

(12) INTERNATIONAL APPLICATION PUBLISHED UNDER THE PATENT COOPERATION TREATY (PCT)

(19) World Intellectual Property
Organization
International Bureau

(43) International Publication Date
19 May 2023 (19.05.2023)



(10) International Publication Number
WO 2023/086928 A3

(51) International Patent Classification:

C12N 15/861 (2006.01) *C12N 9/14* (2006.01)
A61K 35/761 (2015.01) *C12N 15/86* (2006.01)
A61K 48/00 (2006.01) *A61K 35/76* (2015.01)
A61K 38/46 (2006.01)

GH, GM, KE, LR, LS, MW, MZ, NA, RW, SD, SL, ST, SZ, TZ, UG, ZM, ZW), Eurasian (AM, AZ, BY, KG, KZ, RU, TJ, TM), European (AL, AT, BE, BG, CH, CY, CZ, DE, DK, EE, ES, FI, FR, GB, GR, HR, HU, IE, IS, IT, LT, LU, LV, MC, ME, MK, MT, NL, NO, PL, PT, RO, RS, SE, SI, SK, SM, TR), OAPI (BF, BJ, CF, CG, CI, CM, GA, GN, GQ, GW, KM, ML, MR, NE, SN, TD, TG).

(21) International Application Number:

PCT/US2022/079701

(22) International Filing Date:

11 November 2022 (11.11.2022)

(25) Filing Language:

English

(26) Publication Language:

English

(30) Priority Data:

63/278,775 12 November 2021 (12.11.2021) US
63/288,293 10 December 2021 (10.12.2021) US

Published:

- with international search report (Art. 21(3))
- before the expiration of the time limit for amending the claims and to be republished in the event of receipt of amendments (Rule 48.2(h))
- with sequence listing part of description (Rule 5.2(a))

(88) Date of publication of the international search report:

22 June 2023 (22.06.2023)

(71) Applicants: **THE TRUSTEES OF THE UNIVERSITY OF PENNSYLVANIA** [US/US]; Penn Center for Innovation, 3600 Civic Center Blvd., 9th Floor, Philadelphia, PA 19104 (US). **AMICUS THERAPEUTICS, INC.** [US/US]; 3675 Market Street, Philadelphia, PA 19104 (US).

(72) Inventors: **HORDEAUX, Juliette**; 203 Head House Court, Philadelphia, PA 19147 (US). **WILSON, James, M.**; 1831 Delancey Street, Philadelphia, PA 19103 (US). **RASSOULI-TAYLOR, Leida**; 3675 Market Street, Philadelphia, PA 19104 (US). **DO, Hung**; 3675 Market Street, Philadelphia, PA 19104 (US). **TUSKE, Steven**; 3675 Market Street, Philadelphia, PA 19104 (US).

(74) Agent: **KODROFF, Cathy, A.** et al.; Howson & Howson LLP, 325 Sentry Parkway East, Five Sentry East, Suite 160, Blue Bell, PA 19422 (US).

(81) Designated States (unless otherwise indicated, for every kind of national protection available): AE, AG, AL, AM, AO, AT, AU, AZ, BA, BB, BG, BH, BN, BR, BW, BY, BZ, CA, CH, CL, CN, CO, CR, CU, CV, CZ, DE, DJ, DK, DM, DO, DZ, EC, EE, EG, ES, FI, GB, GD, GE, GH, GM, GT, HN, HR, HU, ID, IL, IN, IQ, IR, IS, IT, JM, JO, JP, KE, KG, KH, KN, KP, KR, KW, KZ, LA, LC, LK, LR, LS, LU, LY, MA, MD, MG, MK, MN, MW, MX, MY, MZ, NA, NG, NI, NO, NZ, OM, PA, PE, PG, PH, PL, PT, QA, RO, RS, RU, RW, SA, SC, SD, SE, SG, SK, SL, ST, SV, SY, TH, TJ, TM, TN, TR, TT, TZ, UA, UG, US, UZ, VC, VN, WS, ZA, ZM, ZW.

(84) Designated States (unless otherwise indicated, for every kind of regional protection available): ARIPO (BW, CV,

(54) Title: GENE THERAPY FOR TREATMENT OF MUCOPOLYSACCHARIDOSIS IIIA

(57) Abstract: Provided herein is a recombinant AAV (rAAV) comprising an AAV capsid and a vector genome packaged therein, wherein the vector genome comprises an AAV 5' inverted terminal repeat (ITR), an engineered nucleic acid sequence encoding a functional hSGSH, optionally comprising stabilizing amino acid changes, a regulatory sequence which direct expression of hSGSH in a target cell, and an AAV 3' ITR. Also provided is a pharmaceutical composition comprising a rAAV as described herein in a formulation buffer, and a method of treating a human subject diagnosed with MPS IIIA.



WO 2023/086928 A3