

Publications Univ.-Prof. Dr. med. Joachim Weis

Contents

- List of 20 selected publications
- Chronological list of publications in scientific journals excluding reviews, guidelines and book chapters
- Reviews, guidelines, etc.
- Books, book chapters
- Habilitation treatise (*Habilitationsschrift*)
- Doctoral thesis (*Dissertation*)

20 selected publications

Weis J, Schröder JM: Differential effects of nerve, muscle and fat tissue on regenerating nerve fibers in vivo. *Muscle & Nerve* 12: 723-734, 1989

Weis J, Fine SM, David C, Savarirayan S, Sanes JR: Integration site-dependent expression of a transgene reveals special features of cells associated with neuromuscular junctions. *J Cell Biol* 113: 1385-1397, 1991

Weis J: Jun, fos, myoD, and myogenin proteins are increased in skeletal muscle fiber nuclei after denervation. *Acta Neuropathol* 87: 63-70, 1994

Porter BE, **Weis J**, Merlie JP, Sanes JR: A motoneuron-selective stop signal for neurite outgrowth in the synaptic protein, s-laminin. *Neuron* 14: 549-559, 1995

Weis J, Lie DC, Ragoß U, Züchner S, Karpati G, Yancopoulos GD, DiStefano PS: CNTF receptor alpha expression is increased in acute denervation atrophy of human skeletal muscle. *J Neuropathol Exp Neurol* 57: 850-857, 1998

Brunello AG, Weissenberger J, Kappeler A, Vallan C, Peters M, Rose-John S, **Weis J**: CNS alterations in interleukin-6/soluble interleukin-6 receptor alpha double transgenic mice. *Am J Pathol* 157: 1485-1493, 2000

Weissenberger J, Loeffler S, Afanasieva T, Aguzzi A, Kopf M, **Weis J**: IL-6 is required for glioma development in a mouse model. *Oncogene* 23: 3308-3316, 2004

Bremer J, Baumann F, Tiberi C, Wessig C, Fischer H, Schwarz P, Steele AD, Toyka KV, Nave KA, **Weis J**, Aguzzi A: Axonal prion protein is required for peripheral myelin maintenance. *Nature Neurosci.* 13(3): 310-318, 2010

Brauers E, Dreier A, Roos A, Wormland B, **Weis J***, Krüttgen A*. Differential effects of myopathy-associated caveolin 3-mutants on growth factor signaling. *Am J Pathol.* 177(1): 261-70, 2010 *Equal contribution

Weis J*, Katona I*, Müller-Newen G, Sommer C, Necula G, Hendrich C, Ludolph AC, Sperfeld A-D. Small fiber neuropathy in ALS patients. *Neurology.* 76(23): 2024-2029, 2011. *Equal contribution

Prause J*, Goswami A*, Katona I, Roos A, Schnizler M, Bushuven E, Dreier A, Buchkremer S, Johann S, Beyer C, Deschauer M, Troost D, **Weis J**. Altered localization, abnormal modification and loss of function of Sigma receptor-1 in amyotrophic lateral sclerosis. *Hum Mol Genet.* 22(8): 1581-1600, 2013. *Equal contr.

Roos A, Buchkremer S, Kollipara L, Labisch T, Gatz C, Zitzelsberger M, Brauers E, Nolte K, Schröder JM, Kirschner J, Jesse CM, Goebel HH, Goswami A, Zimmermann R, Zahedi RP, Senderek J, **Weis J**. Myopathy in Marinesco-Sjögren syndrome links endoplasmic reticulum chaperone dysfunction to nuclear envelope pathology. *Acta Neuropathol.* 127(5): 761-777, 2014

Filézac de L'Etang A, Maharjan N, Cordeiro Braña M, Ruegsegger C, Rehmann R, Goswami A, Roos A, Troost D, Schneider BL, **Weis J**, Saxena S. Marinesco-Sjögren syndrome protein SIL1 regulates motor neuron subtype-selective ER stress in ALS. *Nature Neurosci.* 18(2): 227-238, 2015

