

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

### Contents

- 20 selected publications
- Chronological list of publications in scientific journals excluding reviews, guidelines and book chapters
- Reviews, guidelines, etc.
- Books, book chapters
- Others: 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

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

Mair D, Biskup S, Kress W, Abicht A, Brück W, Zechel S, Knop KC, Koenig FB, Tey S, Nikolin S, Eggermann K, Kurth I, Ferbert A, **Weis J**. Differential diagnosis of vacuolar myopathies in the NGS era. *Brain Pathol.* 30(5): 877-896, 2020

Tripathi P, Guo H, Dreser A, Yamoah A, Sechi A, Jesse C, Katona I, Doukas P, Nikolin S, Ernst S, Aronica E, Troost D, Glass H, Hermann A, Steinbusch H, Feller A, Bergmann M, Jaarsma D\*, **Weis J\***, Goswami A\*. Pathomechanisms of ALS8: Altered autophagy and defective RNA binding protein (RBP) homeostasis due to the VAPB-P56S mutation. *Cell Death Dis.* 2021 12(5): 466, 2021. \*equal contribution

Foronda H, Fu Y, Covarrubias-Pinto A, Bocker HT, González A, Seemann E, Franzka P, Bock A, Bhaskara RM, Liebmann L, Hoffmann ME, Katona I, Koch N, **Weis J**, Kurth I, Gleeson JG, Reggiori F, Hummer G, Kessels MM, Qualmann B, Mari M, Dikić I, Hübner CA. Heteromeric clusters of ubiquitinated ER-shaping proteins drive ER-phagy. *Nature.* 2023 May 24. doi: 10.1038/s41586-023-06090-9. Epub ahead of print. PMID: 37225994.

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

### 2023

370. Foronda H, Fu Y, Covarrubias-Pinto A, Bocker HT, González A, Seemann E, Franzka P, Bock A, Bhaskara RM, Liebmann L, Hoffmann ME, Katona I, Koch N, **Weis J**, Kurth I, Gleeson JG, Reggiori F, Hummer G, Kessels MM, Qualmann B, Mari M, Dikić I, Hübner CA. Heteromeric clusters of ubiquitinated ER-shaping proteins drive ER-phagy. *Nature*. 2023 May 24. doi: 10.1038/s41586-023-06090-9. Epub ahead of print. PMID: 37225994.

369. Roos A, van der Ven PFM, Alrohaif H, Kölbel H, Heil L, Della Marina A, **Weis J**, Aßent M, Beck-Wödl S, Barresi R, Töpf A, O'Connor K, Sickmann A, Kohlschmidt N, El Gizouli M, Meyer N, Daya N, Grande V, Bois K, Kaiser FJ, Vorgerd M, Schröder C, Schara-Schmidt U, Gangfuss A, Evangelista T, Röbisch L, Hentschel A, Grüneboom A, Fuerst DO, Kuechler A, Tzschach A, Depienne C, Lochmüller H. Bi-allelic variants of FILIP1 cause congenital myopathy, dysmorphism and neurological defects. *Brain*. 2023 May 10:awad152. doi: 10.1093/brain/awad152. Epub ahead of print. PMID: 37163662.

368. Bremer J, Friemann J, von Stillfried S, Boor P, **Weis J**. Reduced T-cell densities in cranial nerves of patients who died with SARS-CoV-2 infection. *Acta Neuropathologica Communications* 11(1): 10, 2023

367. Igharo D, Thiel JC, Rolke R, Akkaya M, **Weis J**, Katona I, Schulz JB, Maier A. Skin biopsy reveals generalized small fibre neuropathy in hypermobile Ehlers-Danlos syndromes. *Eur J Neurol*. 30(3): 719-728, 2023

366. Yamoah A, Tripathi P, Guo H, Scheve L, Walter P, Johnen S, Müller F, **Weis J\***, Goswami A\*. Early Alterations of RNA Binding Protein (RBP) Homeostasis and ER Stress-Mediated Autophagy Contributes to Progressive Retinal Degeneration in the *rd10* Mouse Model of Retinitis Pigmentosa (RP). *Cells*. 2023 Apr 6;12(7):1094. doi: 10.3390/cells12071094. PMID: 37048167; PMCID: PMC10092976. \*equal contribution

365. Franzka P, Schüler SC, Kentache T, Storm R, Bock A, Katona I, **Weis J**, Buder K, Kaether C, Hübner CA. Impact of Hypermannosylation on the Structure and Functionality of the ER and the Golgi Complex. *Biomedicines*. 2023 Jan 6;11(1):146. doi: 10.3390/biomedicines11010146. PMID: 36672654; PMCID: PMC9856158.

