations in Protein Kinase Cγ associated with Spinocerebellar Ataxia Type 14 (SCA14). ***Ann Neurol.*** **2005**, 58: 720-729.

Klebe S, Stolze H, Grensing K, Volkmann J, Wenzelburger R, Deuschl G. Influence of alcohol on gait in patients with essential tremor. ***Neurology***. **2005**, 65(1):96-101.

Klebe S, Stolze H, Kopper F, Lorenz D, Wenzelburger R, Deuschl G, Volkmann J. Objective assessment of gait after application of intrathecal baclofen in hereditary spastic paraplegia. ***J Neurol.*** **2005**, 252(8):991-3

Klebe S, Stolze H, Kopper F, Lorenz D, Wenzelburger R, Volkmann J, Porschke H, Deuschl G. Gait analysis of sporadic and hereditary spastic paraplegia. ***J Neurol.*** **2004**, 251: 571-8.

*** Co-shared authorship**

Ebersbach G, Ip CW, **Klebe S**, Koschel J, Lorenzl S, Schrader C, Winkler C, Franke C. Management of delirium in Parkinson's disease. J Neural Transm, 2019 Epub ahead of print

Oosterloo M, Bijlsma EK, van Kuijk SM, Minkels F, de Die-Smulders CE; **REGISTRY** Investigators of the European Huntington's Disease Network; Registry Steering committee; Language coordinators; EHDN's associate site in Singapore. Clinical and genetic characteristics of late-onset Huntington's disease. Parkinsonism Relat Disord. 2018 Nov 29.

Ludolph AC, Schuster J, Dorst J, Dupuis L, Dreyhaupt J, Weishaupt JH, Kassubek J, Weiland U, Petri S, Meyer T, Grosskreutz J, Schrank B, Boentert M, Emmer A, Hermann A, Zeller D, Prudlo J, Winkler AS, Grehl T, Heneka MT, Wollebæk, Johannesen S, Göricke B; **RAS-ALS Study Group**. Safety and efficacy of rasagiline as an add-on therapy to riluzole in patients with amyotrophic lateral sclerosis: a randomised, double-blind, parallel-group, placebo-controlled, phase 2 trial. Lancet Neurol. 2018:681-688.

Corvol JC, Artaud F, Cormier-Dequaire F, Rascol O, Durif F, Derkinderen P, Marques AR, Bourdain F, Brandel JP, Pico F, Lacomblez L, Bonnet C, Brefel-Courbon, C, Ory-Magne F, Grabli D, **Klebe S**, Mangone G, You H, Mesnage V, Lee PC, Brice A, Vidailhet M, Elbaz A; DIGPD Study Group. Longitudinal analysis of impulse control disorders in Parkinson disease. Neurology. 2018

Müller K, Brenner D, Weydt P, Meyer T, Grehl T, Petri S, Grosskreutz J, Schuster J, Volk AE, Borck G, Kubisch C, Klopstock T, Zeller D, Jablonka S, Sendtner M, **Klebe S**, Knehr A, Günther K, Weis J, Claeys KG, Schrank B, Sperfeld, AD, Hübers A, Otto M, Dorst J, Meitinger T, Strom TM, Andersen PM, Ludolph AC, Weishaupt JH; German ALS network MND-NET. Comprehensive analysis of the mutation spectrum in 301 German ALS families. J Neurol Neurosurg Psychiatry. 2018, 817-827

Rosenbohm A, Nagel G, Peter RS, Brehme T, Koenig W, Dupuis L, Rothenbacher D, Ludolph AC; **ALS Registry Study Group**. Association of Serum Retinol-Binding, Protein 4 Concentration With Risk for and Prognosis of Amyotrophic Lateral, Sclerosis. JAMA Neurol. 2018:600-607.