Khaminets A, Heinrich T, Mari M, Grumati P, Huebner AK, Akutsu M, Liebmann L, Stolz A, Nietzsche S, Koch N, Mauthe M, Katona I, Qualmann B, **Weis J**, Reggiori F, Kurth I, Hübner CA, Dikic I. Regulation of endoplasmic reticulum turnover by selective autophagy. *Nature.* 522 (7556): 354-358, 2015

Goswami A, Jesse C, Chandrasekar A, Bushuven E, Vollrath J, Dreser A, Katona I, Beyer C, Johann S, Feller A, Grond M, Wagner S, Nikolin S, Troost D, **Weis J**. Accumulation of STIM1 is associated with the degenerative muscle fibre phenotype in ALS and other neurogenic atrophies. *Neuropathol Appl Neurobiol.* 41(3): 304-18, 2015

Jesse CM, Bushuven E, Tripathi P, Chandrasekar A, Simon CM, Drepper C, Yamoah A, Dreser A, Katona I, Johann S, Beyer C, Wagner S, Grond M, Nikolin S, Anink J, Troost D, Sendtner M, Goswami A, **Weis J**. ALS-associated endoplasmic reticulum proteins in denervated skeletal muscle: Implications for motor neuron disease pathology. *Brain Pathol.* 27(6):781-794, 2017

Weis J, Claeys KG, Roos A, Azzedine H, Katona I, Schröder JM, Senderek J. Towards a functional pathology of hereditary neuropathies. *Acta Neuropathol.* 133(4): 493-515, 2017

Dreser A, Vollrath JT, Sechi A, Johann S, Roos A, Yamoah A, Katona I, Boholega S, Wiemuth D, Tian Y, Schmidt A, Vervoorts-Weber J, Dohmen M, Beyer C, Anink J, Aronica E, Troost D, **Weis J***, Goswami A*. The ALS-linked E102Q mutation in Sigma receptor-1 leads to ER stress-mediated defects in protein homeostasis and dysregulation of RNA binding proteins. *Cell Death Differ.* 10:1655-1671, 2017. *Equal contribution

Naumann M, Pal A, Goswami A, Lojewski X, Japtok J, Vehlow A, Naujock M, Günther R, Jin M, Stanslowsky N, Reinhardt P, Sternecker J, Frickenhaus M, Pan-Montojo F, Storkebaum E, Poser I, Freischmidt A, Weishaupt JH, Holzmann K, Troost D, Ludolph AC, Boeckers TM, Liebau S, Petri S, Cordes N, Hyman AA, Wegner F, Grill SW, **Weis J**, Storch A, Hermann A. Impaired DNA damage response signaling by FUS-NLS mutations leads to neurodegeneration and FUS aggregate formation. *Nat Commun.* 9(1): 335, 2018

Bouhy D, Juneja M, Katona I, Holmgren A, Asselbergh B, De Winter V, Hochepeid T, Goossens S, Haigh JJ, Libert C, Ceuterick-de Groote C, Irobi J, **Weis J**, Timmerman V. A knock-in/knock-out mouse model of HSPB8-associated distal hereditary motor neuropathy and myopathy reveals toxic gain-of-function of mutant Hspb8. *Acta Neuropathol.* 135(1): 131-148, 2018

Chronological list of publications in scientific journals excluding reviews, guidelines and comments

2019

338. Dafsari HS, Kocaturk NM, Daimagüler HS, Brunn A, Dötsch J, **Weis J**, Deckert M, Cirak S. Bi-allelic mutations in uncoordinated mutant number-45 myosin chaperone B are a cause for congenital myopathy. *Acta Neuropathol Commun.* 7(1): 211, 2019

337. Altinova H, Hammes S, Palm M, Achenbach P, Gerardo-Nava J, Deumens R, Führmann T, van Neerven SGA, Hermans E, **Weis J**, Brook G. Dense fibroadhesive scarring and poor blood vessel-maturation hamper the integration of implanted collagen scaffolds in an experimental model of spinal cord injury. *Biomed Mater.* 2019 Dec 4. [Epub ahead of print]

336. Rudnik-Schöneborn S, Huemer M, **Weis J**, Sauer E, Meng G. Early onset facioscapulohumeral muscular dystrophy - Long-term follow-up of a patient with total facial diplegia. *Neuromuscul Disord.* 29(12): 973-976, 2019

335. Kulesa M, Weyer-Menkhoff I, Viergutz L, Kornblum C, Claeys KG, Schneider I, Plöckinger U, Young P, Boentert M, Vielhaber S, Mawrin C, Bergmann M, **Weis J**, Ziagaki A, Stenzel W, Deschauer M, Nolte D, Hahn A, Schoser B, Schänzer A. An integrative correlation of myopathology, phenotype, and genotype in late onset Pompe disease. *Neuropathol Appl Neurobiol.* 2019 Sep 23. doi: 10.1111/nan.12580. [Epub ahead of print] PubMed PMID: 31545528.