### 2022

364. von Stillfried S, Bülow RD, Röhrig R, Boor P; German Registry of COVID-19 Autopsies (DeRegCOVID), DeRegCOVID Collaborators. First report from the German COVID-19 autopsy registry. *Lancet Reg Health Eur*. 2022 Feb 18; 15: 100330. doi: 10.1016/j.lanepe.2022.100330. eCollection 2022 Apr. PMID: 35531493

363. von Stillfried S, Bülow RD, Röhrig R, Meybohm P, Boor P; German Registry of COVID-19 Autopsies (DeRegCOVID), DeRegCOVID Collaborators#. Intracranial hemorrhage in COVID-19 patients during extracorporeal membrane oxygenation for acute respiratory failure: a nationwide register study report. *Crit Care*. 2022 Mar 28; 26(1): 83. doi: 10.1186/s13054-022-03945-x. PMID: 35346314

362. Pilotto F, Schmitz A, Maharjan N, Diab R, Odriozola A, Tripathi P, Yamoah A, Scheidegger O, Oestmann A, Dennys CN, Sinha Ray S, Rodrigo R, Kolb S, Aronica E, Di Santo S, Widmer HR, Charlet-Berguerand N, Selvaraj BT, Chandran S, Meyer K, Zuber B, Goswami A, **Weis J**, Saxena S. PolyGA targets the ER stress-adaptive response by impairing GRP75 function at the MAM in C9ORF72-ALS/FTD. *Acta Neuropathol* 2022 144(5):939-966, 2022

361. Maier A, Kapfenberger R, Katona I, **Weis J**, Schulz JB, Rolke R. Nonregional small fibre neuropathy in cases of autoimmune autonomic neuropathy. *J Neurol*. 2022 Sep 9. Epub ahead of print.

360. Altinova H, Achenbach P, Palm M, Katona I, Hermans E, Clusmann H, **Weis J**, Brook GA. Characterization of a Novel Aspect of Tissue Scarring Following Experimental Spinal Cord Injury and the Implantation of Bioengineered Type-I Collagen Scaffolds in the Adult Rat: Involvement of Perineurial-Like Cells? *Int. J. Mol. Sci.* 23(6), 3221, 2022

359. Dohrn MF, Heller C, Zengeler D, Obermaier CD, Biskup S, **Weis J**, Nikolin S, Claeys KG, Schöne U, Beijer D, Winter N, Achenbach P, Gess B, Schulz JB, Mula-hasanovic L. Heterozygous POLG variant Ser1181Asn co-segregating in a family with autosomal dominant axonal neuropathy, proximal muscle fatigability, ptosis, and ragged red fibers. *Neurol Res Pract.* 4(1): 5, 2022

358. Koeppen S, Hense J, Nolte KW, **Weis J**. Immune-mediated neuropathy related to bortezomib in a patient with multiple myeloma. *Arch Pathol Clin Res.* 6: 001-004, 2022

## 2021

357. Ferreira N, Richner M, van der Laan A, Bergholdt Jul Christiansen I, Vægter CB, Nyengaard JR, Halliday GM, **Weis J**, Giasson BI, Mackenzie IR, Jensen PH, Jan A. Prodromal neuroinvasion of pathological  $\alpha$ -synuclein in brainstem reticular nuclei and white matter lesions in a model of  $\alpha$ -synucleinopathy. *Brain Commun.* 3(2): fcab104, 2021

356. Della Marina A, Arlt A, Schara-Schmidt U, Depienne C, Gangfuß A, Kölbl H, Sickmann A, Freier E, Kohlschmidt N, Hentschel A, **Weis J**, Czech A, Grüneboom A, Roos A. Phenotypical and Myopathological Consequences of Compound Heterozygous Missense and Nonsense Variants in SLC18A3. *Cells.* 10(12):3481, 2021