Rosenbohm A, Liu M, Nagel G, Peter RS, Cui B, Li X, Kassubek J, Rothenbacher D, Lulé D, Cui L, Ludolph AC; **ALS Registry Swabia Study Group**. Phenotypic differences of amyotrophic lateral sclerosis (ALS) in China and Germany. J Neurol. 2018:774-782

Liu G, Locascio JJ, Corvol JC, Boot B, Liao Z, Page K, Franco D, Burke K, Jansen IE, Trisini-Lipsanopoulos A, Winder-Rhodes S, Tanner CM, Lang AE, Eberly, S, Elbaz A, Brice A, Mangone G, Ravina B, Shoulson I, Cormier-Dequaire F, Heutink, P, van Hilten JJ, Barker RA, Williams-Gray CH, Marinus J, Scherzer CR; HBS; CamPaIGN; PICNICS; PROPARK; PSG; DIGPD; PDBP. Prediction of cognition in Parkinson's disease with a clinical-genetic score: a longitudinal analysis of nine cohorts. Lancet Neurol. 2017: 620-629

Rosenbohm A, Peter RS, Erhardt S, Lulé D, Rothenbacher D, Ludolph AC, Nagel G; **ALS Registry Study Group**. Epidemiology of amyotrophic lateral sclerosis in Southern Germany. J Neurol. 2017, 749-757

Doppler K, Jentschke HM, Schulmeyer L, Vadasz D, Janzen A, Luster M, Höffken H, Mayer G, Brumberg J, Booij J, Musacchio T, **Klebe S,** Sittig-Wiegand E, Volkmann J, Sommer C, Oertel WH.

Dermal phospho-alpha-synuclein deposits confirm REM sleep behaviour disorder as prodromal Parkinson's disease. Acta Neuropathol, 2017, 133(4):535-545.

Müller SH, Girard SL, Hopfner F, Merner ND, Bourassa CV, Lorenz D, Clark LN, Tittmann L, Soto-Ortolaza AI, **Klebe S**, Hallett M, Schneider SA, Hodgkinson CA, Lieb W, Wszolek ZK, Pendziwiat M, Lorenzo-Betancor O, Poewe W, Ortega-Cubero S, Seppi K, Rajput A, Hussl A, Rajput AH, Berg D, Dion PA, Wurster I, Shulman JM, Srulijes K, Haubenberger D, Pastor P, Vilariño-Güell C, Postuma RB, Bernard G, Ladwig KH, Dupré N, Jankovic J, Strauch K,Panisset M, Winkelmann J, Testa CM, Reischl E, Zeuner KE, Ross OA, Arzberger T, Chouinard S, Deuschl G, LouisED, Kuhlenbäumer G, Rouleau GA. *Brain*. 2016 Oct 20. [Epub ahead of print]

Liu G, Boot B, Locascio JJ, Jansen IE, Winder-Rhodes S, Eberly S, Elbaz A, Brice A, Ravina B, van HiltenJJ, Cormier-Dequaire F, Corvol JC, Barker RA, Heutink P, Marinus J, Williams-Gray CH, Scherzer CR; International Genetics of Parkinson Disease Progression (IGPP) Consortium: Scherzer C, Hyman BT, Ivinson AJ, Trisini-Lipsanopoulos A, Franco D, Burke K, Sudarsky LR, Hayes MT, Umeh CC, Growdon JH, Schwarzschild MA, HungAY, Flaherty AW, Wills AM, Mejia NI, Gomperts SN, Khurana V, Selkoe DJ, Yi T, Page K, Liao Z, Barker R, Foltynie T, Williams-Gray CH, Mason S, Winder-Rhodes S, Barker R, Williams-Gray CH, Breen D, Cummins G, Evans J, Winder-Rhodes S, Corvol JC, Brice A, Elbaz A, Mallet A, Vidailhet M, Bonnet AM, Bonnet C, Grabli D, Hartmann A, **Klebe S**, Lacomblez L, Mangone G, Bourdain F, Brandel JP, Derkinderen P, Durif F, Mesnage V, Pico F, Rascol O, Forlani S, Lesage S, Tahiri K, van Hilten JJ, Marinus J, Liao Z, Page K, Franco D, Duong K, Yi T, Trisini-Lipsanopoulos A, Dong X, Sudarsky LR, Hutten SJ, Amr SS, Shoulson I, Tanner CM, Lang AE, Nalls MA. Ann Neurol. 2016 Nov 80(5): 674 - 85.

