

Neuro aktuell

Ausgabe 7-2020

Literatur

Schwerpunkt: Spinale Muskelatrophie

Titel: Therapeutische Optionen bei erwachsenen Patienten mit spinaler Muskelatrophie (S. 7–13)

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Literatur:

1. Farrar MA & Kiernan MC. The Genetics of Spinal Muscular Atrophy: Progress and Challenges. *Neurotherapeutics* 2015; 12(2): 290–302
2. Lefebvre S et al. Identification and characterization of a spinal muscular atrophy-determining gene. *Cell* 1995; 80(1): 155–165
3. Russman BS. Spinal muscular atrophy: clinical classification and disease heterogeneity. *J Child Neurol* 2007; 22(8): 946–951
4. Lorson CL et al. A single nucleotide in the SMN gene regulates splicing and is responsible for spinal muscular atrophy. *Proc Natl Acad Sci USA* 1999; 96(11): 6307–6311
5. Mailman MD et al. Molecular analysis of spinal muscular atrophy and modification of the phenotype by SMN2. *Genet Med* 2002; 4(1): 20–26
6. Crawford TO et al. Evaluation of SMN protein, transcript, and copy number in the biomarkers for spinal muscular atrophy (BforSMA) clinical study. *PLoS One* 2012; 7(4): e33572
7. Castro D & Iannaccone ST. Spinal muscular atrophy: therapeutic strategies. *Curr Treat Options Neurol* 2014; 16(11): 316
8. Finkel RS et al. Nusinersen versus Sham Control in Infantile-Onset Spinal Muscular Atrophy. *N Engl J Med* 2017; 377(18): 1723–1732
9. Mercuri E et al. Nusinersen versus Sham Control in Later-Onset Spinal Muscular Atrophy. *N Engl J Med* 2018; 378(7): 625–635
10. European Medicines Agency, Assessment Report Spinraza
https://www.ema.europa.eu/en/documents/assessment-report/spinraza-epar-public-assessment-report_en.pdf [letzter Zugriff: 08.09.20 um 17:35]
11. Hagenacker T et al. Nusinersen in adults with 5q spinal muscular atrophy: a non-interventional, multicentre, observational cohort study. *Lancet Neurol* 2020; 19(4): 317–325
12. Wurster CD et al. Intrathecal administration of nusinersen in adolescent and adult SMA type 2 and 3 patients. *J Neurol* 2019; 266(1): 183–194
13. Kizina K et al. Clinical Implication of Dosimetry of Computed Tomography- and Fluoroscopy-Guided Intrathecal Therapy With Nusinersen in Adult Patients With Spinal Muscular Atrophy. *Front Neurol* 2019; 10: 1166. Published 2019 Nov 5
14. Strauss KA et al. Preliminary Safety and Tolerability of a Novel Subcutaneous Intrathecal Catheter System for Repeated Outpatient Dosing of Nusinersen to Children and Adults With Spinal Muscular Atrophy. *J Pediatr Orthop* 2018; 38(10): e610–e617
15. Kirschner J et al. European ad-hoc consensus statement on gene replacement therapy for spinal muscular atrophy [published online ahead of print, 2020 Jul 9]. *Eur J Paediatr Neurol* 2020; S1090–3798(20)30142–2
16. Ziegler A et al. Handlungsempfehlungen zur Gentherapie der spinalen Muskelatrophie mit Onasemnogene Abeparvovec – AVXS-101: Konsensuspapier der deutschen Vertretung der

- Gesellschaft für Neuropädiatrie (GNP) und der deutschen Behandlungszentren unter Mitwirkung des Medizinisch-Wissenschaftlichen Beirates der Deutschen Gesellschaft für Muskelkranke (DGM) e. V. *Nervenarzt* 2020; 91(6): 518–529
17. Wang D et al. Adeno-associated virus vector as a platform for gene therapy delivery. *Nat Rev Drug Discov* 2019; 18(5): 358–378
 18. Mendell JR et al. Single-Dose Gene-Replacement Therapy for Spinal Muscular Atrophy. *N Engl J Med* 2017; 377(18): 1713–1722
 19. Lowes LP et al. Impact of Age and Motor Function in a Phase 1/2A Study of Infants With SMA Type 1 Receiving Single-Dose Gene Replacement Therapy. *Pediatr Neurol* 2019; 98: 39–45
 20. Mendell J et al. AVXS-101 Phase 1 Gene Replacement Therapy Clinical Trial in SMA Type 1: Continued Event Free Survival and Achievement of Developmental Milestones *Neurology* 2018; 90: 29.001
 21. Foust KD et al. Rescue of the spinal muscular atrophy phenotype in a mouse model by early postnatal delivery of SMN. *Nat Biotechnol* 2010; 28(3): 271–274
 22. Jędrzejowska M & Kostera-Pruszczyk A. Spinal muscular atrophy – new therapies, new challenges. *Neurol Neurochir Pol* 2020; 54(1): 8–13
 23. Novartis Media Release: <https://www.novartis.com/news/media-releases/novartis-announces-avxs-101-intrathecal-study-update> [letzter Zugriff: 27.08.20 um 11:00 Uhr]
 24. European Medicines Agency, Zolgensma: EPAR – Product information: https://www.ema.europa.eu/en/documents/product-information/zolgensma-epar-product-information_en.pdf [letzter Zugriff: 26.08.20 um 11:30 Uhr]
 25. Poirier A et al. Risdiplam distributes and increases SMN protein in both the central nervous system and peripheral organs. *Pharmacol Res Perspect* 2018; 6(6): e00447. Published 2018 Nov 29
 26. Roche Media Release: <https://www.roche.com/dam/jcr:916a6937-413a-47b5-9e0e-ac4cad70c71b/en/roche-media-release-230120.pdf> [letzter Zugriff: 27.08.20 um 11:00 Uhr]
 27. <https://neurologienetz.de/fachliches/erkrankungen/neuro-muskulaere-erkrankungen/spinale-muskelatrophien#c3618> [letzter Zugriff: 27.08.20 um 11:00 Uhr]
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Schwerpunkt: Kopfschmerz