355. Hummel C, Leylamian O, Pösch A, **Weis J**, Aronica E, Beyer C, Johann S. Expression and cell type-specific localization of inflammasome sensors in the spinal cord of SOD1(G93A) mice and sALS patients. *Neuroscience.* 463: 288-302, 2021

354. Peters S, Kuespert S, Wirkert E, Heydn R, Jurek B, Johannesen S, Hsam O, Korte S, Ludwig FT, Mecklenburg L, Mrowetz H, Altendorfer B, Poupardin R, Petri S, Thal DR, Hermann A, Weishaupt JH, **Weis J**, Aksoylu IS, Lewandowski SA, Aigner L, Bruun TH, Bogdahn U. Reconditioning the Neurogenic Niche of Adult Non-human Primates by Antisense Oligonucleotide-Mediated Attenuation of TGF $\beta$  Signaling. *Neurotherapeutics.* 18(3):1963-1979, 2021

353. Andereggen L, Mariani L, Beck J, Andres RH, Gralla J, Luedi MM, **Weis J**, Christ E. Lateral one-third gland resection in Cushing patients with failed adenoma identification leads to low remission rates: long-term observations from a small, single-center cohort. *Acta Neurochir (Wien)*. 47(6):840-855, 2021, 2021

352. Kohlschmidt N, Elbracht M, Czech A, Häusler M, Phan V, Töpf A, Huang KT, Bartok A, Eggermann K, Zippel S, Eggermann T, Freier E, Groß C, Lochmüller H, Horvath R, Hajnóczky G, **Weis J**, Roos A. Molecular pathophysiology of human MICU1-deficiency. *Neuropathol Appl Neurobiol*. 47(6):840-855, 2021

351 Freischmidt A, Goswami A, Limm K, Zimyanin VL, Demestre M, Glaß H, Holzmann K, Helferich AM, Brockmann SJ, Tripathi P, Yamoah A, Poser I, Oefner PJ, Böckers TM, Aronica E, Ludolph AC, Andersen PM, Hermann A, **Weis J**, Reinders J, Danzer KM, Weishaupt JH. A serum microRNA sequence reveals fragile X protein pathology in amyotrophic lateral sclerosis. *Brain*. 144(4):1214-1229, 2021

350. Franzka P, Henze H, Jung MJ, Schüler SC, Mittag S, Biskup K, Liebmann L, Kentache T, Morales J, Martínez B, Katona I, Herrmann T, Huebner AK, Hennings JC, Groth S, Gresing LJ, Horstkorte R, Marquardt T, **Weis J**, Kaether C, Mutchinick OM, Ori A, Huber O, Blanchard V, von Maltzahn J, Hübner CA. GMPPA defects cause a neuro-muscular disorder with  $\alpha$ -dystroglycan hyperglycosylation. *J Clin Invest*. 131(9):e139076, 2021

349. Tripathi P, Guo H, Dreser A, Yamoah A, Sechi A, Jesse C, Katona I, Doukas P, Nikolin S, Ernst S, Aronica E, Troost D, Glass H, Hermann A, Steinbusch H, Feller A, Bergmann M, Jaarsma D\*, **Weis J**\*, Goswami A\*. Pathomechanisms of ALS8: Altered autophagy and defective RNA binding protein (RBP) homeostasis due to the VAPB-P56S mutation. *Cell Death Dis*. 2(5):466, 2021. \*equal contribution

348. Deschauer M, Hengel H, Rupprich K, Kreiß M, Schlotter-Weigel B, Grimm M, Admard J, Schneider I, Alhaddad B, Gazou A, Sturm M, Vorgerd M, Balousha G, Balousha O, Falna M, Kirschke JS, Kornblum C, Jordan B, Kraya T, Strom TM, **Weis J**, Schöls L, Schara U, Zierz S, Riess O, Meitinger T, Haack TB. Bi-allelic truncating mutations in VWA1 cause neuromyopathy. *Brain* 144(2):574-583, 2021