334. Krasselt M, Schober R, **Weis J**, Baum P, Baerwald CGO, Seifert O. A Primary Myopathy Complicating Long-lasting Polymyalgia Rheumatica. *J Clin Rheumatol.* 2019 Oct 11. doi: 10.1097/RHU.0000000000001220. [Epub ahead of print] PubMed PMID: 31609812.

333. Stengel H, Vural A, Brunder AM, Heinius A, Appeltshauser L, Fiebig B, Giese F, Dresel C, Papagianni A, Birklein F, **Weis J**, Huchtemann T, Schmidt C, Körtvelyessy, Villmann C, Meinel E, Sommer C, Leyboldt F, Doppler K. Anti-pan-neurofascin IgG3 as a marker of fulminant autoimmune neuropathy. *Neurol Neuroimmunol Neuroinflamm.* 2019 Aug 16;6(5). pii: e603. PubMed PMID: 31454780

332. Farschtschi SC, Kluwe L, Schön G, Friedrich RE, Matschke J, Glatzel M, **Weis J**, Hagel C, Mautner VF. Distinctive low epidermal nerve fiber density in schwannomatosis patients provides a major parameter for diagnosis and differential diagnosis. *Brain Pathol.* 2019 Aug 19. doi: 10.1111/bpa.12780. [Epub ahead of print] PubMed PMID: 31424590.

331. Romeike BFM, Becker K, Großkreutz J, Schulz S, **Weis J**, Cirak S. A family with limb girdle muscular dystrophy type 1B and multiple exostoses. *Clin Neuropathol.* 38(5):225-232, 2019

330. Gatz C, Hathazi D, Münchberg U, Buchkremer S, Labisch T, Munro B, Horvath R, Töpf A, **Weis J**, Roos A. Identification of Cellular Pathogenicity Markers for SIL1 Mutations Linked to Marinesco-Sjögren Syndrome. *Front Neurol.* 10: 562, 2019

329. Joseph S, Vingill S, Jahn O, Fledrich R, Werner HB, Katona I, Möbius W, Mitkovski M, Huang Y, **Weis J**, Sereda MW, Schulz JB, Nave KA, Stegmüller J. Myelinating gliaspecific deletion of Fbxo7 in mice triggers axonal degeneration in the central nervous system together with peripheral neuropathy. *J Neurosci.* 39(28): 5606-5626, 2019

327. Ross JA, Levy Y, Ripolone M, Kolb JS, Turmaine M, Holt M, Lindqvist J, Claeys KG, **Weis J**, Monforte M, Tasca G, Moggio M, Figeac N, Zammit PS, Jungbluth H, Fiorillo C, Vissing J, Witting N, Granzier H, Zanoteli E, Hardeman EC, Wallgren-Pettersson C, Ochala J. Impairments in contractility and cytoskeletal organisation cause nuclear defects in nemaline myopathy. *Acta Neuropathol.* 138(3): 477-495, 2019

327. Fischbach F, Nedelcu J, Leopold P, Zhan J, Clarner T, Nellessen L, Beißel C, van Heuvel Y, Goswami A, **Weis J**, Denecke B, Schmitz C, Hochstrasser T, Nyamoya S, Victor M, Beyer C, Kipp M. Cuprizone-induced graded oligodendrocyte vulnerability is regulated by the transcription factor DNA damage-inducible transcript 3. *Glia.* 67(2): 263-276, 2019

