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

(19) World Intellectual Property Organization
International Bureau

(43) International Publication Date
22 May 2014 (22.05.2014)

WIPO | PCT

(10) International Publication Number
WO 2014/078353 A1

(51) International Patent Classification:
C12P 21/00 (2006.01) *A61K 38/19* (2006.01)

(21) International Application Number:
PCT/US2013/069799

(22) International Filing Date:
13 November 2013 (13.11.2013)

(25) Filing Language: English

(26) Publication Language: English

(30) Priority Data:
61/726,252 14 November 2012 (14.11.2012) US
61/825,227 20 May 2013 (20.05.2013) US

(71) Applicant: **EDIMER PHARMACEUTICALS, INC.** [US/US]; 55 Cambridge Parkway, Cambridge, MA 02142 (US).

(72) Inventors: **HUTTNER, Kenneth M.**; 840 Newton Street, Chestnut Hill, US 02467 (US). **KIRBY, Neil**; 3 Meadowview Lane, Andover, US 01810 (US). **SCHNEIDER, Pascal**; Chemin des Ormeaux 10, Epalinges (CH). **GAIDE, Olivier**; Beaumont 14, Geneva (CH).

(74) Agent: **WARD, Donna T.**; DT Ward, P.C., 142A Main Street, Groton, Massachusetts 01450 (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, CZ, DE, DK, DM, DO, DZ, EC, EE, EG, ES, FI, GB, GD, GE, GH, GM, GT, HN, HR, HU, ID, IL, IN, IR, IS, JP, KE, KG, KN, KP, KR, KZ, LA, LC, LK, LR, LS, LT, LU, LY, MA, MD, ME, 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, SM, ST, SV, SY, TH, TJ, TM, TN, TR, TT, TZ, UA, UG, US, UZ, VC, VN, ZA, ZM, ZW.

(84) Designated States *(unless otherwise indicated, for every kind of regional protection available)*: ARIPO (BW, GH, GM, KE, LR, LS, MW, MZ, NA, RW, SD, SL, 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, 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).

Declarations under Rule 4.17:

— *as to applicant's entitlement to apply for and be granted a patent (Rule 4.17(ii))*

— *as to the applicant's entitlement to claim the priority of the earlier application (Rule 4.17(iii))*

Published:

— *with international search report (Art. 21(3))*

— *with sequence listing part of description (Rule 5.2(a))*

WO 2014/078353 A1

(54) Title: COMPOSITIONS AND METHODS FOR THE TREATMENT OF ECTODERMAL DYSPLASIA

(57) Abstract: The invention relates to pharmaceutical compositions and methods for the treatment of ectodermal displasias via the administration of EDA agonists, in particular EDI200. Use of the compositions and methods described allow for therapeutic dosing and administration regimens in human patients to correct or alter abnormal phenotypes associated with genetic disorders, in particular, XLHED.