Titel: Alkoholinduzierter Kopfschmerz – Pathogenese und Strategien der Prävention (S. 30–35)

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Literatur:

1. Verster JC, Vermeulen SA, Loo AJAEV, et al. Dietary Nutrient Intake, Alcohol Metabolism, and Hangover Severity. *J Clin Med* 2019; 8(9): 1316
2. Goodsell DS. Molecule of the Month: Alcohol Dehydrogenase. 2001. (<https://pdb101.rcsb.org/motm/13>). Zugegriffen: 10.08.2020
3. Kagi JH, Vallee BL. The role of zinc in alcohol dehydrogenase. V. The effect of metal-binding agents on the structure of the yeast alcohol dehydrogenase molecule. *J Biol Chem* 1960; 235: 3188–3192
4. Rocco A, Compare D, Angrisani D, Sanduzzi Zamparelli M, Nardone G. Alcoholic disease: liver and beyond. *World J Gastroenterol* 2014 Oct 28; 20(40): 14652–14659
5. Stephens R, Ling J, Heffernan TM, Heather N, Jones K. A review of the literature on the cognitive effects of alcohol hangover. *Alcohol Alcohol* 2008; 43(2): 163–170

6. Swift R, Davidson D. Alcohol hangover: mechanisms and mediators. *Alcohol Health Res World* 1998; 22(1): 54–60
7. Wiese JG, Shlipak MG, Browner WS. The alcohol hangover. *Ann Intern Med* 2000; 132(11): 897–902
8. Maxwell CR, Spangenberg RJ, Hoek JB, Silberstein SD, Oshinsky ML. Acetate causes alcohol hangover headache in rats. *PLoS One* 2010; 5(12): e15963
9. Van de Loo A, Mackus M, Korte-Bouws G, Brookhuis K, Garssen J, Verster J. Urine ethanol concentration and alcohol hangover severity. *Psychopharmacology (Berl)* 2017; 234(1): 73–77
10. Kim MJ, Lim SW, Kim JH, Choe DJ, Kim JI, Kang MJ. Effect of Mixed Fruit and Vegetable Juice on Alcohol Hangovers in Healthy Adults. *Prev Nutr Food Sci* 2018; 23(1): 1–7
11. Hong YH. Effects of the herb mixture, DTS20, on oxidative stress and plasma alcoholic metabolites after alcohol consumption in healthy young men. *Integr Med Res* 2016; 5(4): 309–316
12. Varella Morandi Junqueira-Franco M, Ernesto Troncon L, Garcia Chiarello P, do Rosário Del Lama Unamuno M, Afonso Jordao A, Vannucchi H. Intestinal permeability and oxidative stress in patients with alcoholic pellagra. *Clin Nutr* 2006; 25(6): 977–983
13. Narasimha VL, Ganesh S, Reddy S, et al. Pellagra and Alcohol Dependence Syndrome: Findings from a Tertiary Care Addiction Treatment Centre in India. *Alcohol Alcohol* 2019; 54(2): 148–151
14. Li Q, Xie G, Zhang W, et al. Dietary nicotinic acid supplementation ameliorates chronic alcohol-induced fatty liver in rats. *Alcohol Clin Exp Res* 2014; 38(7): 1982–1992
15. Li H, Hallows WH, Punzi JS, Pankiewicz KW, Watanabe KA, Goldstein BM. Crystallographic studies of isosteric NAD analogues bound to alcohol dehydrogenase: specificity and substrate binding in two ternary complexes. *Biochemistry* 1994; 33(39): 11734–11744