326. Marrone L, Drexler HCA, Wang J, Tripathi P, Distler T, Heisterkamp P, Anderson EN, Kour S, Moraiti A, Maharana S, Bhatnagar R, Belgard TG, Tripathy V, Kalmbach N, Hosseinzadeh Z, Crippa V, Abo-Rady M, Wegner F, Poletti A, Troost D, Aronica E, Busskamp V, **Weis J**, Pandey UB, Hyman AA, Alberti S, Goswami A, Sternecker J. FUS pathology in ALS is linked to alterations in multiple ALS-associated proteins and rescued by drugs stimulating autophagy. *Acta Neuropathol.* 138(1): 67-84, 2019

325. Altinova H*, Hammes S*, Palm M, Gerardo-Nava J, Achenbach P, Deumens R, Hermans E, Führmann T, Boecker A, van Neerven S, Bozkurt A, **Weis J**, Brook GA. Fibroadhesive scarring of grafted collagen scaffolds interferes with implant–host neural tissue integration and bridging in experimental spinal cord injury. *Regenerative Biomaterials* 6(2): 75-87, 2019, *Equal contribution

324. Karsai G, Kraft F, Haag N, Korenke GC, Hänisch B, Othman A, Suriyanarayanan S, Steiner R, Knopp C, Mull M, Bergmann M, Schröder JM, **Weis J**, Elbracht M, Begemann M, Hornemann T, Kurth I. DEGS1-associated aberrant sphingolipid metabolism impairs nervous system function in humans. *J Clin Invest.* 129(3): 1229-1239, 2019

323. Kölbel H, Abicht A, Schwartz O, Katona I, Paulus W, Neuen-Jacob E, **Weis J**, Schara U. Characteristic clinical and ultrastructural findings in nesprinopathies. *Eur J Paediatr Neurol.* 23(2): 254-261, 2019

322. Phan V, Cox D, Cipriani S, Spendiff S, Buchkremer S, O'Connor E, Horvath R, Goebel HH, Hathazi D, Lochmüller H, Straka T, Rudolf R, **Weis J**, Roos A. SIL1 deficiency causes degenerative changes of peripheral nerves and neuromuscular junctions in fish, mice and human. *Neurobiol Dis.* 124: 218-229, 2019

2018

321. De Paepe B, Zschüntzsch J, Šokčević T, **Weis J**, Schmidt J, De Bleecker JL. Induction of Osmolyte Pathways in Skeletal Muscle Inflammation: Novel Biomarkers for Myositis. *Front Neurol.* 9: 846, 2018

320. Fischbach F, Nedelcu J, Leopold P, Zhan J, Clarner T, Nellessen L, Beißel C, van Heuvel Y, Goswami A, **Weis J**, Denecke B, Schmitz C, Hochstrasser T, Nyamoya S,

Victor M, Beyer C, Kipp M. Cuprizone-induced graded oligodendrocyte vulnerability is regulated by the transcription factor DNA damage-inducible transcript 3. *Glia*. 2018 Dec 3. [Epub ahead of print]

319. Forsberg KME, Zhang Y, Reiners J, Ander M, Niedermayer A, Fang L, Neugebauer H, Kassubek J, Katona I, **Weis J**, Ludolph AC, Del Tredici K, Braak H, Yilmazer-Hanke D. Endothelial damage, vascular bagging and remodeling of the microvascular bed in human microangiopathy with deep white matter lesions. *Acta Neuropathol Commun*. 6(1): 128, 2018

318. Herbelet S, De Vlieghere E, Gonçalves A, De Paepe B, Schmidt K, Nys E, Weynants L, **Weis J**, Van Peer G, Vandesomepele J, Schmidt J, De Wever O, De Bleecker JL. Localization and Expression of Nuclear Factor of Activated T-Cells 5 in Myoblasts Exposed to Pro-inflammatory Cytokines or Hyperosmolar Stress and in Biopsies from Myositis Patients. *Front Physiol*. 9: 126, 2018

317. Vill K, Müller-Felber W, Gläser D, Kuhn M, Teusch V, Schreiber H, **Weis J**, Klepper J, Schirmacher A, Blaschek A, Wiessner M, Strom TM, Dräger B, Hofmeister-Kiltz K, Tacke M, Gerstl L, Young P, Horvath R, Senderek J. SACS variants are a relevant cause of autosomal recessive hereditary motor and sensory neuropathy. *Hum Genet*. 137(11-12): 911-919, 2018