Hopfner F, Ahlf A, Lorenz D, **Klebe S**, Zeuner KE, Kuhlenbäumer G, Deuschl G. Early- and late-onset essential tremor patients represent clinically distinct subgroups. *Mov Dis* **2016,** [Epub ahead of print]

Schüle R, Wiethoff S, Martus P, Karle KN, Otto S, **Klebe S**, Klimpe S, Gallenmüller C, Kurzwelly D, Henkel D, Rimmele F, Stolze H, Kohl Z, Kassubek J, Klockgether T, Vielhaber S, Kamm C, Klopstock T, Bauer P, Züchner S, Liepelt-Scarfone I, Schöls L. Hereditary Spastic Paraplegia -clinico-genetic lessons from 608 patients. *Ann Neurol.* **2016** [Epub ahead of print]

Lesage S, Drouet V, Majounie E, Deramecourt V, Jacoupy M, Nicolas A, Cormier-Dequaire F, Hassoun SM, Pujol C, Ciura S, Erpapazoglou Z, Usenko T, Maurage CA, Sahbatou M, Liebau S, Ding J, Bilgic B, Emre M, Erginel-Unaltuna N, Guven G, Tison F, Tranchant C, Vidailhet M, Corvol JC, Krack P, Leutenegger AL, Nalls MA, Hernandez DG, Heutink P, Gibbs JR, Hardy J, Wood NW, Gasser T, Durr A, Deleuze JF, Tazir M, Destée A, Lohmann E, Kabashi E, Singleton A, Corti O, Brice A; **French Parkinson's Disease Genetics Study (PDG);** International Parkinson's Disease Genomics Consortium (IPDGC). Loss of VPS13C Function in Autosomal-Recessive Parkinsonism Causes Mitochondrial Dysfunction and Increases PINK1/Parkin-Dependent Mitophagy. Am J Hum Genet. 2016: 500-513

Hu HH, Kannengiesser C, Lesage S, André J, Mourah S, Michel L, Descamps V, Basset-Seguin N, Bagot M, Bensussan A, Lebbé C, Deschamps L, Saiag P, Leccia MT, Bressac-de-Paillerets B, Tsalamlal A, Kumar R, **Klebe S**, Grandchamp B, Andrieu-Abadie N, Thomas L, Brice A, Dumaz N, Soufir N. PARKIN Inactivation Links Parkinson's Disease to Melanoma. *J Natl Cancer Inst.* **2015** Dec 17;108(3).

Jahic A, Khundadze M, Jaenisch N, Schüle R, Klimpe S, **Klebe S**, Frahm C, Kassubek J, Stevanin G, Schöls L, Brice A, Hübner CA, Beetz C. The spectrum of KIAA0196 variants, and characterization of a murine knockout: implications for the mutational mechanism in hereditary spastic paraplegia type SPG8. Orphanet J Rare Dis. **2015** 10

Doppler K, Weis J, Karl K, Ebert S, Ebentheuer J, Trenkwalder C, **Klebe, S**, Volkamnn J, Sommer C. Distinctive distribution of phospho-alpha-synuclein in dermal nerves in multiple system atrophy. *Mov Dis* **2015**, 12; 1688 – 92

Strickland AV, Schabhüttl M, Offenbacher H, Synofzik M, Hauser NS, Brunner-Krainz M, Gruber-Sedlmayr U, Moore SA, Windhager R, Bender B, Harms M, **Klebe S**, Young P, Kennerson M, Garcia AS, Gonzalez MA, Züchner S, Schule R, Shy ME, Auer-Grumbach M. Mutation screen reveals novel variants and expands the phenotypes associated with DYNC1H1. *J Neurology*, **2015**, 9; 2124 -34

Pyatigorskaya N, Sharman M, Corvol JC, Valabregue R, Yahia-Cherif L, Poupon F, Cormier-Dequaire F, Siebner H, **Klebe S**, Vidailhet M, Brice A, Lehéricy S.High nigral iron deposition in LRRK2 and Parkin mutation carriers using R2* relaxometry. *Mov Dis* **2015**, 30; 1077 – 84

Weidemann F, Liu D, Hu K, Florescu C, Niemann M, Herrmann S, Kramer B, **Klebe S**, Doppler K, Üçeyler N, Ritter CO, Ertl G, Störk S.The cardiomyopathy in Friedreich's ataxia - New biomarker for staging cardiac involvement. *Int J Cardiol*, **2015**, 194; 50 - 7

Hopfner F, Stevanin G, Müller SH, Mundwiller E, Bungeroth M, Durr A, Pendziwiat M, Anheim M, Schneider SA, Tittmann L, **Klebe S**, Lorenz D, Deuschl G, Brice A, Kuhlenbäumer G. The impact of rare variants in FUS in essential tremor. *Mov Dis* **2015**, Accepted

Mencacci NE, Pittman AM, Isaias IU, Hardy J, **Klebe S**, Bhatia KP, Wood NW. Reply: Parkinson's disease in GTP cyclohydrolase 1 mutation carriers. *Brain* **2014**, Accepted.

