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(71) Applicant: AMBRY GENETICS [US/US]; 15 Argonaut,
Aliso Viejo, CA 92656 (US).(72) Inventors: LI, Xiang; 1700 Bluebird Ln., Carlsbad, CA
92011 (US). LU, Hong; 25339 Via Viejo, Lake Forest, CA
92630 (US). LU, Hsiaomei; 33 Bolinas, Irvine, CA 92602
(US). GONZALEZ, Kelly; 24197 Hollyoak, Apt. J, Aliso
Viejo, CA 92656 (US). PARRA, Melissa; 21003 Dumetz
Road, Woodland Hills, CA 91364 (US). ZENG, Wenqi;
1306 Standish Pl., Arcadia, CA 91006 (US). CHAO,
Elizabeth; 1930 Port Bishop Pl., Newport Beach, CA
93660 (US). DUNLOP, Charles; 2021 Temple Hills, La-
guna Beach, CA 92651 (US).(74) Agent: HILL, James, W.; 4 Park Plaza, Suite 1700,
Irvine, CA 92614 (US).(81) Designated States (unless otherwise indicated, for every
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(54) Title: MOLECULAR GENETIC DIAGNOSTIC SYSTEM

200

NEW RESULT SET FOR FAM_

210 DESCRIPTION

NAME

220 MENDELIAN INHERITANCE MODEL

● RECESSIVE ● DOMINANT

● AUTOSOMAL ● X-LINKED ● Y-LINKED

☐ DE NOVO MUTATION ALLOWED

230 POPULATION FREQUENCY

REMOVE COMMON VARIANTS WITH

FREQUENCY LARGER THAN 0.01

AND

OCCURRENCE LARGER THAN 5

240 GENOTYPES

FATHER (NORMAL) NEGATIVE

MOTHER (NORMAL) NEGATIVE

DAUGHTER (NORMAL) NEGATIVE

250 NORMAL CONTROLS

☐ FAM	PROBAND
☐ FAM	PROBAND
☐ FAM	DAUGHTER
☐ FAM	FATHER
☐ FAM	MOTHER
☐ FAM	PROBAND
☐ FAM	FATHER
☐ FAM	MOTHER
☐ FAM	PROBAND
☐ FAM	FATHER
☐ FAM	MOTHER
☐ FAM	PROBAND
☐ FAM	PROBAND

RUN RESET GO BACK

FIG.2

(57) Abstract: A computer-implemented bioinformatics program annotates human genetic variants by integrating multiple sources of information. The program rapidly filters variants that are unlikely to play a role in the etiology of particular diseases. This filtering may be performed based on such annotations, on clinical profiles and family histories, and on analyses under various inheritance models, in order to classify human variants and identify mutations influencing patients' diseases.