347. Lausberg E, Gießelmann S, Dewulf JP, Wiame E, Holz A, Salvarinova R, Van Karnebeek C, Klemm P, Ohl K, Mull M, Braunschweig T, **Weis J**, Sommer C, Demuth S, Haase C, Debray F-G, Libioulle C, Choukair D, Oommen PT, Borkhardt A, Surowy H, Wiczorek D, Meyer R, Eggermann T, Begemann M, Van Schaffingen E, Häusler M, Tenbrock K, van den Heuvel L, Elbracht M, Kurth, Kraft F. A human multisystem disorder with autoinflammation, leukoencephalopathy and hepatopathy is caused by mutations in C2orf69. *J Clin Invest*. 131(12):e143078, 2021

346. Krasselt M, Schober R, **Weis J**, Baum P, Baerwald CGO, Seifert O. A Primary Myopathy Complicating Long-lasting Polymyalgia Rheumatica. *J Clin Rheumatol*. 27(1): e28-e29, 2021

## 2020

345. Kölbel H, Roos A, van der Ven PFM, Evangelista T, Nolte K, Johnson K, Töpf A, Wilson M, Kress W, Sickmann A, Straub V, Kollipara L, **Weis J**, Furst DO, Schara U.

First clinical and myopathological description of a myofibrillar myopathy with congenital onset and homozygous mutation in FLNC. *Hum Mutat.* 41(9): 1600-1614, 2020

344. Rabenstein M, **Weis J**, Abicht A, Fink GR, Lehmann HC, Wunderlich G. [Multiple acyl-CoA dehydrogenase deficiency/glutaric aciduria type 2: difficult diagnosis, easy to treat]. *Nervenarzt.* 91(4): 349-352, 2020

343. Mair D, Biskup S, Kress W, Abicht A, Brück W, Zechel S, Knop KC, Koenig FB, Tey S, Nikolin S, Eggermann K, Kurth I, Ferbert A, **Weis J**. Differential diagnosis of vacuolar myopathies in the NGS era. *Brain Pathol.* 30(5): 877-896, 2020

342. Mathis S, Vallat JM, **Weis J**. When botany inspired pathology of the peripheral nervous system. *Neurology.* 95(12): 532-536, 2020

341. Hedberg-Oldfors C, Meyer R, Nolte K, Abdul Rahim Y, Lindberg C, Karason K, Thuestad IJ, Visuttijai K, Geijer M, Begemann M, Kraft F, Lausberg E, Hitpass L, Götzl R, Luna EJ, Lochmüller H, Koschmieder S, Gramlich M, Gess B, Elbracht M, **Weis J**, Kurth I, Oldfors A, Knopp C. Loss of supervillin causes myopathy with myofibrillar disorganization and autophagic vacuoles. *Brain.* 143(8): 2406-2420, 2020

340. Yamoah A, Tripathi P, Sechi A, Köhler C, Guo H, Chandrasekar A, Nolte KW, Wruck CJ, Katona I, Anink J, Troost D, Aronica E, Steinbusch H, **Weis J\***, Goswami A\*. Aggregates of RNA Binding Proteins and ER Chaperones Linked to Exosomes in Granulovacuolar Degeneration of the Alzheimer's Disease Brain. *J Alzheimers Dis.* 75(1): 139-156, 2020. \*equal contribution

339. Yilmazer-Hanke D, Mayer T, Müller HP, Neugebauer H, Abaei A, Scheuerle A, **Weis J**, Forsberg KME, Althaus K, Meier J, Ludolph AC, Del Tredici K, Braak H, Kasubek J, Rasche V. Histological correlates of postmortem ultra-high-resolution single-section MRI in cortical cerebral microinfarcts. *Acta Neuropathol Commun.* 8(1): 33, 2020

338. 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.* 15(1): 015012, 2020

337. 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.* 30(2): 386-391, 2020

336. 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* 46(4): 359-374, 2020

335. 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.* 13;15(1):015012, 2020

## 2019

334. 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

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

332. Stengel H, Vural A, Brunder AM, Heinius A, Appeltshauer 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.* 6(5). pii: e603, 2019

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 glia-specific deletion of Fbxo7 in mice triggers axonal degeneration in the central nervous system together with peripheral neuropathy. *J Neurosci.* 39(28): 5606-5626, 2019

328. 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, Buskamp 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]

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