316. Heinen MC, Babler A, **Weis J**, Elsas J, Nolte K, Kipp M, Jahnen-Dechent W, Häusler M. Fetuin-A protein distribution in mature inflamed and ischemic brain tissue. *PLoS One*. 13(11): e0206597, 2018

314. Quade A, **Weis J**, Kurth I, Rolke R, Bienert M, Schradling S, Rohrman D, Yüksel Z, Häusler M. Microangiopathy and mild mixed neuromyopathic alterations in a patient with homozygous PIEZO-2 mutation. *Neuromuscul Disord*. 28(12):1006-1011, 2018

315. Vettermann FJ, Felsberg J, Reifenberger G, Hasselblatt M, Forbrig R, Berding G, la Fougère C, Galldiks N, Schittenhelm J, **Weis J**, Albert NL, Schüller U. Characterization of Diffuse Gliomas With Histone H3-G34 Mutation by MRI and Dynamic 18F-FET PET. *Clin Nucl Med*. 43(12): 895-898, 2018

314. Kork F, Jankowski J, Goswami A, **Weis J**, Brook G, Yamoah A, Anink J, Aronica E, Fritz S, Huck C, Schipke C, Peters O, Tepel M, Noels H, Jankowski V. Golgin A4 in CSF and granulovacuolar degenerations of Alzheimer patients. *Neurology*. 91(19): e1799-e1808, 2018

313. González Coraspe JA, **Weis J**, Anderson ME, Münchberg U, Lorenz K, Buchkremer S, Carr S, Zahedi RP, Brauers E, Michels H, Sunada Y, Lochmüller H, Campbell KP, Freier E, Hatzazi D, Roos A. Biochemical and pathological changes result from mutated Caveolin-3 in muscle. *Skelet Muscle*. 8(1): 28, 2018

312. Arndt P, Leistner ND, Neuss S, Kaltbeitzel D, Brook GA, Grosse J. Artificial urine and FBS supplemented media in cytocompatibility assays for PLGA-PEG-based intravesical devices using the urothelium cell line UROtsa. *J Biomed Mater Res B Appl Biomater*. 106(6): 2140-2147, 2018

311. Laššuthová P, Vill K, Erdem-Ozdamar S, Schröder JM, Topaloglu H, Horvath R, Müller-Felber W, Bansagi B, Schlotter-Weigel B, Gläser D, Neupauerová J, Sedláčková L, Staněk D, Mazanec R, **Weis J**, Seeman P, Senderek J. Novel SBF2 mutations and

clinical spectrum of Charcot-Marie-Tooth neuropathy type 4B2. *Clin Genet.* 94(5): 467-472, 2018

310. Boecker AH, Bozkurt A, Kim BS, Altinova H, Tank J, Deumens R, Tolba R, **Weis J**, Brook GA, Pallua N, van Neerven SGA. Cell-enrichment with olfactory ensheathing cells has limited local extra beneficial effects on nerve regeneration supported by the nerve guide Perimaix. *J Tissue Eng Regen Med.* 12(11): 2125-2137, 2018

309. Wunderlich G, Brunn A, Daimagüler HS, Bozoglu T, Fink GR, Lehmann HC, **Weis J**, Cirak S. Long term history of a congenital core-rod myopathy with compound heterozygous mutations in the Nebulin gene. *Acta Myol.* 37(2): 121-127, 2018

308. Fledrich R, Abdelaal T, Rasch L, Bansal V, Schütza V, Brügger B, Lüchtenborg C, Prukop T, Stenzel J, Rahman RU, Hermes D, Ewers D, Möbius W, Ruhwedel T, Katona I, **Weis J**, Klein D, Martini R, Brück W, Müller WC, Bonn S, Bechmann I, Nave KA, Stassart RM, Sereda MW. Targeting myelin lipid metabolism as a potential therapeutic strategy in a model of CMT1A neuropathy. *Nat Commun.* 9(1): 3025, 2018

307. Lohmann P, Piroth MD, Sellhaus B, **Weis J**, Geisler S, Oros-Peusquens AM, Mohlberg H, Amunts K, Shah NJ, Galldiks N, Langen KJ. Correlation of Dynamic O-(2-[18F]Fluoroethyl)-L-Tyrosine Positron Emission Tomography, Conventional Magnetic Resonance Imaging, and Whole-Brain Histopathology in a Pretreated Glioblastoma: A Postmortem Study. *World Neurosurg.* 119: e653-e660, 2018

