

PubMed

Format: Summary Sort by: Best Match Per page: 20

Search results

Items: 1 to 20 of 518

- [Two breakthrough gene-targeted treatments for spinal muscular atrophy: challenges remain.](#)
1. Sumner CJ, Crawford TO.
J Clin Invest. 2018 Aug 1;128(8):3219-3227. doi: 10.1172/JCI121658. Epub 2018 Jul 9. Review.
PMID: 29985170 **Free PMC Article**
- [Mechanism of Splicing Regulation of Spinal Muscular Atrophy Genes.](#)
2. Singh RN, Singh NN.
Adv Neurobiol. 2018;20:31-61. doi: 10.1007/978-3-319-89689-2_2. Review.
PMID: 29916015 **Free PMC Article**
- [Nusinersen in the Treatment of Spinal Muscular Atrophy.](#)
3. Goodkey K, Aslesh T, Maruyama R, Yokota T.
Methods Mol Biol. 2018;1828:69-76. doi: 10.1007/978-1-4939-8651-4_4. Review.
PMID: 30171535
- [Spinal muscular atrophy 5Q - Treatment with nusinersen.](#)
4. Brazilian Medical Association, Silvinato A, Bernardo WM.
Rev Assoc Med Bras (1992). 2018 Jun;64(6):484-491. doi: 10.1590/1806-9282.64.06.484. Review.
PMID: 30304304 **Free Article**
- [Expanded Carrier Screening.](#)
5. Gregg AR.
Obstet Gynecol Clin North Am. 2018 Mar;45(1):103-112. doi: 10.1016/j.ogc.2017.10.005. Review.
PMID: 29428278
- [AAV gene delivery to the spinal cord: serotypes, methods, candidate diseases, and clinical trials.](#)
6. Hardcastle N, Boulis NM, Federici T.
Expert Opin Biol Ther. 2018 Mar;18(3):293-307. doi: 10.1080/14712598.2018.1416089. Epub 2017 Dec 18. Review.
PMID: 29249183
- [Diagnosis and management of spinal muscular atrophy: Part 1: Recommendations for diagnosis, rehabilitation, orthopedic and nutritional care.](#)
7. Mercuri E, Finkel RS, Muntoni F, Wirth B, Montes J, Main M, Mazzone ES, Vitale M, Snyder B, Quijano-Roy S, Bertini E, Davis RH, Meyer OH, Simonds AK, Schroth MK, Graham RJ, Kirschner J, Iannaccone ST, Crawford TO, Woods S, Qian Y, Sejersen T; SMA Care Group.
Neuromuscul Disord. 2018 Feb;28(2):103-115. doi: 10.1016/j.nmd.2017.11.005. Epub 2017 Nov 23. Review.
PMID: 29290580 **Free Article**
- [Selective modulation of the androgen receptor AF2 domain rescues degeneration in spinal bulbar muscular atrophy.](#)
8. Badders NM, Korff A, Miranda HC, Vuppala PK, Smith RB, Winborn BJ, Quemin ER, Sopher BL, Dearman J, Messing J, Kim NC, Moore J, Freibaum BD, Kanagaraj AP, Fan B, Tillman H, Chen PC, Wang Y, Freeman BB III, Li Y, Kim HJ, La Spada AR, Taylor JP.
Nat Med. 2018 May;24(4):427-437. doi: 10.1038/nm.4500. Epub 2018 Mar 5.
PMID: 29505030 **Free PMC Article**

