

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

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- Reviews, guidelines, etc.
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- 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

Bouhy D, Juneja M, Katona I, Holmgren A, Asselbergh B, De Winter V, Hochepped 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

Mair D, Biskup S, Kress W, Abicht A, Brück W, Zechel S, Knop KC, Koenig FB, Tey S, Nikolin S, Eggemann 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 (accepted for publication). *equal contribution

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

Publications 2021

354. 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*. 2021 Epub ahead of print. PMID: 33781799

353. 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 β Signaling. *Neurotherapeutics*. 2021 Apr 15 Epub ahead of print. PMID: 33860461.

352. 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)*. 2021 Epub ahead of print. PMID: 33811521

351. 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*. 2021 Epub ahead of print. PMID: 33428302

350. 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

349. 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 neuromuscular disorder with α -dystroglycan hyperglycosylation. *J Clin Invest*. 131(9):e139076, 2021

348. 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

347. 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

346. 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, Libiouille C, Choukair D, Oommen PT, Borkhardt A, Surowy H, Wieczorek D, Meyer R, Eggermann T, Begemann M, Van Schaftingen 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

345. 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

Publications 2020

344. 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**, Fürst 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

343. 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

342. 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

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

340. 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

339. 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

338. 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, Kas-subek J, Rasche V. Histological correlates of postmortem ultra-high-resolution single-section MRI in cortical cerebral microinfarcts. *Acta Neuropathol Commun.* 8(1): 33, 2020

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.* 15(1): 015012, 2020

336. 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

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* 46(4): 359-374, 2020

334. 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

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333. 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

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

331. 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.* 6(5). pii: e603, 2019

330. 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

329. 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

328. 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

326. 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

325. 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

324. 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

323. 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

322. 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

321. 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

320. 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

319. 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]

318. 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

317. 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
316. 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
315. 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, Rohrmann 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
313. 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
312. 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
311. 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, Hathazi D, Roos A. Biochemical and pathological changes result from mutated Caveolin-3 in muscle. *Skelet Muscle.* 8(1): 28, 2018
310. 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
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308. 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
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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

306. 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

305. 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

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303. 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

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