306. Issop Y, Hathazi D, Khan MM, Rudolf R, **Weis J**, Spendiff S, Slater CR, Roos A, Lochmüller H. GFPT1 deficiency in muscle leads to myasthenia and myopathy in mice. *Hum Mol Genet.* 27(18): 3218-3232, 2018

305. Nikoubashman O, Heringer S, Feher K, Brockmann MA, Sellhaus B, Dreser A, Kurtenbach K, Pjontek R, Jockenhövel S, **Weis J**, Kießling F, Gries T, Wiesmann M. Development of a Polymer-Based Biodegradable Neurovascular Stent Prototype: A Preliminary In Vitro and In Vivo Study. *Macromol Biosci.* 18(7): e1700292, 2018

304. Yuan X, Klein D, Kerscher S, West BL, **Weis J**, Katona I, Martini R. Macrophage depletion ameliorates peripheral neuropathy in aging mice. *J Neurosci.* 38(19): 4610-4620, 2018

303. Dusanic M, Dekomien G, Lücke T, Vorgerd M, **Weis J**, Epplen JT, Köhler C, Hoffjan S. Novel Nonsense Mutation in SLC39A13 Initially Presenting as Myopathy: Case Report and Review of the Literature. *Mol Syndromol.* 9(2): 100-109, 2018

302. 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.* 89: 817-821, 2018

301. Lehmann S, Esch E, Hartmann P, **Goswami A**, Nikolin S, **Weis J**, Beyer C, Johann S. Expression profile of pattern recognition receptors in skeletal muscle of SOD1((G93A)) amyotrophic lateral sclerosis (ALS) mice and sporadic ALS patients. *Neuropathol Appl Neurobiol.* 37: 121-127, 2018

300. Radke J, Koll R, Preuße C, Pehl D, Todorova K, Schönemann C, Allenbach Y, Aronica E, de Visser M, Heppner FL, **Weis J**, Doostkam S, Maisonobe T, Benveniste O, Goebel HH, Stenzel W. Architectural B-cell organization in skeletal muscle identifies subtypes of dermatomyositis. *Neurol Neuroimmunol Neuroinflamm.* 5(3): e451, 2018

299. Naumann M, Pal A, Goswami A, Lojewski X, Japtok J, Vehlow A, Naujock M, Günther R, Jin M, Stanslawski N, Reinhardt P, Sternecker J, Frickenhaus M, Pan-Montojo F, Storkebaum E, Poser I, Freischmidt A, Weishaupt J, Holzmann K, Troost D, Ludolph A, Boeckers TM, Liebau S, Petri S, Cordes N, Hyman A, Wegner F, Grill S, **Weis J**, Storch A, Hermann A. Impaired DNA damage response signaling by FUS-NLS mutations leads to neurodegeneration and aggregation formation. *Nature Commun.* 2018; 9: 335.

298. Brenner D, Yilmaz R, Müller K, Grehl T, Petri S, Meyer T, Grosskreutz J, Weydt P, Ruf W, Neuwirth C, Weber M, Pinto S, Claeys KG, Schrank B, Jordan B, Knehr A, Günther K, Hübers A, Zeller D; German ALS network MND-NET, Kubisch C, Jablonka S, Sendtner M, Klopstock T, de Carvalho M, Sperfeld A, Borck G, Volk AE, Dorst J, **Weis J**, Otto M, Schuster J, Del Tredici K, Braak H, Danzer KM, Freischmidt A, Meitinger T, Strom TM, Ludolph AC, Andersen PM, Weishaupt JH. Hot-spot KIF5A mutations cause familial ALS. *Brain.* 141: 688-697, 2018

297. Bouhy D, Juneja M, Katona I, Holmgren A, Asselbergh B, De Winter V, Hochepeid T, Goossens S, Haigh JJ, Libert C, Ceuterick-de Groote C, Irobi J, **Weis J**, Timmerman V. A knock-in/knock-out mouse model of HSPB8-associated distal hereditary motor neuropathy and myopathy reveals toxic gain-of-function of mutant Hspb8. *Acta Neuropathol.* 135(1): 131-148, 2018

