



OPTIMIZING OUTCOMES FOR ADULTS WITH SPINAL MUSCULAR ATROPHY

PATIENT-CENTRIC STRATEGIES FOR THE MULTIDISCIPLINARY TREATMENT TEAM

Bibliography

- Butchbach ME. Copy number variations in the survival motor neuron genes: implications for spinal muscular atrophy and other neurodegenerative diseases. *Front Mol Biosci*. 2016;3:7.
- ClinicalTrials.gov. A study for participants with spinal muscular atrophy (SMA) who previously participated in nusinersen (ISIS 396443) investigational studies (SHINE). Identifier: NCT02594124. ClinicalTrials website. <https://clinicaltrials.gov/ct2/show/NCT02594124>. Accessed August 2022.
- ClinicalTrials.gov. A study of risdiplam (RO7034067) in adult and pediatric participants with spinal muscular atrophy (JEWELFISH). Identifier: NCT03032172. Clinical Trials website. <https://clinicaltrials.gov/ct2/show/NCT03032172>. Accessed August 2022.
- Cure SMA. Voice of the patient report: spinal muscular atrophy. January 10, 2018. Cure SMA website. <https://www.curesma.org/wp-content/uploads/2018/01/SMA-VoP-for-publication-1-22-2018.pdf>. Accessed August 2022.
- Cruz R, Lenz M, Belter L, et al. Voice of the patient report: spinal muscular atrophy. January 10, 2018. Cure SMA website. <https://www.curesma.org/wp-content/uploads/2018/01/SMA-VoP-for-publication-1-22-2018.pdf>. Accessed January 2023.
- Darba J, Marsa A. Patient characteristics and hospitalisation costs of spinal muscular atrophy in Spain: a retrospective multicentre database analysis. *BMJ Open*. 2019;9(11):e031271.
- Darras BT, Masson R, Mazurkiewicz-Betdzińska M, et al. Risdiplam-treated infants with type 1 spinal muscular atrophy versus historical controls. *N Engl J Med*. 2021;385(5):427–435.
- Farrar MA, Carey KA, Paguinto S-G, et al. Financial, opportunity and psychosocial costs of spinal muscular atrophy: an exploratory qualitative analysis of Australian carer perspectives. *BMJ Open*. 2018;8(5):e020907.
- FDA-approved drug: nusinersen. Revised December 2016. U.S. Food and Drug Administration website. https://www.accessdata.fda.gov/drugsatfda_docs/label/2016/209531lbl.pdf. Accessed August 2022.
- FDA-approved drug: onasemnogene abeparvovec-xioi. Revised October 2021. U.S. Food and Drug Administration website. <https://www.fda.gov/media/126109/download>. Accessed August 2022.
- FDA-approved drug: risdiplam. Revised April 2021. U.S. Food and Drug Administration website. https://www.accessdata.fda.gov/drugsatfda_docs/label/2021/213535s001lbl.pdf. Accessed August 2022.
- Finkel RS, Mercuri E, Darras BT, et al. Nusinersen versus sham control in infantile-onset spinal muscular atrophy. *N Engl J Med*. 2017;377(18):1723–1732.
- Hagenacker T, Wurster CD, Günther R. Nusinersen in adults with 5q spinal muscular atrophy: a non-interventional, multicentre, observational cohort study (see appendix). *Lancet Neurol*. 2020;19(4):317–325.
- Kolb SJ, Kissel JT. Spinal muscular atrophy. *Neurol Clin*. 2015;33(4):831–846.
- LaMarca NH, Golden L, John RM, et al. Diabetic ketoacidosis in an adult patient with spinal muscular atrophy type II: further evidence of extraneural pathology due to survival motor neuron 1 mutation? *J Child Neurol*. 2013;28(11):1517–1520.
- Maggi L, Bello L, Bonanno S, et al. Nusinersen safety and effects on motor function in adult spinal muscular atrophy type 2 and 3. *J Neurol Neurosurg Psychiatry*. 2020;91(11):1166–1174.
- Mazzella A, Curry M, Belter L, et al. "I have SMA, SMA doesn't have me": a qualitative snapshot into the challenges, successes, and quality of life of adolescents and young adults with SMA. *Orphanet J Rare Dis*. 2021;16(1):96.
- Mercuri E, Darras BT, Chiriboga CA, et al. Nusinersen versus sham control in later-onset spinal muscular atrophy. *N Engl J Med*. 2018;378(7):625–635.

- 
- Mercuri E, Deconinck N, Mazzone ES, et al. Safety and efficacy of once-daily risdiplam in type 2 and non-ambulant type 3 spinal muscular atrophy (SUNFISH part 2): a phase 3, double-blind, randomised, placebo-controlled trial [published correction appears in *Lancet Neurol*. 2022;21(2):e2] [published correction appears in *Lancet Neurol*. 2022;21(3):e3]. *Lancet Neurol*. 2022;21(1):42–52.
- Mercuri E, Finkel RS, Muntoni F, et al. Diagnosis and management of spinal muscular atrophy: part 1: recommendations for diagnosis, rehabilitation, orthopedic and nutritional care. *Neuromuscul Disord*. 2018;28(2):103–115.
- Mongioli P, Dilek N, Garland C, et al. Patient reported impact of symptoms in spinal muscular atrophy (PRISM-SMA). *Neurology*. 2018;91(13):e1206–e1214.
- Rouault F, Christie-Brown V, Broekgaarden R, et al. Disease impact on general well-being and therapeutic expectations of European type II and type III spinal muscular atrophy patients. *Neuromuscul Disord*. 2017;27:428–38.
- Ozge Ozkaya. Spinal Muscular Atrophy: SMA Type 4. Rare Disease Advisor Website. Available at: <https://www.rarediseaseadvisor.com/hcp-resource/spinal-muscular-atrophy-type-4/#:~:text=Spinal%20muscular%20atrophy%20%28SMA%29%20type%204%20is%20the,be%20approximately%201%2F300%2C000.%203%20SMA%20Type%204%20Causes>. Last reviewed July 2021. Accessed January 2023.
- Sporer SM, Smith BG. Hip dislocation in patients with spinal muscular atrophy. *J Pediatr Orthop*. 2003;23(1):10–14.
- Talbot K, Tizzano EF. The clinical landscape for SMA in a new therapeutic era. *Gene Therapy*. 2017;24:529-533.
- Verhaart IEC, Robertson A, Leary R, et al. A multi-source approach to determine SMA incidence and research ready population. *J Neurol*. 2017;264(7):1465–1473.
- Verhaart IEC, Robertson A, Wilson IJ. et al. Prevalence, incidence, and carrier frequency of 5q-linked spinal muscular atrophy—a literature review. *Orphanet J Rare Dis*. 2017;12(124).
- Walter MC, Chiriboga C, Duong T, et al. Improving care and empowering adults living with SMA: a call to action in the new treatment era. *J Neuromuscul Dis*. 2021;8(4):543–551.
- Wan HWY, Carey KA, D'Silva A, et al. Health, wellbeing and lived experiences of adults with SMA: a scoping systematic review. *Orphanet J Rare Dis*. 2020;15(1):70.