Méneret A, Depienne C, Riant F, Trouillard O, Bouteiller D, Cincotta M, Bitoun P, Wickert J, Lagroua I, Westenberger A, Borgheresi A, Doummar D, Romano M, Rossi M, Defebvre L, Meirleir L, Espay A, Fiori S, **Klebe S**, Quélin C, Rudnik-Schöneborn S, Plessis G, Dale R, Sklower Brooks S, Dziezyc K, Pollak P, Golmard JL, Vidailhet M, Brice A, and Roze E. Congenital mirror movements: mutational analysis of RAD51 and DCC in 26 cases. *Neurology* **2014**, 82; 1999 - 2002

Karle KN, Schüle R, **Klebe S**, Otto S, Frischholz C, Liepelt-Scarfone I, Schöls L. Electrophysiological characterisation of motor and sensory tracts in patients with hereditary spastic paraplegia (HSP). *Orphanet J Rare Dis.* **2013,** 8: 158.

Nalls MA, Duran R, Lopez G, Kurzawa-Akanbi M, McKeith IG, Chinnery PF, Morris CM, Theuns J, Crosiers D, Cras P, Engelborghs S, De Deyn PP, Van Broeckhoven C, Mann DM, Snowden J, Pickering-Brown S, Halliwell N, Davidson Y, Gibbons L, Harris J, Sheerin UM, Bras J, Hardy J, Clark L, Marder K, Honig LS, Berg D, Maetzler W, Brockmann K, Gasser T, Novellino F, Quattrone A, Annesi G, De Marco EV, Rogaeva E, Masellis M, Black SE, Bilbao JM, Foroud T, Ghetti B, Nichols WC, Pankratz N, Halliday G, Lesage S, **Klebe S**, Durr A, Duyckaerts C, Brice A, Giasson BI, Trojanowski JQ, Hurtig HI, Tayebi N, Landazabal C, Knight MA, Keller M, Singleton AB, Wolfsberg TG, Sidransky E. A Multicenter Study of Glucocerebrosidase Mutations in Dementia With Lewy Bodies. ***JAMA Neurol.*** **2013** Apr 15:1-9.

Lesage S, Condroyer C, **Klebe S**, Lohmann E, Durif F, Damier P, Tison F, Anheim M, Honoré A, Viallet F, Bonnet AM, Ouvrard-Hernandez AM, Vidailhet M, Durr A, Brice A; French Parkinson's

Disease Genetics Study Group. EIF4G1 in familial Parkinson's disease: pathogenic mutations or rare benign variants? ***Neurobiol Aging.*** **2012**, 2233.e1-2233.e5

Dufke C, Schlipf N, Schüle R, Bonin M, Auer-Grumbach M, Stevanin G, Depienne C, Kassubek J, **Klebe S**, Klimpe S, Klopstock T, Otto S, Poths S, Seibel A, Stolze H, Gal A, Schöls L, Bauer P. A high-throughput resequencing microarray for autosomal dominant spastic paraplegia genes. ***Neurogenetics,*** **2012,** 215-27.

Depienne C, Bouteiller D, Méneret A, Billot S, Groppa S, **Klebe S**, Djarmati A, Bhatia K, Cincotta M, Brochard V, Flamand-Roze C, Carpentier W, Meunier S, Marie Y, Gaussen M, Stevanin G, Wehrle R, Charbonnier-Beaupel F, Corvol JC, Vidailhet M, Klein C, Dusart I, Brice A, Roze E. RAD51 Haploinsufficiency Causes Congenital Mirror Movements in Humans. *AJHG***, 2012**, 301-7

Lesage S, Condroyer C, **Klebe S**, Honoré S, Tison F, Brefel-Courbon C, Dürr A, Brice A. Identification of VPS35 mutations replicated in French families with Parkinson disease. ***Neurology,*** **2012**, 1449-50

