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- [68] Kariyawasam DS, D'Silva AM, Sampaio H, Briggs N, Herbert K, Wiley V, et al. Newborn screening for spinal muscular atrophy in Australia: a non-randomised cohort study. *Lancet Child Adolesc Health*. 2023;7(3):159-70.
- [69] ClinicalTrials.gov. Safety and Efficacy of Intravenous OAV101 (AVXS-101) in Pediatric Patients with Spinal Muscular Atrophy (SMA) (SMART). 2021 [updated 2023 Aug 18; cited 2023 Nov 21]. Available from: <https://clinicaltrials.gov/ct2/show/NCT04851873>.
- [70] ClinicalTrials.gov. Safety and Efficacy of Intravenous OAV101 (AVXS-101) in Pediatric Patients With Spinal Muscular Atrophy (SMA) (OFELIA). 2021 [updated 2023 Nov 14; cited 2023 Nov 21]. Available from: <https://clinicaltrials.gov/ct2/show/NCT05073133>.
- [71] Servais L, De Vivo DC, Kirschner J, Mercuri E, Muntoni F, Tizzano EF, et al. P.79 Effectiveness and safety of onasemnogene abeparvovec in older patients with spinal muscular atrophy: Real-world outcomes from the RESTORE registry. Poster presented at: Muscular Dystrophy Association Clinical and Scientific Congress. 2022. [updated 2022; cited 2023 Apr 1]. Available from: <https://www.mdaconference.org/abstract-library/effectiveness-and-safety-of-onasemnogene-abeparvovec-in-older-patients-with-spinal-muscular-atrophy-real-world-outcomes-from-the-restore-registry/>.
- [72] Servais L, Benguerba K, De Vivo D, Kirschner J, Mercuri E, Muntoni F, et al. P.103 Onasemnogene abeparvovec (OA) treatment outcomes by patient weight at infusion: Initial findings from the RESTORE registry. *Neuromuscul Disord*. 2022;32(1):S87.
- [73] Chand DH, Mitchell S, Sun R, LaMarca N, Reyna SP, Sutter T. Safety of onasemnogene abeparvovec for patients with spinal muscular atrophy 8.5kg or heavier in a global managed access program. *Pediatr Neurol*. 2022;132:27-32.
- [74] Ertl HCJ. Immunogenicity and toxicity of AAV gene therapy. *Front Immunol*. 2022;13:975813.
- [75] Kishimoto TK, Samulski RJ. Addressing high dose AAV toxicity - 'one and done' or 'slower and lower'? *Expert Opin Biol Ther*. 2022;22(9):1067-71.
- [76] Chand D, Mohr F, McMillan H, Tukov FF, Montgomery K, Kleyn A, et al. Hepatotoxicity following administration of onasemnogene abeparvovec (AVXS-101) for the treatment of spinal muscular atrophy. *J Hepatol*. 2021;74(3):560-6.
- [77] Wijngaarde CA, Blank AC, Stam M, Wadman RI, van den Berg LH, van der Pol WL. Cardiac pathology in spinal muscular atrophy: a systematic review. *Orphanet J Rare Dis*. 2017;12(1):67.
- [78] Chand DH, Zaidman C, Arya K, Millner R, Farrar MA, Mackie FE, et al. Thrombotic microangiopathy following onasemnogene abeparvovec for spinal muscular atrophy: a case series. *J Pediatr*. 2021;231:265-68.
- [79] De Sanctis R, Coratti G, Pasternak A, Montes J, Pane M, Mazzone ES, et al. Developmental milestones in type I spinal muscular atrophy. *Neuromuscul Disord*. 2016;26(11):754-9.
- [80] Dangouloff T, Vrščaj E, Servais L, Osredkar D, SMA NBS World Study Group. Newborn screening programs for spinal muscular atrophy worldwide: Where we stand and where to go. *Neuromuscul Disord*. 2021;31(6):574-82.
- [81] McMillan HJ, Proud CM, Farrar MA, Alexander IE, Muntoni F, Servais L. Onasemnogene abeparvovec for the treatment of spinal muscular atrophy. *Expert Opin Biol Ther*. 2022;22(9):1075-90.
- [82] Al-Zaidy SA, Mendell JR. From clinical trials to clinical practice: practical considerations for gene replacement therapy in SMA type 1. *Pediatr Neurol*. 2019;100:3-11.
- [83] Kirschner EA, Proud CM, Farrar MA, Kwon JM, Saito K, Desguerre I, McMillan HJ. Expert recommendations and clinical considerations in the use of onasemnogene abeparvovec gene therapy for spinal muscular atrophy. *Muscle Nerve*. 2021;64(4):413-27.
- [84] Faulkner E, Spinner DS, Ringo M, Carroll M. Are global health systems ready for transformative therapies? *Value Health*. 2019;22(6):627-41.
- [85] Faulkner E, Ringo M, Spinner D, Carroll, Latif E, Miller TM. Guiding principles of rare disease care and patient access. *Global Genes and The Child Neurology Foundation*. September 2020. [updated 2022; cited 2023 Apr 1]. Available at: <https://globalgenes.org/white-paper/access-to-critical-therapies-guiding-principles-of-rare-disease-care-and-patient-access/>.