- [Measuring quality of life in children with spinal muscular atrophy: a systematic literature review.](#)
9. Vaidya S, Boes S.
Qual Life Res. 2018 Dec;27(12):3087-3094. doi: 10.1007/s11136-018-1945-x. Epub 2018 Jul 24. Review. Erratum in: [Qual Life Res. 2018 Sep 14](#);.
PMID: 30043243
- [Cell cycle inhibitors protect motor neurons in an organoid model of Spinal Muscular Atrophy.](#)
10. Hor JH, Soh ES, Tan LY, Lim VJW, Santosa MM, Winanto, Ho BX, Fan Y, Soh BS, Ng SY.
Cell Death Dis. 2018 Oct 27;9(11):1100. doi: 10.1038/s41419-018-1081-0.
PMID: 30368521 **Free PMC Article**
- [Ambulatory function in spinal muscular atrophy: Age-related patterns of progression.](#)
11. Montes J, McDermott MP, Mirek E, Mazzone ES, Main M, Glanzman AM, Duong T, Young SD, Salazar R, Pasternak A, Gee R, De Sanctis R, Coratti G, Forcina N, Fanelli L, Ramsey D, Milev E, Civitello M, Pane M, Pera MC, Scoto M, Day JW, Tennekoon G, Finkel RS, Darras BT, Muntoni F, De Vivo DC, Mercuri E.
PLoS One. 2018 Jun 26;13(6):e0199657. doi: 10.1371/journal.pone.0199657. eCollection 2018.
PMID: 29944707 **Free PMC Article**
- [Update on muscle disease.](#)
12. Witherick J, Brady S.
J Neurol. 2018 Jul;265(7):1717-1725. doi: 10.1007/s00415-018-8856-1. Epub 2018 Apr 18. Review.
PMID: 29671051
- [Genome-wide CRISPR-Cas9 Screen Identifies Leukemia-Specific Dependence on a Pre-mRNA Metabolic Pathway Regulated by DCPS.](#)
13. Yamauchi T, Masuda T, Canver MC, Seiler M, Semba Y, Shboul M, Al-Raqad M, Maeda M, Schoonenberg VAC, Cole MA, Macias-Trevino C, Ishikawa Y, Yao Q, Nakano M, Arai F, Orkin SH, Reversade B, Buonamici S, Pinello L, Akashi K, Bauer DE, Maeda T.
Cancer Cell. 2018 Mar 12;33(3):386-400.e5. doi: 10.1016/j.ccell.2018.01.012. Epub 2018 Feb 22.
PMID: 29478914 **Free PMC Article**
- [Targeting RNA structure in SMN2 reverses spinal muscular atrophy molecular phenotypes.](#)
14. Garcia-Lopez A, Tessaro F, Jonker HRA, Wacker A, Richter C, Comte A, Berntenis N, Schmucki R, Hatje K, Petermann O, Chiriano G, Perozzo R, Sciarra D, Konieczny P, Faustino I, Fournet G, Orozco M, Artero R, Metzger F, Ebeling M, Goekjian P, Joseph B, Schwalbe H, Scapozza L.
Nat Commun. 2018 May 23;9(1):2032. doi: 10.1038/s41467-018-04110-1.
PMID: 29795225 **Free PMC Article**
- [Molecular Factors Involved in Spinal Muscular Atrophy Pathways as Possible Disease-modifying Candidates.](#)
15. Maretina MA, Zheleznyakova GY, Lanko KM, Egorova AA, Baranov VS, Kiselev AV.
Curr Genomics. 2018 Aug;19(5):339-355. doi: 10.2174/1389202919666180101154916. Review.
PMID: 30065610 **Free PMC Article**
- [Recent Advances and Clinical Applications of Exon Inclusion for Spinal Muscular Atrophy.](#)
16. Son HW, Yokota T.
Methods Mol Biol. 2018;1828:57-68. doi: 10.1007/978-1-4939-8651-4_3. Review.
PMID: 30171534
- [The role of survival motor neuron protein \(SMN\) in protein homeostasis.](#)
17. Chaytow H, Huang YT, Gillingwater TH, Faller KME.
Cell Mol Life Sci. 2018 Nov;75(21):3877-3894. doi: 10.1007/s00018-018-2849-1. Epub 2018 Jun 5. Review.
PMID: 29872871 **Free PMC Article**
- [Motor milestone assessment of infants with spinal muscular atrophy using the hammersmith infant neurological Exam-Part 2: Experience from a nusinersen clinical study.](#)
- 18.

Bishop KM, Montes J, Finkel RS.

Muscle Nerve. 2018 Jan;57(1):142-146. doi: 10.1002/mus.25705. Epub 2017 Jun 14.

PMID: 28556387

[Precision Medicine through Antisense Oligonucleotide-Mediated Exon Skipping.](#)

19. Li D, Mastaglia FL, Fletcher S, Wilton SD.

Trends Pharmacol Sci. 2018 Nov;39(11):982-994. doi: 10.1016/j.tips.2018.09.001. Epub 2018 Sep 30. Review.

PMID: 30282590

[Using Systems Biology and Mathematical Modeling Approaches in the Discovery of Therapeutic Targets for](#)

20. [Spinal Muscular Atrophy.](#)

Butchbach MER.

Adv Neurobiol. 2018;21:267-281. doi: 10.1007/978-3-319-94593-4_10. Review.

PMID: 30334226 **Free PMC Article**

[Back to top](#)