Klimpe S, Schüle R, Kassubek J, Otto S, Kohl Z, **Klebe S**, Klopstock T, Ratzka S, Karle K, Schöls L. Disease severity affects quality of life of hereditary spastic paraplegia patients. *Eur J Neurol.* **2012**, 168-71

Depienne C, Cincotta M, Billot B, Bouteiller D, Groppa S, Brochard V, Flamand C, Hubsch C, Meunier S, Giovanelli F, **Klebe S**, Corvol JC, Vidailhet M, Brice A, Roze E. Congenital mirror movements: a new DCC mutation, and evidence of genetic heterogeneity. ***Neurology*****, 2011.** 260-4.

Schlipf NA, Beetz C, Schüle R, Stevanin G, Erichsen AK, Forlani S, Zaros C, Karle K, **Klebe S**, Klimpe S, Durr A, Otto S, Tallaksen CM, Riess O, Brice A, Bauer P, Schöls L. A total of 220 patients with autosomal dominant spastic paraplegia do not display mutations in the SLC33A1 gene (SPG42). ***Eur J Hum Genet*****, 2010,** 1065-7.

Schüle R, Schlipf N, Synofzik M, **Klebe S**, Klimpe S, Hehr U, Winner B, Lindig T, Dotzer A, Riess O, Winkler J, Schöls L, Bauer P. **F**requency and phenotype of SPG11 and SPG15 in complicated hereditary spastic paraplegia. ***J Neurol Neurosurg Psychiatry.*** **2009,** 80(12):1402-4.

Schüle R, Brandt E, Karle KN, Tsaousidou M, **Klebe S**, Klimpe S, Auer-Grumbach M, Crosby AH, Hübner CA, Schöls L, Deufel T, Beetz C.Analysis of CYP7B1 in non-consanguineous cases of hereditary spastic paraplegia. ***Neurogenetics***. **2009**, 10(2):97-104

Lorenz D, Papengut F, Frederiksen H, Kopper F, **Klebe S**, Christensen K, Schreiber S, Deuschl G. Evaluation of a screening instrument for essential tremor. ***Mov Disord.*** **2008**, 23(7): 1006-12

Beetz C, Schüle R, **Klebe S**, Klimpe S, Klopstock T, Lacour A, Otto S, Sperfeld AD, van de Warrenburg B, Schöls L, Deufel T. Screening of hereditary spastic paraplegia patients for alterations at NIPA1 mutational hotspots. ***J Neurol Sci.*** **2008**, 268(1-2):131-5.

Elleuch N, Bouslam N, Hanein S, Lossos A, Hamri A, **Klebe S**, Meiner V, Birouk N, Lerer I, Grid D, Bacq D, Tazir M, Zelenika D, Argov Z, Durr A, Yahyaoui M, Benomar A, Brice A, Stevanin G. Refinement of the SPG15 candidate interval and phenotypic heterogeneity in three large Arab families. ***Neurogenetics.*** **2007**, (4):307-15.

Nolte D, **Klebe S**, Baron R, Deuschl G, Müller U. Codon 101 of PRKCG, a preferential mutation site in SCA14. ***Mov Disord.*** **2007**, 22(12):1831-2.

Bouslam N, Bouhouche A, Benomar A, Hanein S, **Klebe S**, Azzedine H, Di Giandomenico S, Boland-Augé A, Santorelli FM, Durr A, Brice A, Yahyaoui M, Stevanin G. A novel locus for autosomal recessive spastic ataxia on chromosome 17p. ***Hum Genet.*** **2007**, 121(3-4):413-20.

Stevanin G, Paternotte C, Coutinho P, **Klebe S**, Elleuch N, Loureiro JL, Denis E, Cruz VT, Durr A,

Prudhomme JF, Weissenbach J, Brice A, Hazan J. A novel locus for autosomal recessive spastic paraplegia (SPG32) maps to chromosome 14q12-q21. ***Neurology.*** **2007**, 68(21):1837-40