296. Breuer T, Bleilevens C, Rossaint R, Marx G, Gehrenkemper J, Dierksen H, Delpierre A, **Weis J**, Gayan-Ramirez G, Bruells CS. Dexmedetomidine Impairs Diaphragm Function and Increases Oxidative Stress but Does Not Aggravate Diaphragmatic Atrophy in Mechanically Ventilated Rats. *Anesthesiology.* 128(4):784-795, 2018

295. Labisch T, Buchkremer S, Phan V, Kollipara L, Gatz C, Lentz C, Nolte K, Vervoorts-Weber J, González Coraspe JA, Sickmann A, Carr S, Zahedi RP, **Weis J**, Roos A. Tracking effects of SIL1 increase: taking a closer look beyond the consequences of elevated expression level. *Mol Neurobiol.* 2018; 55(3): 2524-2546

294. Quade A, Wiesmann M, **Weis J**, Kurth I, Jalaie H, Rohrbach M, Häusler M. Stroke in Ehlers-Danlos Syndrome Kyphoscoliotic Type: Dissection or Vasculitis? *Pediatr Neurol.* 28(12):1006-1011, 2018

2017

293. Wang H, Salter CG, Refai O, Hardy H, Barwick KES, Akpulat U, Kvarnung M, Chioza BA, Harlalka G, Taylan F, Sejersen T, Wright J, Zimmerman HH, Karakaya M, Stüve B, **Weis J**, Schara U, Russell MA, Abdul-Rahman OA, Chilton J, Blakely RD, Baple EL, Cirak S, Crosby AH. Choline transporter mutations in severe congenital myasthenic syndrome disrupt transporter localization. *Brain.* 2017;140(11):2838-2850.

292. Bozkurt A, Claeys KG, Schradning S, Rödler JV, Altinova H, Schulz JB, **Weis J**, Pallua N, van Neerven SGA. Clinical and biometrical 12-month follow-up in patients after

reconstruction of the sural nerve biopsy defect by the collagen-based nerve guide Neuromaix. *Eur J Med Res.* 22(1): 34, 2017

291. Dreser A, Vollrath JT, Sechi A, Johann S, Roos A, Yamoah A, Katona I, Boholega S, Wiemuth D, Tian Y, Schmidt A, Vervoorts-Weber J, Dohmen M, Beyer C, Anink J, Aronica E, Troost D, **Weis J***, Goswami A*. The ALS-linked E102Q mutation in Sigma receptor-1 leads to ER stress-mediated defects in protein homeostasis and dysregulation of RNA binding proteins. *Cell Death Differ.* 10 24(10):1655-1671, 2017. *equal contribution

290. Cordts I, Bodart N, Hartmann K, Karagiorgou K, Tzartos JS, Mei L, Reimann J, Van Damme P, Rivner MH, Vigneron A, **Weis J**, Schulz JB, Tzartos SJ, Claeys KG. Screening for lipoprotein receptor-related protein 4-, agrin-, and titin-antibodies and exploring the autoimmune spectrum in myasthenia gravis. *J Neurol.* 264(6): 1193-1203, 2017

289. Schnitzler LJ, Schreckenbach T, Nadaj-Pakleza A, Stenzel W, Rushing EJ, Van Damme P, Ferbert A, Petri S, Hartmann C, Bornemann A, Meisel A, Petersen JA, Tousseyn T, Thal DR, Reimann J, De Jonghe P, Martin JJ, Van den Bergh PY, Schulz JB, **Weis J**, Claeys KG. Sporadic late-onset nemaline myopathy: clinico-pathological characteristics and review of 76 cases. *Orphanet J Rare Dis.* 12(1): 86. doi: 10.1186/s13023-017-0640-2, 2017

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287. Kollipara L, Buchkremer S, Coraspe JAG, Hathazi D, Senderek J, **Weis J**, Zahedi RP, Roos A. In-depth phenotyping of lymphoblastoid cells suggests selective cellular vulnerability in Marinesco-Sjögren syndrome. *Oncotarget.* 28;8(40):68493-68516, 2017

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