

(51) International Patent Classification:
G01N 33/48 (2006.01) **G01N 33/50** (2006.01)(21) International Application Number:
PCT/US2009/053373(22) International Filing Date:
11 August 2009 (11.08.2009)

(25) Filing Language: English

(26) Publication Language: English

(30) Priority Data:
61/088,203 12 August 2008 (12.08.2008) US
61/186,673 12 June 2009 (12.06.2009) US
61/224,647 10 July 2009 (10.07.2009) US(71) Applicant (for all designated States except US): **SHIRAZ PHARMACEUTICALS, INC.** [US/US]; One Science Drive, Durham, North Carolina 27708 (US).

(72) Inventor; and

(75) Inventor/Applicant (for US only): **ROSES, Allen D.** [US/US]; 53525 Bickett, Chapel Hill, North Carolina 27517 (US).(74) Agent: **MYERS BIGEL SIBLEY & SAJOVEC, P.A.**; P.O. Box 37428, Raleigh, North Carolina 27627 (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, 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, IS, JP, KE, KG, KM, 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, PE, PG, PH, PL, PT, RO, RS, RU, SC, SD, SE, SG, SK, SL, SM, ST, SV, SY, 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, LS, MW, MZ, NA, SD, SL, SZ, TZ, UG, ZM, ZW), Eurasian (AM, AZ, BY, KG, KZ, MD, RU, TJ, TM), European (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, SE, SI, SK, SM, TR), OAPI (BF, BJ, CF, CG, CI, CM, GA, GN, GQ, GW, ML, MR, NE, SN, TD, TG).

Published:

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

— with amended claims (Art. 19(1))

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

[Continued on next page]

(54) Title: METHOD OF IDENTIFYING DISEASE RISK FACTORS

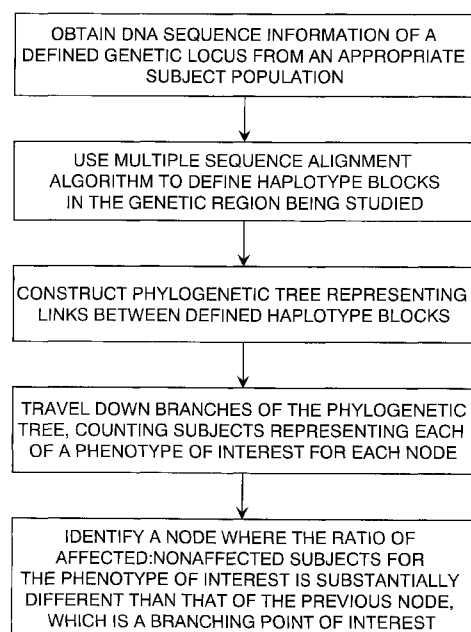


FIG. 1

(57) Abstract: Provided herein is a method for identifying a genetic variant that is associated with development of a condition of interest (e.g., Alzheimer's disease), and genetic variants so identified. Methods of treatment with an active agent (e.g., with a particular active agent and/or at an earlier age) is also provided, upon detecting a genetic variant described herein. In some embodiments, the genetic variant is a deletion/insertion polymorphism (DIP) of the TOMM40 gene. Kits for determining if a subject is at increased risk of developing late onset Alzheimer's disease is also provided. Kits for determining if a subject is responsive to treatment for a condition of interest with an active agent are further provided.



(88) Date of publication of the international search report: 22 April 2010 Date of publication of the amended claims: 13 January 2011