Deuschl G, Schade-Brittinger C, Krack P, Volkmann J, Schäfer H, Bötzel K, Daniels C, Deutschländer C, Dillmann U, Eisner W, Gruber D, Hamel W, Herzog J, Hilker R, **Klebe S**, Kloß M, Koy J, Krause M,, Kupsch A, Lorenz D, Lorenzl S, Mehdorn M, Moringlane JR, Oertel W, Pinsker MO, Reichmann H, Reuss A, Schneider GH, Schnitzler A, Steude U, Sturm V, Timmermann L, Tronnier V, Trottenberg T, Wojtecki L, Wolf E, Poewe W, Voges J. The German Parkinson Study Group, Neurostimulation Section. Deep brain stimulation for Parkinson's disease – a 6-month randomized, controlled trial. ***NEJM.*** **2006**, 355: 896-908

Lossos A, Stevanin G, Meiner V, Argov Z, Bouslam N, Newman JP, Gomori JM, **Klebe S**, Lerer I, Elleuch N, Silverstein S, Durr A, Abramsky O, BenNariah Z, Brice A. Hereditary spastic paraplegia with thin corpus callosum in Israel: homogeneous phenotype despite genetic heterogeneity. ***Arch Neurol.*** **2006**, 63: 1468-1471.

Bouslam N, Benomar A, Azzedine H, Bouhouche A, Namekawa M, **Klebe S**, Charon C, Durr A, Ruberg M, Brice A, Yahyaoui M, Stevanin G.Mapping of a new form of pure autosomal recessive spastic paraplegia (SPG28). ***Ann Neurol.*** **2005**, 57:567-71.

Stolze H, **Klebe S**, Baecker C, Zechlin C, Friege L, Pohle S, Deuschl G. Prevalence of gait disorders in hospitalized neurological patients. ***Mov Disord.*** **2005**, (1):89-94.

Stolze H, **Klebe S**, Zechlin C, Baecker C, Friege L, Deuschl G. Falls in frequent neurological diseases--prevalence, risk factors and aetiology. ***J Neurol.*** **2004**, 251: 79-84

Wenzelburger R, Kopper F, Frenzel A, Stolze H, **Klebe S**, Brossmann A, Kuhtz-Buschbeck J, Golge M, Illert M, Deuschl G. Hand coordination following capsular stroke. ***Brain.*** **2005**, 128:64-74.

Herzog J, Volkmann J, Krack P, Kopper F, Potter M, Lorenz D, Steinbach M, **Klebe** S, Hamel W, Schrader B, Weinert D, Muller D, Mehdorn HM, Deuschl G. Two-year follow-up of subthalamic deep brain stimulation in Parkinson's disease. ***Mov Disord.*** **2003**, 18(11):1332-7.

Stolze H, **Klebe** S, Petersen G, Raethjen J, Wenzelburger R, Witt K, Deuschl G. Typical features of cerebellar ataxic gait. ***J Neurol Neurosurg Psychiatry.*** **2002**, 73(3):310-2.

Wenzelburger R, Zhang BR, Pohle S, **Klebe** S, Lorenz D, Herzog J, Wilms H, Deuschl G, Krack P. Force overflow and levodopa-induced dyskinesias in Parkinson's disease. ***Brain.*** **2002**, 125(Pt 4):871-9.

Stolze H, **Klebe** S, Poepping M, Lorenz D, Herzog J, Hamel W, Schrader B, Raethjen J, Wenzelburger R, Mehdorn HM, Deuschl G, Krack P. Effects of bilateral subthalamic nucleus stimulation on parkinsonian gait. ***Neurology.*** **2001**, 57(1):144-6.

Books

Deuschl, G., Fietzek, U., Klebe, S. and Volkmann, V. (2003) Clinical neurophysiology and pathophysiology of Parkinson tremor. In: Hallet M (Ed). Handbook of Clinical Neurophysiology, Movement Disorders Vol 1, Elsevier

Klebe S, Bösch S, Bürk K. Ataxien (2011) In: Oertel W, Deuschl G, Poewe W (Eds.). Parkinson-Syndrome und andere Bewegungsstörungen. Erste Auflage, Thieme

Klebe S, Bürk K. Hereditäre Spastische Spinalparalyse (2011) In: Oertel W, Deuschl G, Poewe W (Eds.). Parkinson-Syndrome und andere Bewegungsstörungen. Erste Auflage, Thieme

Klebe, S, Anheim Mathieu. Genetics of Recessive Ataxias (2015) In: Susanne A. Schneider, Jose M. Tomas Bras (Eds.). Movement Disorder Genetics. Erste Auflage, Springer